School of Psychology and Speech Pathology

Early speech motor and language skills in Childhood Apraxia of Speech: Evidence for a core deficit in speech motor control?

Chantelle D Highman

This thesis is presented for the Degree of
Doctor of Philosophy
Of
Curtin University of Technology

February 2010

Declaration

To the best of my knowledge and belief this thesis contains no material previously published by any other person except where due acknowledgement has been made.

This thesis contains no material which has been accepted for the award of any other degree or diploma in any university.

Signature: ……..

Date: ……..

ACKNOWLEDGEMENTS

This research would not have been possible if not for the team of supportive people surrounding me in various forms over the last few years. I consider myself very fortunate to have been able to explore a topic that stemmed from my clinical role, and I am grateful for the support I have received along the way.

I am indebted to the constant support of my supervisors, Dr Neville Hennessy and Dr Suze Leitão, during this process. Suze, thankyou for the original encouragement to explore my clinical areas of interest through a PhD. Your constant moral support and encouragement has underpinned my ability to finally finish. Thank you also for your expert assistance with conceptual, clinical, methodological and editorial aspects of the project. Neville, thankyou for your thoroughness, eye for detail, and persistence with many aspects of this project. I appreciate the many areas where you offered invaluable input, especially with regards to theoretical and statistical aspects of the research. I was also very fortunate to have Professor Jan Piek as an associate supervisor on this project. Not only have I been able to extend my knowledge of motor control in typical and disordered development, but the research process was very much demystified by way of Jan's involvement and constant encouragement – thankyou Jan!

Sincere thanks go to the staff and PhD students in the School of Psychology and Speech Pathology at Curtin – the friendly faces that offered advice, practical support, and administrative and IT assistance. I am grateful to Mellanie Sherwood for sharing her knowledge and experience in the area of CAS, and for so enthusiastically being involved in Study 1. The many other speech pathologists who assisted with recruitment and access to retrospective data are also thanked. Thanks go to the child health nurses who assisted with recruitment of infants, and to the early childhood teachers (particularly Rosalie Wallace) for graciously allowing me access to their students. I'm very grateful to Jan Marshall and Kate Bethel from the Child and Adolescent Health Branch of the Department of Health Western Australia for access to and assistance with the retrospective data. Acknowledgement is also made to

Speech Pathology Australia for kick-starting this project with a small clinical research grant to initiate Study 1. I am very grateful to Dr Natalie Gasson for her constructive comments and suggestions on an earlier draft of this thesis. Leesa Santa Maria and Anne-Marie Lewis are thanked for their assistance with establishing inter-rater reliability.

Thanks are due to my colleagues and friends at Andrea Way Child Development Centre. Your interest in, and, at times, distraction from, my research, helped to maintain the momentum! I am also very grateful to my clients and their families, especially those who were involved in the dyspraxia support group. Your open and honest discussions about your children taught me much about CAS, and planted the all-important research question in my mind. I appreciate the involvement of the children and families who participated in this research, especially those families whose infants took part. Thankyou for allowing me into your homes and your busy lives.

My family and friends have been patient, understanding and, above-all, tolerant of the time taken up by this project. Thankyou all for being there when I needed you, and for not being there when that is what was required. Narelle, thanks for your absolute enduring support and understanding, and Danielle, for your ability to encourage from afar. To my wonderful parents, Vivian and Bill Skett, thankyou for your constant encouragement, love and support. You have always instilled in me an understanding that hard work pays off in the end, and it is because of your continual support that I have been able to pursue my goals.

Finally, the biggest applause when I finally finish will come from my husband Shane and our daughter, Elise; two people who have dealt on a daily basis with the thesis' presence (or my absence). I am so grateful to Shane for his never-ending patience, constant encouragement and much needed emotional support, and for being so much fun to be around in the moments where I needed a break from it all. You have literally shared the ups and downs of this process with me, every step of the way, and have helped me to enjoy the successes along the way. And to Elise, who makes me smile every single day, thankyou for coming into our lives and giving me a constant reminder of what it's really all about!

ABSTRACT

Children with childhood apraxia of speech (CAS) present with significant speech production deficits, the effects of which often persist well into late childhood (American Speech-Language-Hearing Association, 2007; Lewis, Freebairn, Hansen, Iyengar, & Taylor, 2004). Debate has historically surrounded whether the features of CAS are the result of an impairment in linguistic or speech motor systems, or both (American Speech-Language-Hearing Association, 2007). Most research, however, has failed to explicitly consider a developmental perspective of the disorder, arguably limiting the associated interpretations that often (implicitly) assume an established underlying system (Maassen, 2002). One of the key tenets of such a developmental perspective is the possibility of an original core deficit in one system, with negative consequences for aspects of the system that subsequently develop.

A mixed-methodology paradigm was employed in the present research in order to explore the core deficit in CAS. Similar paradigms have been applied to the study of dyslexia (Koster et al., 2005; Lyytinen et al., 2001; Viholainen et al., 2006) and autism spectrum disorders (Coonrod & Stone, 2004; Dawson, Osterling, Meltzoff, & Kuhl, 2000; Iverson & Wozniak, 2007), but have yet to be applied to CAS.

Study 1 sought to quantify parental report of vocalisation behaviours in children with a clinical diagnosis of CAS. The parents of 20 children with suspected CAS (sCAS) completed a questionnaire focussing on the prelinguistic development of their children as infants. Responses were compared to those from parents of 20 children with Specific Language Impairment (SLI) and 20 children with typically developing (TD) speech and language development. The sCAS children were reported to be significantly less vocal, less likely to have babbled, later in the emergence of first words and later in the emergence of two-word combinations than the TD children. However, the SLI children were reported similarly on many (but not all) items. Despite this similarity, the sCAS group were unique in terms of the presence of reported babbling (35% were reported *not* to have babbled at all, compared to the TD

and SLI children who were all recalled as having babbled in infancy), and the emergence of two word combinations (significantly later than both the TD *and* SLI groups). In addition, the motor milestones of age of crawling and age of walking were significantly correlated with age of emergence of two-word combinations in the sCAS group, suggesting commonly constrained speech and motor development. Overall, the results provided preliminary support for the notion of atypical prelinguistic vocal development in children with sCAS, and highlighted the importance of further research on the topic.

Study 2 applied a retrospective data paradigm in exploring the prelinguistic vocal development of children with CAS. Nine clinically-ascertained children, aged 3 to 4 years and presenting with a range of speech and language profiles (including 3 with suspected CAS), were characterised in terms of operationally-defined CAS characteristics in the first stage (2A) of this study. The battery of tasks included standardised speech and language assessments as well as non-standardised tasks targeting speech production ability. A group of 21 age-matched children with typically developing speech and language skills provided comparison data for the non-standardised tasks. This phase of the study documented CAS characteristics in five of the nine clinical sample participants, with two of these children showing all five of the features investigated. Study 2B examined the early speech, language and motor development of the clinical sample children, via analysis of data available retrospectively for this unique group of children. Their infant profiles were compared to those of 205 infants who had been part of the same community program that the clinical sample had been involved in (and thus had infant data available) but who did not have identified ongoing speech and language issues. Single case comparisons (Crawford & Garthwaite, 2005) revealed that the child with the greatest number and severity of CAS features at preschool age demonstrated significantly poorer expressive skills and a significant dissociation in receptive-expressive abilities in infancy, compared to the typically developing children. Profiles for the other clinical sample children varied considerably.

In the third study (Study 3), the development of infants with a family history of CAS (n = 8) was compared to that of infants with no such familial risk (n =8) to further examine the proposed core deficit in CAS. Early speech, language and motor

development was tracked at 9, 12, 15, 18 and 24 months. The siblings as a group demonstrated significantly poorer expressive language, speech sound development and fine motor ability than the comparison group, consistent with the notion of a verbal trait deficit (Lewis, Freebairn, Hansen, Taylor et al., 2004). At two years of age, two siblings (and none of the comparison infants) showed clinically-important delays in speech and language development. Inspection of their profiles suggested one infant (SIB2) to present with features consistent with putative early features of CAS (Davis & Velleman, 2000); the other (SIB1) to present with language difficulties not suggestive of CAS. Analysis of their vocalisation samples revealed that while SIB2's rate of vocalisations at 9 months was not different to that of the comparison group, the nature of the vocalisations were different. While all comparison infants were using canonical syllables at 9 months, SIB2 had not entered this important stage until 12 months, and showed a significantly reduced proportion of canonical syllables at this age (2.5% compared to the comparison infants, who averaged 17%, with none producing less than 6%). Acoustic analyses performed on prelinguistic canonical syllables showed that while duration did not differ, a restricted use of the F1:F2 planar space was noted for SIB2 compared to the typically developing infants, suggesting limited vowel production. Furthermore, a particularly strong correlation between F1 and F2 was observed, suggesting stronger coupling of the articulators. Importantly, the vocalisation data, together with data from standardised assessments, showed a dissociation between speech motor and conceptualiser areas, with a deficit in speech motor control evident in the context of intact conceptual skills for this infant. In contrast, SIB1 (who showed a language-delayed profile at 2 years, with no CAS features) did not evidence the types of anomalies identified for SIB2.

Taken together, the results of the present research provide support for the viability of a speech motor control deficit account of CAS, when interpreted in a developmental context. As such, they highlight the importance of the prelinguistic period and longitudinal investigations in examining the underlying core deficit in CAS, and suggest important implications for theoretical and clinical conceptualisations of the disorder.

TABLE OF CONTENTS

LIST OF TABLES

LIST OF FIGURES

OVERVIEW

"Spoken language is one of the greatest achievements of childhood, for it opens the door to a variety of educational and social experiences" (Kent, 2000 p. 391). Not only is it one of the most important, speech is also one of the fastest discrete human motor skills, involving many muscle fibres and relying on precise neuronal control. Unfortunately, not all children learn to speak with the ease that is expected. This research focuses on one group of children who have a particular and persistent difficulty with speaking – those with childhood apraxia of speech (CAS).

This thesis explored the argument that one of the main difficulties in identifying the core deficit in childhood apraxia of speech relates to the interactive nature of development. In particular, that the array of features observed in this clinical population relates, in part, to the unfolding nature of speech and language development. Many current conceptualisations of CAS do not explicitly consider this issue, arguably limiting the progression of research into diagnosis and treatment. However, considering a developmental model of speech and language development allows hypotheses relating to the very earliest features of the disorder to be proposed and tested (Hodge, 1994; Maassen, 2002; Stackhouse & Snowling, 1992). The present thesis applied a combination of methodologies to address these hypotheses.

Childhood apraxia of speech is described and discussed in detail in the first chapter, providing a background for the current research. The chapter also explores the pitfalls of using models of established systems to interpret CAS, and presents key arguments for using a developmental model to interpret findings relating to the disorder. The proposed underlying deficit in CAS is discussed, and potential manifestations at the very earliest stages of speech development are presented.

Chapters 2, 3 and 4 document the three studies comprising the present research, each addressing hypotheses relating to the core deficit in CAS from different standpoints. Study 1 describes the retrospective parent report of early vocalisations in children

with CAS, compared to that of children with specific language impairment (SLI) and those with typically developing speech and language skills. This study, which was conducted early in the PhD process, sought to quantify parent report of vocal behaviours in children with CAS, establishing the viability of launching into the time-intensive studies that followed. The following chapter (Study 2) considers data from children with various speech and language profiles, including some with CAS features. Operationally defined CAS features were described to characterise the sample, before corresponding infant data available for these children are investigated. Chapter 4 then describes a prospective, longitudinal study (Study 3) of infants who are siblings of children with CAS. In this study, general development and communication assessments, vocalisation and acoustic data are presented as evidence addressing the core thesis outlined herein. The final chapter draws together the results of the three studies, and in light of the strengths and limitations of the present research, considers the theoretical and clinical implications of these findings.

CHAPTER 1

CHILDHOOD APRAXIA OF SPEECH

"Developmental apraxia of speech is a label in search of a population" (Guyette and Diedrich, 1981, p 39).

"The Committee recommends that childhood apraxia of speech be recognized as a type of childhood (pediatric) speech sound disorder that warrants research and clinical attention"(American Speech-Language-Hearing Association, 2007, p. 3).

In the space of over twenty five years, and despite controversy and debate surrounding the phenotype (Chappell, 1984; Hall, 2003a; Shriberg, Aram, & Kwiatkowski, 1997a), nature (Crary & Towne, 1984; Hall, 2003b; Stackhouse, 1992) and differentially diagnostic features (Davis, Jakielski, & Marquardt, 1998; Shriberg, Aram, & Kwiatkowski, 1997b) of CAS, the importance of uncovering the underlying deficit giving rise to the varied symptoms observed in children with the disorder remains. During this time frame, there have been significant leaps in our understanding of typical speech and language development, and of communication disorders in general. Despite much debate, there is presently consensus among CAS researchers for the existence of the disorder, and for the urgent need for research into diagnosis, early features, and treatment (American Speech-Language-Hearing Association, 2007).

Terminology

Many terms have been used to label the speech sound disorder that is the focus of this thesis. The particular label applied often (but not always) reflects the researcher's theoretical background and assumptions about the disorder, and/or geographical and historical influences. Developmental verbal dyspraxia (DVD), developmental apraxia of speech (DAS), speech dyspraxia, apraxia of speech (AOS), developmental articulatory dyspraxia and childhood apraxia of speech (CAS) are

terms that have been used variously over the years and across continents, presumably labelling the same type of speech disorder in children (American Speech-Language-Hearing Association, 2007; Shriberg, Aram, & Kwiatkowski, 1997a; Stackhouse, 1992).

The term *praxis* refers to "the ability to plan and execute a skilled movement" (Goodgold-Edwards & Cermak, 1990, p. 431). Borrowing from the adult acquired communication disorders field, the term *apraxia* was initially applied to reflect the similarities with apraxic conditions observed in adults post-stroke or other neurological insult. The prefix *dys-* (in *dyspraxia*), however, is often used to reflect that some praxis is still possible (i.e., there is not a total loss in function). This variant also parallels motor or movement dyspraxia, the term used by occupational therapists to describe motor planning deficits (Dewey 1995). Childhood apraxia of speech (CAS), a term that until recently was most widely used in the United States, reflects and emphasises that although the disorder is identified in childhood, it is not 'developmental' in the sense of being a condition the child will 'grow out of' or overcome as development progresses.

The qualifier *suspected* has also been used in labels of CAS (Shriberg et al., 1997a), highlighting the tentative nature of the diagnosis in many cases, the absence of a clear set of validated diagnostic criteria, and to enable increased consistency amongst international researchers and clinicians. Following this convention[1], the term 'suspected childhood apraxia of speech (sCAS)' was adopted in the first study of this thesis to describe participants with features consistent with CAS. 'Childhood apraxia of speech (CAS)' is used when discussing the disorder in its conceptual and theoretical sense, except where direct quotes from other sources are used.

[1] The draft childhood apraxia of speech technical report released by ASHA initially used this terminology (ASHA, 2006)

Definition and Description

Numerous and wide-ranging definitions of CAS have been used in the research literature. Definitions have focussed on describing the clinical characteristics thought to define the disorder and core deficits hypothesised to underlie these symptoms. Many definitions have emphasised the motor planning aspects of speech production that children with the disorder have difficulty with (e.g., a "disorder in the programming of articulatory movements", Marquardt, Sussman, Snow, & Jacks, 2002, p. 31). Some definitions have included specific exclusionary criteria, highlighting, for example, that the speech difficulties occur in the context of normal intellectual functioning or in the absence of any frank neurological impairment (Williams & Inghman, 1981), whilst others have focussed only on inclusionary features.

For the purposes of the present research, the following working definition proposed by the American Speech-Language-Hearing Association (2007) in the recent CAS technical report will be used:

> *Childhood apraxia of speech (CAS)* is a neurological childhood (pediatric) speech sound disorder in which the precision and consistency of movements underlying speech are impaired in the absence of neuromuscular deficits (e.g., abnormal reflexes, abnormal tone). CAS may occur as a result of known neurological impairment, in association with complex neurobehavioral disorders of known or unknown origin, or as an idiopathic neurogenic speech sound disorder. The core impairment in planning and/or programming spatiotemporal parameters of movement sequences results in errors in speech sound production and prosody. (p. 3)

The definition recognises key features commonly observed in children with CAS and focuses on the presumed core deficit in speech movement ability. Whilst CAS may occur in conjunction with other disorders (American Speech-Language-Hearing Association, 2007; Hodge, 1994), in its idiopathic form (the focus of the present thesis) there are no identified etiological causes. In typical accounts of CAS, affected children are thought to know what they want to say, but have extreme difficulty

producing intelligible speech, despite the absence of structural abnormalities or peripheral weakness.

Symptomatology

A broad range of characteristics have been suggested to be part of the symptom complex of CAS. These include inconsistency and high variability in speech production, prosodic anomalies (such as a tendency to stress unstressed syllables), vowel errors, increasing error rate with increasing length and phonetic complexity, sequencing difficulties, limited phonemic repertoires, and simple syllable shapes (Davis et al., 1998; Marquardt, Jacks, & Davis, 2004). Additional features such as groping behaviours, heightened awareness of the unintelligibility of the child's own speech and the spontaneous development of gesture systems to compensate are also often reported (Forrest, 2003; Hall, 2003c).

In parallel with debate and variations in terminology and definitions, there has been much discussion about the core features that comprise the disorder. Depending on whether the focus is on identification of CAS as a diagnostic category (and thus inclusivity) or more specifically on differential diagnosis (and thus features which differentiate the disorder from those with similar symptoms), varied core features have been proposed. Some features reflect clinical observations, and at times are difficult to operationally define and measure (groping, for example). Others focus on the presumed deficit underlying the symptoms (e.g., sequencing difficulties). Many lists include all common symptoms observed in children with CAS, regardless of whether they are also features of other speech sound disorders (e.g., consonant errors), whilst others focus on only those features thought to be differentially diagnostic and thus specific to CAS (e.g., vowel errors).

Diagnostic Issues

The diagnosis of CAS by speech pathologists has typically been relegated to a process of exclusion, whereby diagnostic 'checklists' are used to distinguish the disorder from other speech and/or language impairments such as specific language impairment, and (particularly) phonological disorder/s (Hodge, 1994; Stackhouse, 1992). Shriberg and colleagues (2003) highlighted the constraints and psychometric

issues associated with diagnosis via checklists, both for finding a phenotype and genotype for the disorder, and for identifying the population for clinical and research purposes. Highlighting such issues, Forrest (2003) reported 50 different characteristics described by speech pathologists in establishing a diagnosis of CAS. Of these, however, six were predominant: inconsistent productions, general oral-motor difficulties, groping, inability to imitate sounds, increasing difficulty with increased utterance length, and poor sequencing of sounds.

Systematic research programs have attempted to reveal a differentially diagnostic 'marker' or set of markers for CAS. Shriberg and colleagues (Shriberg, 2003; Shriberg et al., 1997a; Shriberg et al., 1997b; Shriberg, Aram, & Kwiatkowski, 1997c; Shriberg, Campbell et al., 2003; Shriberg, Green, Campbell, McSweeny, & Scheer, 2003) have investigated a number of measures in terms of their ability to differentially mark CAS, including phonological, prosodic and acoustic features. Of all of the potential diagnostic markers investigated, the assignment of lexical stress was the only differentiating feature for CAS found in the cohort investigated. (Shriberg, Aram, & Kwiatkowski, 1997c). Importantly, however, this differentiating feature was only present for half of the children who had been suspected to have CAS.

In addition to prosodic features such as lexical stress, candidate characteristics that have been proposed as being potentially differentially diagnostic have included vowel errors, inconsistency, sequencing difficulties, and increasing errors with increasing length and complexity (American Speech-Language-Hearing Association, 2007). These features are considered in more detail in Chapter 3 in the context of participant description for Study 2.

Many features proposed to be characteristic of CAS, however, are also reported in the general paediatric speech-impaired population (McCabe, Rosenthal, & McLeod, 1998). For example, children with phonological disorder/s may similarly show a limited phonemic repertoire (Broomfield & Dodd, 2004). Inconsistency in production, often reported to be specific to CAS, is a characteristic of a proposed subtype of phonological disorder – inconsistent phonological disorder – in one diagnostic classification system (Dodd, 1995). Not all classification systems

acknowledge this subtype, however (Shriberg, 2003). Differentiating CAS from phonological disorder has been a major research and clinical challenge, despite some consensus for the existence of different types of speech-sound disorder (Shriberg, 2003). Developmental dysarthria, although previously considered as sharing less overlapping features with CAS, has also been identified as being potentially co-morbid in many children with CAS (American Speech-Language-Hearing Association, 2007), furthering the diagnostic challenge. Mirroring conceptualisations of other developmental disorders, a number of researchers have also proposed the notion of a continuum of features for CAS (Crary & Towne, 1984). Such conceptualisations suggest that rather than representing discrete categories, speech-sound disorders may reflect a continuum of motoric involvement (Strand, 2003).

Maassen (2003) commented that the overlapping symptomatology for various speech disorders often limit the inferences that can be drawn from them. Despite this overlap (or perhaps *because* of it), there is still a strong desire to differentiate CAS from other speech and language difficulties, both clinically and in the research literature. One key factor is the belief that the nature of the underlying deficit may be different, as well as the genotypes (Shriberg et al., 2003). Clinically, the diagnosis of CAS has implications for prognosis and therapy (Hall, 2003d). Progress is often reported to be slower for children with CAS (Hall, 2003d). Children who do not receive an accurate diagnosis may receive treatments that fail to target the nature of the deficit (Strand & Debertine, 2000; Velleman, 2002). Given the protracted and broad nature of the disorder's effects, the long term needs of the child need to be considered from an early age (Lewis, Freebairn, Hansen, Iyengar, & Taylor, 2004).

Despite attempts to identify differentially diagnostic markers for CAS, at present a validated set of features that reliably differentiate CAS from other speech sound disorders is lacking (American Speech-Language-Hearing Association, 2007). However, expert opinion, based on a systematic review of the literature, currently suggests features such as impaired performance on tasks involving multiple syllables (e.g., diadochokinesis, non-word repetition and multisyllabic word production tasks) and tasks involving prosodic variables as being more specific to CAS (American Speech-Language-Hearing Association, 2007).

Reflecting the lack of a set of validated differentially diagnostic features, the method of identification of CAS participants for research studies has varied. Many researchers have identified participants based on the presence of a list of commonly reported features (Lewis, Freebairn, Hansen, Iyengar et al., 2004; Skinder, Strand, & Mignerey, 1999). How these features are quantified and measured, and the exact number of features required to meet the diagnosis are often not specified. Even in instances where a certain number of features are specified (Davis et al., 1998), participants may vary in which features they demonstrate. The ad hoc committee on CAS conceded that even the features that have gained consensus in the literature may not be necessary and sufficient for a diagnosis of CAS (i.e., it is not necessarily the case that all features must be present for a CAS diagnosis to be valid). Furthering the difficulty in identifying homogeneous groups, the features characteristic of CAS also change over time (American Speech-Language-Hearing Association, 2007). It is also likely that a number of features are cross-correlated – that is, some reported features may reflect the same underlying deficit, yet be reported in slightly different ways. Reporting a high incidence of vowel errors and limited vowel phonemic inventory, for example, are features likely to be closely associated.

Expert researcher or clinician opinion is another method used in identifying participants for CAS research. In many studies, researchers have used participants for whom a 'clear' diagnosis of CAS has been established by either the referring speech pathologists, or the researching speech pathologists (Jacks, Marquardt, & Davis, 2006; Munson, Bjorum, & Windsor, 2003). In some studies, participants are initially identified via clinical diagnosis, but are described further or additional criteria are applied (Nijland, Maassen, & van der Meulen, 2003; Nijland, Maassen, van der Meulen et al., 2002, 2003). Although there is some overlap, the additional criteria (and importantly, the way they are measured) may not be consistent across research groups (reflecting the lack of a set of validated diagnostic criteria). It has been suggested that many clinical features proposed to be diagnostic of CAS may eventually be shown to be those with scientific validity (American Speech-Language-Hearing Association, 2007). Identification of participants via expert opinion may therefore be a justifiable method in the absence of validated criteria (American Speech-Language-Hearing Association, 2007; Dodd, 2007).

Another method of participant identification in CAS research avoids making a-priori assumptions about the nature of the speech sound disorder and instead investigates patterns of task performance in a relatively heterogeneous group of children (Peter & Stoel-Gammon, 2008). Such studies, although informative, are limited in their ability to investigate specific hypotheses regarding each disorder. Regardless of how CAS is identified, researchers have acknowledged the need for detailed participant description, especially while diagnostic criteria are still being confirmed (American Speech-Language-Hearing Association, 2007).

Co- morbidities and Associated Features

A number of additional symptoms or features have been commonly reported in cases of CAS. These include language deficits, difficulties with literacy-related tasks, and motor coordination impairments. Deficits have been interpreted as either being commonly co-morbid with CAS, secondary effects of the core deficit underlying CAS, or part of the symptom complex of the disorder itself.

Language Deficits

Expressive language difficulties are commonly reported in children with CAS (Hall, 2003c). Language areas reported to be affected include vocabulary acquisition and expansion (Davis & Velleman, 2000; Hall, 2003c), general expressive language ability (Lewis, Freebairn, Hansen, Iyengar et al., 2004), and syntactic skills (Ekelman & Aram, 1983). For example, the use of grammatical markers was impaired in a group of 8 children with CAS studied by Ekelman and Aram (1983). Language impairments (defined by clinician report) were present in 9 out of 11 children with CAS studied by Thoonen et al. (1997). Although most accounts of CAS acknowledge the frequent presence of language difficulties, the nature of and explanation for the linguistic impairments is often debated. Some researchers have hypothesised an over-arching deficit in organising and sequencing linguistic units to account for the syntactic and speech production errors observed in CAS (Velleman & Strand, 1994).

Whether receptive language deficits co-occur in children with CAS has been a more controversial issue. Some studies investigating CAS have explicitly excluded children who show evidence of receptive language difficulties, reflecting beliefs that the core underlying deficit (in CAS) does not involve or impact on comprehension. Inclusion criteria for Ekelman and Aram's (1983) study, for example, included a requirement for normal lexical comprehension. Similarly, normal receptive language was an inclusion criterion for Marion et al.'s (Marion, Sussman, & Marquardt, 1993) and Skinder, Strand and Mignerery's (1999) studies of CAS children. In contrast, other researchers have included children with receptive language difficulties (Shriberg et al., 1997b), either interpreting these impairments as being co-morbid or a sequalae of expressive difficulties. Despite the inconsistency in inclusion criteria, most researchers and clinicians report a 'receptive-expressive gap', with many children with CAS reported to demonstrate a relative strength in receptive language (Lewis, Freebairn, Hansen, Iyengar et al., 2004; Stackhouse, 1992).

Pre-literacy/literacy Difficulties

A number of studies have documented literacy-related difficulties for children with CAS (Hall, 2003c; Lewis, Freebairn, Hansen, Iyengar et al., 2004; Marion et al., 1993). Difficulties with phonological awareness tasks, themselves linked to subsequent literacy acquisition, have been commonly reported. Children with CAS have been shown to perform poorly on rhyme detection, judgement and production tasks; word segmentation tasks; spelling and decoding; and other literacy-related skills (Lewis, Freebairn, Hansen, Iyengar et al., 2004; Marion et al., 1993). There is also evidence to suggest that phonological awareness skills in children with CAS can be improved with targeted therapy (Moriarty & Gillon, 2006). However, most researchers acknowledge that the literacy difficulties experienced by children with CAS are likely to be directly related to impaired phonological awareness skills, as is seen in children with other speech sound disorders, rather than a core part of the symptom complex of CAS itself (cf. Marion et al., 1993).

Motor Co-ordination Deficits

Another reported observation of children with CAS is that many affected children also show difficulties with motor co-ordination (Davis & Velleman, 2000; Hall, 2003c; Hill, 2001). There have been few studies that have explicitly examined this

issue specifically in children with CAS. In a rare investigation of motor skills in children with a range of speech disorders that included children with CAS, Bradford and Dodd (1996) reported children with a diagnosis of CAS to display significant difficulty on fine motor subtests of a standardised assessment. An increased rate of motor coordination difficulties has also been found in children with a range of speech and language disorders, however (Archibald & Alloway, 2008; Hill, 2001), making the nature of the association between motor impairment and CAS less clear. Some studies suggest motor co-ordination deficits to be more prevalent in children with speech, rather than language, impairments (Bishop, 2002). Proposed explanations for the apparent co-morbidity of CAS and other movement / motor co-ordination disorders will be discussed later in this chapter.

Other Co-morbidities

Difficulties with feeding, oral-motor movements, nasal resonance, and perceptual skills are also described in some reports of children with CAS (Hall, 2003c). These observations are usually based on clinical descriptions and few studies have examined such features in detail. Of those that have been researched, deficits in auditory perception (Bridgeman & Snowling, 1988), and fine-grained auditory discrimination of consonants (Groenen, Maassen, Crul, & Thoonen, 1996) and vowels (Maassen, Groenen, & Crul, 2003) have been documented. Whether these associated characteristics are part of the core symptom complex of CAS, or are secondary consequences, is not yet clear.

Prevalence

In the absence of epidemiologically-ascertained population data, the prevalence of CAS in the general population has been estimated at approximately 1 to 2 in 1000, or 0.1 - 0.2% (Shriberg et al., 1997a), based on clinical referral data. The prevalence in clinical samples is often reported to be higher (e.g., 3.4 - 4.3% in a US study, Delany & Kent, 2004, as cited in American Speech-Language-Hearing Association, 2007). By comparison, language impairment is thought to affect approximately 7% of school-age children (Tomblin, Records, Buckwalter, Zhang, & Smith, 1997), and speech sound disorders estimated to affect 3 to 4% (Shriberg & Tomblin, 1999).

CAS is therefore a challenging disorder to research, given the relative infrequency of its occurrence.

Heritability

CAS, like other speech and language impairments, tends to run in families, with more males affected than females (Maassen, 2002).The apparent high heritability of CAS has been investigated by few researchers, however. Early research suggested that as many as 67% of children with CAS have a first degree relative with a speech and/or language disorder (Morley, 1965). This familial aggregation of speech and language deficits in individuals with CAS has been subsequently supported in the small number of studies that have addressed the issue. Thoonen and colleagues (Thoonen, Maassen, Gabreels, Schreuder, & de Swart, 1997), for example, found that six out of their 11 children with CAS had a family history of speech and language disorders. More recent research has suggested that as many as 86% of children with CAS have at least one first-degree relative with a speech/language disorder (Lewis, Freebairn, Hansen, Iyengar et al., 2004).

Whilst it is clear that many, if not most, individuals with CAS have a family history of speech and/or language disorder, the heritability of CAS as a specific disorder is still under investigation. When looking at familial aggregation in CAS research, a number of factors affect the interpretation of results. Apart from the issue of establishing a differential diagnosis in the participants under investigation, familial aggregation studies also face the challenge of how to identify 'affected' family members. Most research considers a broad phenotype, interpreting any speech and/or language disorder as indicating 'affectedness'. Deficits in family members are identified either by direct testing or by way of self- or parent- report (Lewis, Freebairn, Hansen, Taylor et al., 2004). Rarely, a more specific phenotype of an unambiguous CAS diagnosis is required for family members to be considered affected, which is challenging to document for siblings and, especially, parents. In the only such study to date, Lewis et al. (2004) reported that two of their 22 participants with CAS had a sibling with features consistent with the same diagnosis. This represents an affection rate of 9%, considerably higher than that estimated for the general population (Shriberg et al., 1997a).

Genetic studies also suggest CAS to be heritable. Programmatic research has been conducted on a large British family (known as the KE family), in which 15 of the 30 members (over 4 generations) are reported to present with a speech and language deficit that was initially characterised as 'verbal dyspraxia' (Alcock, Passingham, Watkins, & Vargha-Khadem, 2000b; Alcock, Passingham, Watkins, & Vargha-Khadem, 2000a; Watkins, Dronkers, & Vargha-Khadem, 2002; Watkins, Vargha-Khadem et al., 2002). An abnormality (specifically, a translocation) in the FOXP2 gene has been identified in these individuals. Whilst these studies have suggested an autosomal-dominant mode of genetic transmission for CAS, a number of cognitive, cranio-facial and other anomalies have since been reported in the KE family members (Vargha-Khadem, Gadian, Copp, & Mishkin, 2005). Thus it is not clear how the results may generalise to other cases of CAS.

However, FOXP2 has also been implicated as having a primary role in speech and language ability (Corballis, 2004; Vernes et al., 2006). When this gene is disrupted, vocal learning capacity has been shown to be limited in humans and songbirds alike (Haesler et al., 2007). MacDermot et al. (2005) reported an anomaly in FOXP2 in a child with CAS, and found the same irregularity in the child's sibling and mother. Although such genetic anomalies have not been universally found in clinical populations (MacDermot et al., 2005), research such as this supports the notion of a possible genetic basis for CAS.

Theoretical Perspectives

The identification of CAS as a theoretical and clinical diagnostic category has been preceded and greatly influenced by the adult neurological literature and associated theoretical perspectives. Reference to adult patients demonstrating isolated speech production deficits in the absence of muscular weakness were made as early as the mid 1800s. The term apraxia was used the following century to describe motor planning difficulties affecting motor movements of the limbs, and the possibility of an analogous difficulty in speech production motivated a re-think of diagnostic categories associated with acquired speech-language disorders (Duffy, 2002). Darley (1968, as cited in Duffy, 2002), formalised the first set of clinical characteristics for

the disorder which was neither aphasia nor dysarthria, termed apraxia of speech (AOS): groping for correct positioning of articulators, clumsiness in finding correct patterns of movement for polysyllabic words, and phonemic near misses and retrials, all in the context of normal auditory comprehension and written expression. A motor planning or programming deficit was hypothesised to underlie the observed speech characteristics. Although motoric explanations have continued as the dominant theoretical perspective of AOS, alternative linguistic accounts hypothesising higher-level deficits have also been proposed at various points (Duffy, 2002).

A developmental equivalent to AOS was identified in the early 1950s, when Morley and colleagues described a group of 12 children who displayed difficulty ('clumsiness' or 'awkwardness') with complex and rapid articulatory movements, but who otherwise showed normal ability to produce voluntary movements of the lips, tongue and palate. They applied the term articulatory dyspraxia and offered the following definition:

> A defect of articulation which occurs when movements of the muscles used for speech…. appear normal for involuntary and spontaneous movements …. or even for voluntary imitation of movements …, but are inadequate for the complex and rapid movements used for articulation and reproduction of sequences of sounds used in speech (Morley, 1965, as cited in Stackhouse, 1992, p. 20).

Description of potential neurological correlates and diagnostic characteristics of CAS ensued. In parallel with the debate about the nature of and explanation for acquired apraxia, researchers deliberated on potential core deficits underlying CAS. As with AOS, the predominant perspective identified CAS as a motoric disorder, affecting motor planning ability. However, in line with AOS, the high frequency with which language deficits were identified in children with CAS often led to debate as to whether the seemingly motoric symptoms of CAS could be more parsimoniously explained by linguistic or higher-order explanations. As seen below, debate about the nature of the underlying deficit has continued.

Underlying Deficit/s

Both historically and in more current research, models of adult spoken word production are often referred to when attempting to interpret the features and underlying core deficit/s of CAS. Figure 1 outlines an adapted version of one such model, the WEAVER (Word-Form Encoding by Activation and VERification) model (Roelofs, 1997), expanding on Levelt's (1989) earlier model. In this model, speech production begins with conceptualisation, where ideas, thoughts and intentions are specified. Lexical concepts appropriate to the intention are activated and corresponding lemmas (which consist of syntactic information) are selected. The selected lemmas are slotted into the appropriate section within the utterance's syntactic frame. Within this frame, the internal structure of the word is accessed, in a process referred to as morpho-phonological encoding. Information about the word's morphological properties, its metrical shape (number of syllables and main stress position), and segmental aspects is retrieved during this process. The output is a phonological word – a section of speech that contains one main stressed syllable and any associated weak syllables, with segmental content of the syllables specified. Although the real life boundaries between linguistic and speech motor processes are (justifiably) less clear than is depicted in a model, these processes (from conceptualisation to the retrieval of the phonological word) are often conceptualised as being linguistic in nature.

During the next stage of the WEAVER model, phonetic encoding (the equivalent of 'motor planning' in other models), the gestural score of the phonological word is specified. This part of the production process involves accessing a repository of gestural scores (the syllabary) that is assumed to be available for frequently used syllables. Gestural scores specify articulatory gestures (such as lip protrusion) and their temporal relationships. The articulation system executes the gestural score, resulting in overt speech (Browman & Goldstein, 1992). These processes (transforming the phonological word into overt speech) are typically considered as those involved in speech motor control.

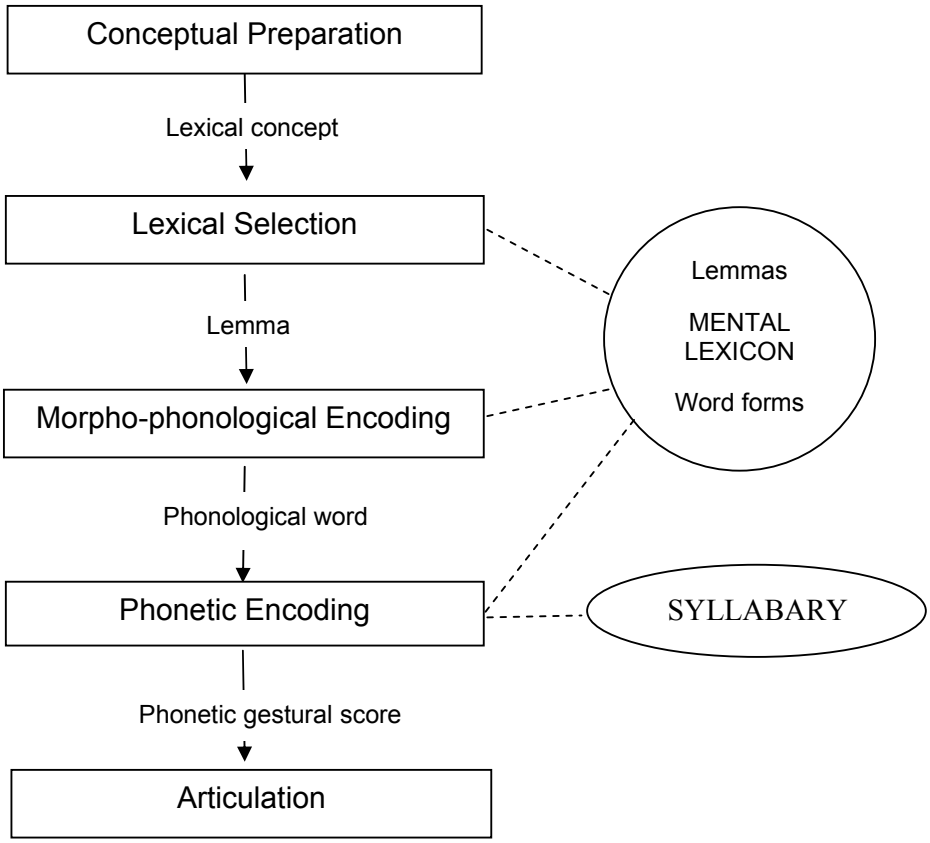


Figure 1. An adapted version of the WEAVER model of speech production (Roelofs, 1997).

In this and many other models of speech and language processing, language impairment (in children) and aphasia (in adults) are hypothesised to reflect deficits in processing or representations at the levels of conceptualisation, lexical selection and access, and/or morpho-phonological encoding (Dell, Schwartz, Martin, Saffran, & Gagnon, 1997; Gathercole & Baddeley, 1990). Dysarthria, in contrast, is typically conceptualised as a speech sound disorder caused by weakness of the peripheral musculature, and thus is isolated to the level of articulation (execution in other models). Historically, apraxia has often been conceptualised as the disorder in-between – a difficulty caused not by language impairment or musculature weakness, but by deficits primarily in motor planning (e.g., phonetic encoding in the present model). There have been many alternative explanations, however. The main long-running debate in the CAS literature has involved whether the underlying deficit is primarily of a 'motoric' or 'linguistic' nature (e.g., Crary & Towne, 1984). In the context of the WEAVER model, the question can be conceptualised as whether CAS

can be attributed to 'lower-level' speech motor control processes involving phonetic encoding and articulation (e.g., compiling, accessing, and executing gestural scores), or to 'higher-level' linguistic processes involved in, for example, forming or accessing and retrieving phonological representations.

Speech Motor Control Deficits

A deficit in speech motor control has been implicated by a number of researchers examining CAS (e.g., Kent, 2000; Nijland, Maassen, & van der Meulen, 2002). Kent (2000) described speech motor control as encompassing the processes and systems involved in transforming a phonologic representation of language into an acoustic signal (comprised of phonetic encoding and articulation in the WEAVER model). Nijland and collaborators demonstrated that children with CAS produced "idiosyncratic coarticulation patterns", reflected in F2 (second formant) ratios that were different to normally speaking children and adults (Nijland, Maassen, van der Meulen et al., 2002); as well as poorer compensation for a bite block (Nijland, Maassen, & van der Meulen, 2002; Nijland, Maassen, & van der Meulen, 2003). These studies suggest that the gestural control associated with perceptually normal sound production is deviant in children with CAS, consistent with a deficit in the phonetic realisation of speech sounds. Acknowledged by the researchers, the specificity of conclusions to CAS are limited because the studies involved comparison of children with CAS and age-matched typically developing children (i.e., no comparison was made with children with other speech difficulties).

A growing body of research demonstrating prosodic anomalies in children with CAS lends further support to a deficit in speech motor control. It has long been noted that children with CAS don't 'just' have difficulties with the segmental aspects of speech. Often, these children are described clinically to have 'staccato' speech and to be perceived as putting equal stress on multisyllabic words (Odell & Shriberg, 2001). These prosodic issues often persist even when other aspects of speech (e.g., phonological inventory and syllable shape use) have improved (Velleman & Shriberg, 1999). As indicated earlier, the assignment of lexical stress was the only measure found by Shriberg and colleagues to differentiate a group of children with CAS from those with phonological delays. Maassen (2002) suggested that

inappropriate stress "might stand out as the first candidate to serve as a diagnostic marker" for children with CAS (p. 262).

In investigating such prosodic anomalies, Skinder, Strand and Mignerey (1999) found that their group of 5 children with CAS were perceived by trained listeners to less accurately mark syllabic stress than the control children. However, this perceptual difference was highly variable, and not reflected in the acoustic measures of fundamental frequency and amplitude. Similarly, Munson, Bjorum, and Windsor (2003) investigated the perceptual and acoustic parameters of stress assignment in children with CAS and those with phonological delays. The CAS children were judged as being less able to produce correct stress on nonwords of varying stress patterns. Despite this, the difference was not reflected in the acoustic measures, which included vowel duration, fundamental frequency at vowel midpoint, and intensity at vowel midpoint.

Consistent with the perceptual findings above, Nijland et al (2003) found that 5 year old children with CAS did not make durational differences between iambic (weak-strong pattern of stress) and spondaic (equal stress) utterances, whereas normally speaking children did. Similarly, Skinder, Connaghan, Strand and Betz (2000) found both perceptual and acoustic correlates (in peak fundamental frequency and amplitude) of a lexical stress deficit in 4-8 year old children with CAS. Furthermore, Odell and Shriberg (2001) demonstrated that children with CAS produced a high proportion of utterances that were deemed to have inappropriate stress (specifically – excessive-equal stress).

However, finding differences in gestural control or prosodic anomalies in children with CAS, whilst suggesting a deficit in speech motor control, does not clearly distinguish which aspect/s of speech motor control is/are impaired. The deficit could potentially lie with establishing or forming appropriate gestural scores (motor programs), similar to the deficit proposed by Varley and Whiteside (2001) to be involved in acquired apraxia of speech. An inability to program extended units of speech may have consequences for producing appropriate rhythmic patterns. It could also lie with later stages of execution. Peters, Hulstijn and Lieshout (2000) have proposed an additional stage subsequent to phonetic encoding, involving integration

of segmental and suprasegmental (i.e., prosodic) features prior to execution. Temporal (rate and force) parameters within the particular speech context are set at this stage. It is possible therefore that the prosodic difficulties displayed by children with CAS reflect a deficit at this level. However, stress related anomalies could also be reflective of higher level linguistic impairment, as described below.

Linguistic Level Deficits

A number of researchers have suggested that linguistic deficits, rather than motoric factors, may account for the symptoms in CAS. For example, deficits in the quality of or access to phonological representations, (i.e., the representations stored in the lexicon or the process of morpho-phonological encoding in the WEAVER model) have been proposed. Marion and collaborators (Marion et al., 1993) found that children with CAS were significantly poorer at producing, recognising and judging rhyming words than typically developing children, and argued that an underlying deficiency in phonological representations is the core locus of the deficit in CAS. Similarly, Marquardt, Sussman, Snow and Jacks (2002) reported difficulties identifying the number of syllables in words and judging positions of sounds within syllables in children with CAS. Forrest and Morrisette (1999) found that the pattern of feature retention (i.e., the features of the adult target sounds that are 'retained' when the children make 'errors') in children with phonological delays were the same as children with CAS, with both groups of children retaining voice, then manner, then place of articulation last. This suggested similarities in the quality (or lack of) of phonological representations of the two groups.

Furthermore, the lexical stress deficit noted by Shriberg and colleagues was initially interpreted as evidence of a deficit involving phonological representations (Shriberg et al., 1997c). Citing evidence of limited groping and self-correction attempts, it was initially argued that the linguistic representations, including stress marking, may be underspecified in children with CAS. In the WEAVER model, the metrical shape of lexical items are accessed during morpho-phonological encoding; it was at this level that Shriberg and colleagues initially posited the deficit.

It is clear from the literature (and from clinical observations) that children with CAS often have difficulties on a range of phonological awareness tasks. However, many studies fail to compare the CAS children to children with other speech and language impairments, limiting the specificity of the claim that an impoverished phonological representation system is the core deficit in CAS. In fact, a large body of research has demonstrated that children with speech and language impairments also show data suggestive of phonological processing deficits, for example, poorly specified phonological representations (e.g., Leitão & Fletcher, 2004; Sutherland & Gillon, 2007). Moreover, a core linguistic deficit may not account for the range of difficulties seen in children with CAS, and, particularly, the developmental trajectory of the disorder.

Motoric and Linguistic, or Motoric Deficits only?

Despite the early prominence of the speech motor control deficit account of CAS, it appeared to be limited in its ability to explain the various clinical features associated with the disorder. For instance, a growing body of research highlighted that children with CAS almost always demonstrate difficulties with language and literacy (Bahr, Velleman, & Ziegler, 1999; Dewey, 1995; Ekelman & Aram, 1983; Hall, 2003c; Lewis, Freebairn, Hansen, Iyengar et al., 2004; Stackhouse, 1992), fuelling the debate as to whether this reflects co-morbidity, or is in fact part of the symptomatology of CAS itself.

Most recently, the traditional motor versus linguistic debate surrounding CAS has been advanced by calls to frame the debate as being about motor *and* linguistic, versus motor-*only,* deficits in CAS (American Speech-Language-Hearing Association, 2007). As will be seen in the next section, an extension to this approach may be the need to distinguish between *original* core deficits and deficits observed *after* a period of development (Maassen, 2002; Stackhouse, 1992).

The Need for a Development Perspective

Despite attempts to locate the 'underlying' deficit or deficits in CAS, multiple potential loci of impairment have been identified. In addition to wide-ranging patterns of symptoms, the nature of these difficulties is often observed to change over time, across task demands, and varies considerably between children (Hodge & Hancock, 1994; Shriberg et al., 1997a). Indeed, variability has been described as a 'constant' in CAS (Maassen, 2002). This variation within and between groups of children with CAS has been variously interpreted as suggesting co-morbidity (Hall, 2003c), confounded methodology (Guyette & Diedrich, 1981), the existence of sub-types (Crary, 1993), or that CAS is a sub-type of another disorder or disorders (Dodd, 1995). This inconsistency and lack of validated differentially diagnostic behavioural markers is somewhat difficult to interpret within standard applications of models, without implying additional explanations.

In contrast to acquired apraxia of speech, where the pattern of difficulty occurs because of neurological insult after (presumably) previously having normal speech and language skills, CAS emerges within a developing system (Hodge, 1994; Maassen, 2002; Stackhouse, 1992). Components of the speech and language system can not be assumed to be already in place in their entirety, especially at the onset of the disorder's characteristics. Despite the apparent sense of this statement, much previous research has, either explicitly or implicitly, attempted to interpret the pattern of impairment in the context of models of established systems.

A case in point is the long-running debate, described above, concerning whether the underlying deficit in CAS is of a 'motoric' or 'linguistic' nature (Crary & Towne, 1984). As outlined in the previous section, researchers have found evidence supporting both linguistic and speech motor impairments in children with CAS. These disparate explanations of CAS suggest multiple levels of deficit. Rather than necessarily implying co-morbidity, however, an alternative and logical approach is to view the disorder from a developmental perspective (Maassen, 2002; Stackhouse, 1992).

Not specific to CAS, a number of researchers have highlighted the problematic nature of applying a cognitive neuropsychological approach to developmental phenomena (Bishop, 1997; Karmiloff-Smith, 1998; Karmiloff-Smith, 1999; Karmiloff-Smith, Scerif, & Ansari, 2003; Karmiloff-Smith, Scerif, & Thomas, 2002). Bishop (1997) emphasised the inappropriateness of using static models (with associated assumptions of modularity) in developmental contexts. For many developmental disorders, it is rare to find a highly selective impairment. Rather, the observation of a range of patterns of performance is, in Bishop's words, "inevitable, given the interdependence of different stages of processing upon one another in the course of development" (p. 904). Karmiloff-Smith and colleagues have also highlighted this issue, describing the developmental process as having a significant effect on the resultant phenotype at various stages of development (Karmiloff-Smith, 1998; Karmiloff-Smith, 1999; Karmiloff-Smith, Scerif, & Ansari, 2003; Karmiloff-Smith, Scerif, & Thomas, 2002; Paterson, Brown, Gsodl, Johnson, & Karmiloff-Smith, 1999). The pattern of deficits observed in a child at one particular time-point, therefore, does not necessarily equate to modules or processing components with an original causal role.

Such a developmental approach is consistent with dynamic systems theory, where developmental outcomes depend on the cooperative interactions between many systems (McCune, 1992). Development within the organism (in this case the infant) is a function of the interactions of many subsystems, including the central nervous system and the environment (Piek, 2006). Change occurs when instability in attractor states potentiates a shift to another state, and individual variation is explained by dynamic interaction within the system.

Developmental Perspectives of Communication Development

In the area of communication development, researchers have begun to acknowledge the dynamic and interactive nature of developing systems, and the difficulties disentangling language and speech motor control processes. This is in line with a shift in thinking from differentiating phonological and speech motor control processes, to a "deliberate blurring of the boundaries" between the representation of speech sounds, and the motor functions used to produce them (Kent, 2000, p. 391).

Indeed, within speech and language systems, there is growing evidence for the interaction between levels of representation in early development. A direct and dynamic relationship between speech motor/ phonetic skill and 'language' development (including the development of phonology) has been demonstrated (Mitchell, 1995). Smith and Goffman (Goffman & Smith, 1999; Smith & Goffman, 2003, 2004), for example, have presented considerable evidence that speech motor skill contributes to the emergence of linguistic units. In infancy, the production of 'vocal motor schemes' (consistent phonetic patterning for a particular consonant) has been shown to be related to lexical acquisition (McCune & Vihman, 2001). Furthermore, continuities observed in individual profiles in babbling through to first words provide additional support for the importance of phonetic development as providing the foundation for phonological development and vocabulary acquisition (Stoel-Gammon, 1989, 1992), and thus also the interactive nature of development. The application of this knowledge to studies of developmental disorders such as CAS, however, has been limited.

A Developmental Perspective of CAS

Rather than necessarily indicating co-morbidity, the broad range of symptoms observed in children with CAS can be accommodated by acknowledging the interactive nature of development in a dynamic system (Karmiloff-Smith, 1998). In such a developmental and interactive context, we might expect to find evidence of diverse symptoms in those with the disorder. As the developing system is dynamic and interactive, an impairment at one level of the emerging system has the potential to influence subsequent development of other areas. Consideration of the available evidence in CAS supports this notion. As outlined in the previous section, children with CAS present with varied profiles, and the nature of specific symptoms varies with age and within and across individuals. Deficits at multiple levels have been suggested, including linguistic and speech motor levels. The disorder, however, has rarely been investigated with specific reference to a developmental model of speech and language. The utilisation of such a model which emphasises and describes the processes involved in normal development may provide a more acceptable explanation of the core deficit and the nature of changes over time.

Developmental Models of Speech Production

There are a number of models that explicitly conceptualise the gradual development of speech and language processes in the infant and developing child. These models, although varying in terminology and specificity, all attempt to capture the unfolding system, and propose an initially simplified system in the developing child. Most models emphasise either speech motor development or language development; few combine the two. The following models, however, describe the developing system in its emerging and dynamic state.

Levelt et al. (Levelt, Roelofs, & Meyer, 1999)

An adapted version of Levelt's (1989) model of speech production is one such developmental model. Maassen (2002) and Zeigler and Maassen (2004), based on suggestions by Levelt et al. (1999), proposed a simplified version of Levelt's model as a useful framework for interpreting information about early speech and language development. As shown in Figure 2, in contrast to the adult system (Figure 1), in the early stages of development the infant is proposed to have a somewhat simplified system, comprising of a conceptual system and a set of syllabic articulatory gestures. These two systems are initially independent.

The conceptual system includes abstract conceptual knowledge, such as object permanence, as well as emerging lexical concepts, which are initially auditory in nature (i.e., the beginning of a receptive vocabulary). In addition to the conceptual system, the infant also has an emerging speech motor system, initially comprising of a restricted set of syllabic articulatory gestures. The infant, on producing these syllables, attends to the acoustic output and gradually builds a core repository of speech motor patterns – forming the *protosyllabary*.

In this model, during the first stage of intentional speech, there is a direct connection between the conceptual system and the protosyllabary. That is, real word production evolves from a coupling of the speech motor patterns and the lexical concepts. The infant's first words often comprise previously babbled syllables (Locke, 2004) and support exists for similarities in consonants produced in babbling and first words (e.g., Kent, Mitchell, & Sancier, 1991; McCune & Vihman, 2001). It is only under

the pressure of a growing vocabulary that the word form lexicon and phonological encoding systems develop. In contrast to the adult system, the infant system has not yet established the areas that are often implicated in CAS.

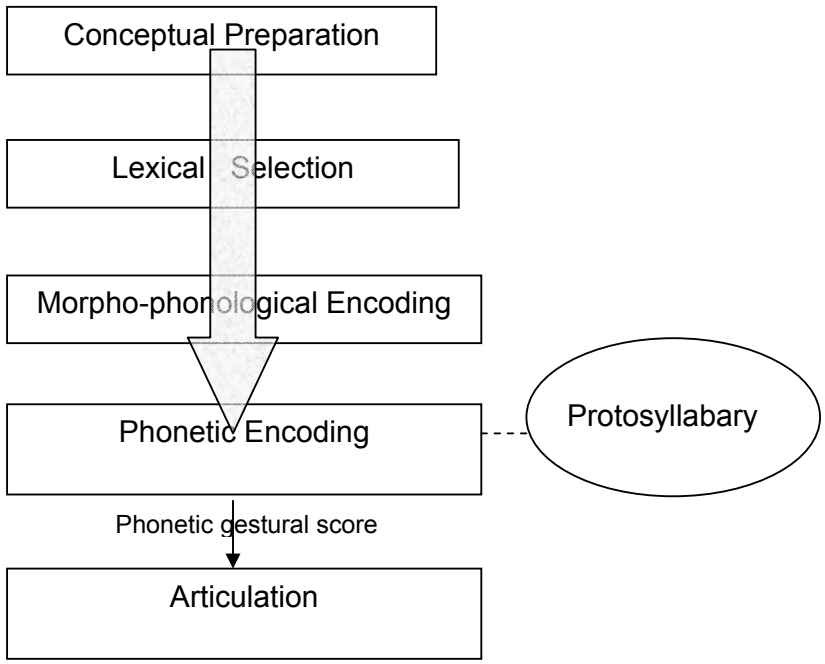


Figure 2. The simplified speech system proposed to be present in infancy (Maassen, 2002; Zeigler and Maassen, 2004).

DIVA (Guenther, 1995; Guenther, Ghosh, & Tourville, 2006)

The notion of the setting up of the protosyllabary finds parallels with neural network models of speech development, such as the DIVA model (Directions into Velocities of Articulators, Guenther, 1995; Guenther et al., 2006). In this model, the processes involved in learning to speak are simulated, and cortical correlates for each process suggested. Critically, a babbling phase like that in infancy sets up the available sequences for later production. During this phase, semi-random movements of the articulators produce auditory and somatosensory feedback to the model. This

information (articulatory, auditory and somatosensory), when combined, forms the basis for learning and tuning the mappings between sensory and motor functions.

Using the input of speech presented to it (analogous to the infant being exposed to language by its parent/s), the model then learns the auditory targets for words and syllables. The resultant 'speech sound map cells' can generate the motor commands to produce the syllable. "After babbling, the model can quickly learn to produce new sounds from audio samples provided to it, and it can produce arbitrary combinations of the sounds it has learned" (Guenther et al., 2006, p. 282).

Westermann and Miranda's (2004) computational model

The developing system is also highlighted in Westermann and Miranda's (2004) recently proposed computational model of sensorimotor coupling in speech development. In this model, articulatory parameters and auditory perception set up subsequent motor and perceptual representations. During a babbling phase, auditory and articulatory parameters are coupled in an experience-dependent way. That is, when the model generates motor parameters it *babbles* and *listens* to the resulting output, developing connections between two parameters. An important prediction of this model is that the absence of normal babbling will result in abnormal production and perception patterns later on.

Stackhouse and Wells (1997)

Finally, Stackhouse and Wells' model (1997) also provides a developmental perspective of speech and language acquisition. The infant system begins with fewer input and output processes and, through experience and development, gradually expands. Initially 'input' and 'output' systems are separate, only to be coupled on the infant's production of first words. Furthermore, phonological representations are not established until the infant has developed motor programming and motor planning systems, active during babbling.

A commonality among the models described above is the emphasis on the developmental nature of speech and language acquisition. Such a developmental perspective has important implications for understanding disorders such as CAS, although few researchers have explicitly considered this issue when interpreting the

disorder (cf. Maassen, 2002; Stackhouse, 1992; Strand, 2002). The models also suggest an important role for prelinguistic vocalisations (particularly babbling), in setting up the speech motor patterns for subsequent meaningful speech production. This important aspect of development is the focus of the section below.

Prelinguistic Vocal Development

Despite individual variation, the vocalisations of typically developing infants show a general progression, from reflexive vocal noises to those that are increasingly speech-like in manner of production and resultant sound (Locke, 2002, 2004; Nathani, Ertmer, & Stark, 2006; Oller, 2000). Physiological, cognitive, perceptual, motoric and social-emotional developments are thought to underlie such progressions (Locke, 2002, 2004). Although various systems for detailing the changes that occur have been proposed, a general developmental sequence has been identified (Nathani, Ertmer, & Stark, 2006).

Vocalisations in the first three months from birth include cries and reflexive sounds such as burps, coughs and hiccups that lack the acoustic property of full resonance (Oller, 2000). Typical vocalisations during this time include faint, low-pitched grunt-like sounds with muffled resonance, termed quasi-resonant nuclei (Nathani et al., 2006). By three months, babies are able to control phonation to produce raspberries and vowel-like sounds with full resonance (Nathani et al., 2006; Oller, 2000).

Vowels and vowel glides are observed to emerge between 3 and 8 months of age (Nathani et al., 2006). Between 6 and 10 months of age an important milestone in infant vocal development is reached – canonical babbling (Oller, 2000). The infant produces canonical syllables – ‘adult like’ syllables containing a closant (consonant like sound) and vocant (vowel like sound). Such syllables are readily and reliably identified by parents (Oller, Eilers, & Basinger, 2001) and often are produced in reduplicated strings (Mitchell & Kent, 1990). The emergence of canonical babbling in normal development is robust to factors such as socio-economic status and ambient language (Oller et al., 2001; Oller, Eilers, Neal, & Schwartz, 1999). Importantly, it is a behaviour with similarities to other rhythmical movements (Thelen, 1981), and tends to co-occur with object banging (Ejiri & Masataka, 2001).

The emergence of canonical syllables is thought to represent one of the earliest ventures into speech motor control (Moore & Ruark, 1996). According to the Frame-Content theory proposed by Davis and MacNeilage (Davis & MacNeilage, 1995; MacNeilage, 1998; MacNeilage & Davis, 1990; MacNeilage, Davis, Kinney, & Matyear, 2000; MacNeilage & Davis, 2000, 2001), babbling productions represent rhythmical oscillations of the jaw, and form the basis of later articulations. Rather than being productions of individual consonant and vowel like sounds, each syllable is the result of the combined mandibular oscillation and vocalisation. In the context of the adapted Levelt model (Levelt, 1989), these initial productions are those that are used to set up the protosyllabary.

There are suggestions that the canonical syllables produced by the (initially independent) speech motor system play a role in subsequent sensory-motor development, and even neuronal growth. Levelt's adapted model (Levelt et al., 1999) emphasises the perceptuo-motor (or sensory-motor) development that occurs when the infant both produces 'syllabic articulatory gestures' (i.e., babbles) and hears the auditory consequences of such output. Similarly, in the DIVA model (Guenther, 2006), a feedback and feedforward loop exists between the input and output mappings. Moreover, theories encompassing the evolutionary and biological bases of emerging speech production suggest a dynamic relationship between neurological maturation and experience, and a sensitive period for such sensorimotor integration (Locke, 2004; Locke & Pearson, 1992). Observations of increased dendritic branching in the vocal-motor and manual areas of the left hemisphere at 5-6 months of age may suggest that babbling is both *enabled by* and *facilitates* such brain growth. Indeed, Locke and Pearson (1992) suggested that "…babbling may stimulate some additional brain growth of the type that is needed for vocal learning." (p. 113).

Following the emergence of canonical babbling, typically developing infants gradually produce more phonotactically varied vocalisations prior to, and overlapping with, the emergence of first words (Nathani et al., 2006). Vocalisations typical at this stage (between 9 and 12 months) include diphthongs, syllables with more complex phonotactic patterns such as vowel-consonant (VC), VCV, and CCV, and jargon strings.

Proposed Core Deficit in CAS

The important advances in vocal development that occur in the typically developing system, and the prelinguistic period in general, may be the key to investigating the nature of the core deficit underlying CAS. A number of researchers have suggested a relatively 'low level' impairment in speech motor control as being responsible for the deficits observed in children with CAS (Maassen, 2002; Strand, 2002). In contrast to the purported limitations of such accounts in explaining concomitant language difficulties, a developmental perspective suggests otherwise. Maassen (2002), for example, proposed a relatively 'low level' impairment for CAS with flow on effects to the establishment of higher level linguistic processes. Specifically, it was proposed that CAS is an impairment in "perceptuomotor control and perceptuomotor learning" (p. 263). This account proposes a core speech motor (or articulatory motor) control deficit in CAS, affecting the development of auditory-perceptual links, and the forming of corresponding representations. Such a deficit is not of the peripheral nature of a dysarthria, where low muscle tone, for example, causes an inability to move the articulators adequately (as is the case in some types of cerebral palsy, for example).

In this hypothesis, the infant with CAS[2] does not have the typical syllabic articulatory gestures available, which restricts the development of the protosyllabary. While the typically developing infant's system continues to develop, using previously established syllables to produce meaningful speech, this natural progression is impaired by articulatory motor difficulties in the infant with CAS. In terms of the DIVA model (Guenther, 1995), the infant has a reduced capacity to form systematic mappings between articulatory movements and auditory consequences. Establishing phoneme-specific mappings and representations is thus also impaired. In typical development, the usual rapid vocabulary growth that occurs between 18 and 30 months overtaxes the protosyllabary, leading to the establishment of systems for phonological and morphological encoding (Levelt et al., 1999; Stackhouse & Wells, 1997). Articulatory gestures for each word can no longer be stored economically as holistic units, necessitating their dismantling into smaller units.

[2] The term 'infant with CAS' is used in the conceptual sense, and does not imply the ability to diagnose CAS prelinguistically

Thus, if the protosyllabary does not contain a rich repository of speech motor patterns, negative flow on effects for subsequent linguistic development would result.

This account of CAS, by taking a developmental perspective, is able to accommodate the varied pattern of deficits that is noted in children with features of the disorder. Whilst historically, a 'lower-level' account of CAS has been viewed as being inadequate in accounting for the broad range of observed features in CAS, interpretation of the deficit in the context of the complex and dynamic interaction of speech and language processes *predicts* that additional deficits will also be observed, *especially* after a period of development (Maassen, 2002). If speech motor / phonetic skill facilitates the development of phonology and an expressive vocabulary (Maassen, 2002), a restricted phonemic inventory and limited vocabulary expansion would be *expected* in children with CAS. Limited vocabulary acquisition would delay or restrict the development of the word form lexicon and associated phonological encoding system. The lack of babbling (resulting from the original speech motor impairment) would have effects not only on production, but also perception (Westermann & Miranda, 2004).

The potential impact of a low level articulatory motor deficit on subsequent speech and language development is highlighted by cross-discipline research supporting a sensitive period for sensorimotor integration during vocal motor learning (Haesler et al., 2007; Pytte & Suthers, 2000). Studies of vocal learning in birdsong development show that interruptions during the imitative motor learning phase negatively affect vocal learning (Pytte & Suthers, 2000). Interestingly, and highly relevant to CAS given the literature relating to the FOXP2 gene, interference to the normal FOXP2 levels in zebra finches (who share neural parallels to humans in terms of vocalisation development) impairs vocal imitation and subsequent vocalisations (Haesler et al., 2007). Not only do affected birds show an impaired ability to imitate tutor's songs, evident very early on and persisting into adulthood, but syllable production is abnormally variable, consistent with observations of inconsistency in CAS. Thus, in children with CAS, a core impairment in speech motor control may affect both the development of the protosyllabary, as well as normal sensorimotor integration and vocal *learning*.

Proposed Anomalies in Early Development in CAS

The notion of a core deficit in speech motor control in CAS, proposed by many investigators but rarely interpreted within a developmental model of speech production, predicts atypical vocalisation development in the prelinguistic period. Given that babbling has been identified as one of the earliest behaviours of speech motor control (Kent, 2000), if impaired speech motor control is the core deficit in CAS, the impairment would be expected to be evident pre-linguistically.

Furthermore, due to the initial independence of the speech motor and conceptual systems, such a deficit would theoretically be present in the context of an intact conceptual system (Maassen, 2002). Due to the dynamic nature of development, the 'best' time to observe such a deficit would be very early in development, before the speech motor control system interacts completely with higher language levels.

Atypical Prelinguistic Vocal Development in CAS

Numerous researchers have suggested atypical prelinguistic vocal development in children with CAS, yet limited empirical accounts exist. Davis and Velleman (2000) discussed frequently reported characteristics of CAS and proposed behavioural correlates of these features for infants and toddlers. The proposed features relate to phonetic, phonological, language, motor and general characteristics. In parallel with observations in preschool and school age children with CAS, a restricted phonetic repertoire and lack of variety in consonants and vowels was proposed. Limited vocal output and a lack of babbling and consonant-vowel combinations were also suggested. Although yet to be thoroughly investigated in infants and toddlers, many of these features find support from the research literature and theoretical models of language development.

Hall (2003a) described the clinical observation that many parents report children with CAS to have been quiet babies who did not coo or babble as expected. Similarly, Maassen (2002) suggested delayed or absent babbling histories in children with CAS. Description by Tate (1991, as cited in Shriberg, Aram & Kwiatkowski, 1997b) of three infants later considered to have CAS included the observation that they were 'quiet' babies with limited vocalisations.

Velleman (1994) reported case studies of two preschool children with CAS, both with reported histories of delayed or decreased babbling. One was reported to babble only from 12 months of age, with word production not emerging until 16 months; the other, reported to be milder in CAS symptoms, was reported to babble 'infrequently' around 7 to 12 months, with first words at 12 months but subsequent delays in expressive language development. Information about other areas of infant development was not provided.

Atypical Prelinguistic Vocal Development in Late Talkers

Atypical prelinguistic vocal development may not be specific to CAS, however (Oller, 2000). Reports of late talkers suggest atypical development in this broader group of children with communication difficulties (Stoel-Gammon, 1989). Longitudinal observation of two infants who at 2 years of age presented with restricted phonological and lexical development suggested a relationship between prespeech vocalisations and later language ability (Stoel-Gammon, 1989). One of these late talkers infrequently produced canonical babble until 24 months of age; the other produced only one type of consonant in his babbles (velar stops). In a larger study focussing on 2 year olds with expressive language delay, the proportion of consonant to vowel babble was the strongest predictor of language outcome 5 months later (Whitehurst, Smith, Fischel, Arnold, & Lonigan, 1991).

Research on larger samples of children also suggests continuity in communication development from infancy to the second year of life, in typical and disordered acquisition alike (Reilly et al., 2007). In a longitudinal investigation of over 1700 children, communication development at 12 months of age was the strongest predictor of language ability at 24 months (Reilly et al., 2007). Consonant inventory at 18 to 22 months has also been shown to be related to expressive language (Watt, Wetherby, & Shumway, 2006). In a study of over 3400 infants, Oller and colleagues (Oller et al., 1999) reported persistently smaller expressive vocabularies from 18 to 30 months in infants who were not producing canonical babbling by 10 months of age.

While atypical prelinguistic vocalisation development may also feature in some cases of more general language delay, the source of the impairment may differ to that proposed for CAS. Various theoretical accounts of language delay and SLI exist, proposing deficits in perceptual (Leonard, McGregor, & Allen, 1992), linguistic (Rice & Wexler, 1996) or more general processing domains (Millar, Kail, & Leonard, 2001), or in the ability to integrate such domains (Evans, 2001). In the context of Levelt's simplified model of early language development, a core deficit in the conceptualiser and/or linguistic processes is often proposed. Although some children with general language delays may also have histories of atypical prelinguistic vocal development, the nature and pattern of their early profiles may be different. There may be a general delay in the communication system, for example, with no dissociation between conceptual and speech motor domains. Even for children with phonological disorders (the features of which often overlap with those for CAS), the source of difficulty is often hypothesised to be linguistic and/or cognitive-linguistic in nature (Dodd & McIntosh, 2008). If CAS has a core motoric origin, the profile may be different to that for other disorders, despite overlap in symptomatology.

In summary, a core deficit in speech motor control, interpreted in a developmental context, presents as a plausible theoretical account of CAS. It accommodates the presence of speech motor and linguistic impairments observed in children with the disorder, and also predicts specific evidence of the core deficit in the prelinguistic period. Evidence supporting such a deficit is lacking, however.

Rationale for the Present Research

CAS has been identified as an important speech disorder with significant consequences for affected children and their families. Previous research has been limited by the lack of a validated set of differentially diagnostic features and large variability in the presentation of children thought to have the disorder. Relatively few researchers have conceptualised the disorder with explicit reference to developmental models. However, when the interactive and dynamic nature of early communication development is considered, a relatively low level speech motor control deficit may accommodate the range of features observed.

The hypothesis of a core lower level impairment in CAS, with flow on effects to the establishment of higher level linguistic systems predicts that the core deficit may be evident very early on in development (Maassen, 2002). Indeed, it is often assumed that the neurological impairment presumed to underlie CAS is present from birth (American Speech-Language-Hearing Association, 2007; Maassen, 2002). However, more specific to this hypothesis is the prediction of abnormal prelinguistic vocal development, prior to the production of first words. Levelt's adapted model (Levelt et al., 1999) proposes the initial independence of the emerging speech motor control and conceptual systems at this stage of development. In the case of CAS, it is therefore theoretically plausible for an infant to show dissociation between the two areas, with impaired speech motor control but intact development of the conceptualiser. As development progresses and the systems are coupled with the production of first words, effects on the developing lexical system would be observed, and later linguistic aspects would be negatively affected.

Irrespective of whether such an isolated deficit accounts for every clinical case of CAS, the existence of a dissociated pattern has yet to be reported prelinguistically. Differences in early vocal development have not been thoroughly demonstrated, and evidence of a pattern of selective impairment coupled with intact abilities in other domains (in the prelinguistic period) is lacking.

A number of factors contribute to this lack of evidence. There is still little consensus on the differentially diagnostic features of CAS, and so called 'pure' cases are rare. Even though investigations of CAS focus on childhood, an enormous amount of development has already taken place by the age usually studied. Most studies of CAS focus on children over the age of four (American Speech-Language-Hearing Association, 2007). According to developmental models of speech and language processing, the interaction that takes place would mean that untangling the original loci of underlying deficits would be impractical. Furthermore, we are unable to diagnose CAS in infants and toddlers (Davis & Velleman, 2000). There are no published longitudinal studies of the developmental progression of CAS from pre-speech (Zeigler & Maassen, 2004). Large scale longitudinal studies investigating the

emergence and risk factors of speech and language impairment have not reported specifically on CAS (Reilly et al., 2007).

Aims and Research Questions

The present research aimed to address the lack of research on CAS which explicitly considers a developmental perspective. The main objective was to examine a theoretical account of CAS in the context of a developmental model of speech production. Specifically, the research aimed to test a speech motor control deficit account of CAS. When interpreted from a developmental perspective, this notion posits articulo-motor deficits in the context of intact conceptual development in infancy.

Based on this premise, the following broad research questions were explored:

1. Do children with CAS show deficits in early vocalisation development consistent with a speech motor control account of the disorder?
2. Do infants at risk of CAS show a profile consistent with evidence of a dissociation between conceptual and speech motor control abilities in early development?

Reflecting the exploratory nature of the research, and consistent with the current state of the literature in CAS, it was acknowledged that the present research would provide preliminary information regarding the broad research questions.

Methodological Overview and Rationale

Whilst studies focussing on readily-identifiable disorders (such as cleft palate or Down's syndrome) can identify and track affected individuals from infancy, research investigating later-diagnosed disorders such as CAS must employ alternative methods to document early features and developmental trajectories. The use of retrospective parent report, analysis of early home videos, and longitudinal investigation of at-risk samples are examples of approaches applied to the study of other complex developmental disorders, most notably in the study of Autism

Spectrum Disorders (Bryson et al., 2007; Coonrod & Stone, 2004; Dawson, Osterling, Meltzoff, & Kuhl, 2000; Iverson & Wozniak, 2007; Landa & Garrett-Mayer, 2006; Matson, Wilkins, & Gonzalez, 2007; Sivberg, 2003; Wetherby et al., 2004).When used in isolation, inferences drawn from the results of such methods are limited. However, when used in combination or when the results are built upon and corroborated, features worthy of further empirical investigation can be identified.

In this vein, the present research utilises a combination of methodologies: retrospective parent report, analysis of retrospective infant data available from a separate community based program, and longitudinal investigation of an at-risk sample. Study 1 was designed as a preliminary investigation of the first broad aim, using parent report information relating to the prelinguistic period in children with sCAS. Although clinical anecdotes give some indication of report tendencies, empirical research quantifying parental recollections of vocal development in children with sCAS has been lacking. In this study, parents of children with a clinical diagnosis of sCAS completed a questionnaire reporting on early vocalisation behaviours and developmental milestones. In order to investigate whether vocal development is reported similarly for children with a related developmental disorder that may have a different origin, responses were compared to those from parents of children with Specific Language Impairment (SLI), as well as a group of children with typically developing speech and language skills. Based on theoretical and clinical predictions, it was hypothesised that parents of children with a clinical diagnosis of CAS would report reduced or absent babbling, reduced vocalisations, and delayed language milestones in infancy, compared to typically developing infants. Reflecting its limited scope, Study 1 is presented near to its published form, with minor editing to avoid repetition within the thesis (Chapter 2).

Study 2 investigated the core deficit in CAS via analysis of retrospective infant data available for a unique clinical sample of children with varying speech and language profiles, including those with CAS features. Although researchers in the ASD field have used retrospective data to investigate early profiles of affected children (Watson, Baranek, & DiLavore, 2003), this approach has yet to be applied to the study of CAS. To address the diagnostic challenges relating to participant identification in CAS research, the sample was first characterised with respect to

operationally defined CAS features. Infant data for these same children, including measures of communication, motor, and cognitive development, were investigated using single case methodology, described further below (Crawford & Garthwaite, 2002; Crawford, Garthwaite, & Gray, 2003; Crawford, Garthwaite, Howell, & Gray, 2004; Crawford & Howell, 1998; Crawford, Howell, & Garthwaite, 1998). It was hypothesised that children with a high degree of CAS features at 3-4 years of age would show correlates of a speech motor control deficit in data from 9 months of age.

In study 3, single case methodology (as well as group comparisons) was used to further investigate the core deficit in CAS. In this study, detailed perceptual and acoustic data of prelinguistic vocalisations, and data from communication and developmental measures, were examined in a longitudinal investigation of infant siblings (of children with sCAS). "To date, no longitudinal studies are available in which children with DAS are followed from babbling to early speech" (Zeigler & Maassen, 2004, p. 436). Researchers studying CAS have yet to employ paradigms used in investigations of other complex developmental disorders. Study 3 contributes to this research need, and investigates the second broad research aim of examining whether a profile of dissociation between speech motor and conceptual abilities in early development is present in children who may be at increased 'risk' of CAS. It was hypothesised that, if any of the siblings showed a profile suggestive being at heightened risk of CAS, an isolated speech motor control deficit would be observed in the context of intact conceptual development.

While familial aggregation and genetic studies do not conclusively point to the heritability of CAS as a certainty, they do support the approach of utilising family history as a method for identifying infants who, by way of genetics, are at increased risk of the disorder. An affection rate of 9%, considerably larger than that estimated for the general population, has been suggested for siblings with CAS (Lewis, Freebairn, Hansen, Taylor et al., 2004). The large majority of infant siblings of children with CAS will not have CAS themselves (e.g., 20 of the 22 children in Lewis et al.'s research did not have features consistent with CAS, although most evidenced a range of speech and language difficulties). However, the use of family history in combination with observation of proposed early CAS-related features

(Davis & Velleman, 2000) presents a practical method for identifying infants for longitudinal investigation.

Similar approaches for identifying 'at-risk' infants have been used in the study of other complex developmental disorders. Studies of ASD (Iverson & Wozniak, 2007; Landa & Garrett-Mayer, 2006) and dyslexia (Koster et al., 2005; Lyytinen et al., 2001), for example, have reported the development of infant siblings of children with the disorders under investigation. In such studies, overall group performances are described in addressing the possibility of a broader phenotype. In most cases, the infant siblings' data are informative despite the inability to confirm a diagnosis for any individual until many years later.

Despite the suggestion of a genetic component in CAS, and the use of family history paradigms in other developmental disorders, few researchers have reported longitudinal investigations of CAS. Davis, Jacks and Marquardt (2005) reported a longitudinal study of vowel development in three children with CAS from 4;6 to 7;7 years of age, allowing for documentation of the nature of their impairment over that age range. Lewis and colleagues (Lewis, Freebairn, Hansen, Iyengar et al., 2004) reported longitudinal data for 10 children with CAS that they followed from 4-6 years of age for a period of four years, describing the changing phenotype in school aged children. These studies, whilst being longitudinal in nature, focussed on children who have already been diagnosed with CAS, and therefore do not report on the potentially informative prelinguistic to early speech period.

A core deficit in speech motor control in CAS predicts atypical vocalisation development, a restricted phonetic repertoire, limited syllable shapes, and acoustic patterns consistent with impaired speech motor control. Thus, an affected infant may be expected to show delayed or absent babbling (Maassen, 2002), reduced frequency of canonical syllables, limited consonant and vowel inventory, and limited phonotactic variation, consistent with features proposed by Davis and Velleman (2000). Moreover, deficits in speech motor control are often reflected in acoustic analyses of speech production (Kent, 2000). If a core deficit in speech motor control underlies the symptoms observed in CAS, this may also be reflected in acoustic measures of the initial syllabic gestures produced by the infant. Acoustic measures of

duration, fundamental frequency, and analyses of vowel formants 1 and 2 may reveal subtle differences that reflect impaired speech motor control, and were therefore also investigated in Study 3.

Single Case Methodology

Despite the tradition of employing group comparisons in psychological and human communication sciences, researchers have highlighted the inadequacy of such an approach in instances where there is large variability between individuals within the groups, there are small numbers of participants, and where individual patterns are of particular importance (Bishop, 1997; Caramazza, 1986), as is often the case in CAS research. Due in part to the low prevalence of the disorder, most group studies have included relatively small numbers of participants with CAS, for example 5 or 6 children (American Speech-Language-Hearing Association, 2007). Inspection of individual patterns of performance within these groups often shows large variability on numerous measures. Many studies of CAS have therefore focussed on the performance of individuals (e.g., Davis et al., 2005). Following procedures utilised in the neuropsychological literature (Crawford & Howell, 1998), single case methodology was used in the present thesis where individual cases were of interest.

An associated major limitation in investigating a low-incidence disorder such as CAS is the difficulty in applying standard statistical procedures to the data (Crawford & Garthwaite, 2002; Crawford, Garthwaite, & Gray, 2003; Crawford, Garthwaite, Howell, & Gray, 2004; Crawford & Howell, 1998; Crawford, Howell, & Garthwaite, 1998). Statistical techniques appropriate for comparing small numbers of disordered participants to a larger, but still modest, group of typically developing participants were employed in Studies 2 and 3. In testing whether an individual shows a statistically significant 'deficit' on a particular measure, the control sample statistics are treated as statistics rather than as population parameters as is the case when *z* scores are used (Crawford & Garthwaite, 2002; Crawford & Howell, 1998). The 'abnormality' or rarity of a participant's score is indicated by the obtained *p* value of the modified t-test. Investigations have demonstrated that this modified t-test procedure controls the Type I error rate regardless of the control sample size, and is robust even when used with highly skewed data (Crawford & Garthwaite, 2002; Crawford & Howell, 1998).

Furthermore, in order to test for a dissociation or differential deficit in an individual, the Revised Standardised Difference Test (RSDT) method (Crawford & Garthwaite, 2005) was applied. In this procedure, strict criteria are used to identify instances where an individual's score on one measure shows a deficit, but their score on another measure does not show a deficit *and* the difference between the two scores exceeds the difference scores in the comparison sample (i.e., a classical dissociation). The Type 1 error rate is again suitably controlled regardless of the size of the control sample and correlation between the two measures (Crawford & Garthwaite, 2005).

The nature of exploring a relatively under-studied area (namely, the prelinguistic period in CAS) involves a high number of statistical analyses being applied to the range of measures investigated within this thesis. Because hypotheses were theory-driven, it was decided *a*-priori to interpret results against the standard per-test alpha level of .05 rather than systematically apply a Bonferroni correction. Although this increases the risk of making a Type 1 error (Tabachnick & Fidell, 2001), the threat of ignoring potentially informative results relevant for guiding future larger scale research was considered imperative.

Summary

Each of the studies in the present thesis aims to examine CAS in a developmental context. Given the difficulties inherent in interpreting developmental disorders such as CAS in the context of established models of speech production, the theoretical importance of investigating children with CAS at earlier timepoints is significant. Information about the developmental picture of children with CAS or at risk of CAS would offer insight into the development of speech and language processes, not only in disordered systems, but in normal development.

CHAPTER 2

STUDY 1. RETROSPECTIVE PARENT REPORT OF EARLY VOCAL BEHAVIOURS IN CHILDREN WITH SUSPECTED CHILDHOOD APRAXIA OF SPEECH (sCAS) [1]

Introduction

This study aimed to apply a retrospective parent report paradigm to quantify the nature of hypothesised differences in early vocalisations and development for children with CAS. Retrospective parent report paradigms, despite having some potential methodological limitations related to recall ability and reliability, have been used in investigations of developmental disorders including autism and developmental delay. For example, both interview and questionnaire formats have been used with parents of children with autism to investigate the nature of reported early signs and concerns (Coonrod & Stone, 2004; Sivberg, 2003). We are not aware of any similar formal studies on children with CAS, although clinical anecdotes give some indication of parental report tendencies (Shriberg & Campbell, 2002). The questionnaire developed for the present study included items relating to vocalizations and babbling, as well as motor milestones and other 'features' often reported to co-occur with CAS (Davis & Velleman, 2000, Shriberg et al., 1997, Stackhouse, 1992). Despite the potential limitations of a retrospective parent report design, examining and quantifying parent report of early development may indicate a starting point in terms of identifying commonly reported early features of this challenging disorder.

[1] This study appears as an article in Child Language Teaching and Therapy (SAGE). See Appendix B for a statement of copyright permission and authorship. The abstract has been removed and the introduction edited to avoid repetition with Chapter 1

Method

Participants

Participants were parents (all mothers) of children with a clinical diagnosis of sCAS[2] (*n = 20)*, diagnosed SLI (*n = 20)*, and typically developing speech and language (TD, *n = 20)*. The children did not have any identified medical, physical or intellectual impairment. Hearing was normal for all children according to clinician and/or audiology report. Specific audiological reports were available for the sCAS children, which showed that all had normal hearing acuity and middle ear function at the time of their assessment.

The children with sCAS were identified from the caseload of a specialist second opinion clinic which caters for the state of Western Australia. A qualified Speech Pathologist with over 20 years experience in motor speech disorders provides assessment and treatment to children who are suspected (by their managing therapist) to have CAS. The 20 children (referred to the clinic over a period of 2 ½ years) identified for this study therefore represent the number of children identified by their managing therapist as having features consistent with CAS and *also* diagnosed with sCAS at the second opinion clinic using spontaneous speech samples, single word naming and elicitation of the same words in phrases/spontaneous speech, oral motor examination including DDK, stimulability testing in isolation and syllables, thorough case history taking, and formal and/or informal language assessment.

Because the children with sCAS were not directly assessed for this study, specific data are limited. Notwithstanding current debate as to the diagnostic criteria for sCAS (e.g. ASHA, 2006) case-note information was reviewed in terms of commonly reported characteristic features. The study children displayed: a limited consonant and vowel phonetic inventory, predominant use of simple syllable shapes, frequent omission errors, high incidence of vowel errors, altered suprasegmental characteristics, variability/lack of consistent patterns of output, increased errors on longer sequences, and groping/lack of willingness to imitate (Davis & Velleman, 2000). Table 1 lists summary clinical information for the children in the sCAS

[2] Following recommendations from the American Speech-Language-Hearing Association's Childhood Apraxia of Speech Draft Technical Report (ASHA, 2006) which recognized the lack of validated diagnostic criteria for CAS, the children with CAS are referred to as having *suspected* CAS (*s*CAS)

Table 1

Participant Characteristics for the sCAS Group

	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20
Chron Age (years; months)	5;0	3;1	4;0	4;5	4;3	4;0	3;4	4;11	3;6	3;0	4;11	3;6	3;0	5;0	3;8	3;6	4;3	4;2	3;9	4;1
Receptive Language	wnl	wnl	wnl	wnl	wnl	wnl	wnl	wnl	wnl	wnl	wnl	wnl	wnl	wnl	wnl	wnl	mild	mod	wnl	wnl
Expressive Language	mild	mild	sev	mod	mild	sev	mod	sev	mild	sev	wnl	sev	mod	mod	mild	sev	sev	sev	mild	sev
Intelligibility Rating	sev	sev	sev	sev	mod	mild	sev	sev	mild	sev	sev	mod	mod	sev	sev	sev	sev	sev	mod	sev
Features of CAS																				
Limited consonant inventory	+	+	-	+	-	-	+	+	-	+	+	-	+	+	+	+	+	+	+	+
Limited vowel inventory	+	+	+	+	+	-	+	+	-	+	-	+	+	+	+	+	+	+	-	-
High incidence of vowel errors	+	+	-	+	-	-	-	+	-	+	-	+	-	+	+	+	+	+	-	-
Diphthong errors	+	+	+	+	+	-	+	+	-	+	+	+	+	+	+	+	+	+	+	+
Simple syllable shapes	+	+	+	+	+	-	-	+	+	+	+	+	+	+	+	+	+	+	+	+
Frequent omission errors	+	+	-	+	+	-	+	+	+	+	+	-	+	-	+	+	+	+	+	+
Increased difficulty as complexity ↑	+	+	-	+	+	+	+	+	+	-	+	+	-	-	+	+	+	+	+	+
Groping	+	+	+	+	+	+	+	+	+	-	+	-	-	+	+	+	+	+	+	+
Token to token variability	+	+	+	+	-	+	+	+	+	+	+	+	+	-	+	+	+	+	+	+
Variability	+	+	-	+	+	+	+	+	+	+	+	+	+	-	+	+	+	+	-	-
Non-speech oral difficulties	+	+	+	+	-	-	+	+	+	-	-	+	+	+	+	+	+	-	-	+
Automatic vs volitional advantage	+	+	+	+	+	-	-	+	+	+	+	+	+	+	+	+	-	+	+	+
Altered prosody	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+	+
Slow initial response to therapy	+	+	-	+	+	+	+	+	+	+	+	-	-	+	+	+	+	+	+	+

Note: wnl = within normal limits, sev = severely impaired, mod = moderately impaired. Characteristics were derived from clinical records.

group. Consistent with evidence of considerable variability in the clinical presentation of children with sCAS (ASHA, 2006), Table 1 shows a wide range in the characteristics of this clinically identified sample. The number of 'features' present for each child ranges from 6 to 14 (mean, M = 11.4). The most common features are altered prosody, diphthong errors, simple syllable shapes, and token-to-token variability. The children also vary in intelligibility and language skills, although most show a receptive to expressive gap in language.

Children with SLI were identified through placement at a Language Development Centre, which services children with a primary specific language impairment. Placement requires normal nonverbal/performance and adaptive behaviour skills in the presence of significant language difficulties, assessed on standardised and informal assessments. Mean receptive language and expressive language standard scores were 66.3 (SD = 12.9) and 67.1 (SD = 11.9), respectively (Clinical Evaluation of Language Fundamentals – Preschool, Wiig et al., 1992). Note that the majority of children showed moderate to severe impairments in both receptive and expressive language. Children displaying concomitant phonological difficulties were not included in the present study, to limit overlapping speech features with the sCAS children.

The TD children were identified through primary schools who had taken part in speech pathology screenings. The study children had passed the speech and language screenings, comprising standardised and informal assessments, and did not display any academic, cognitive or motor difficulties.

All children were from monolingual English speaking homes. Chronological age and gender for the three groups are displayed in Table 2. Children in the sCAS group were younger than the SLI group, $t(38) = 5.6$, $p < .001$ and the TD group $t(38) = 4.5$, $p < .001$, reflecting differences in the convenience sampling applied in order to identify the children whose parents could be approached for this study. The implications of this average age difference of 12 months will be considered in the discussion. The TD and SLI groups were not significantly different in age, $t(38) = .25$, $p = .80$. There was no significant difference in the proportion of males/females in each group, x^2 (2, N = 60) = 3.73, $p = .155$.

Table 2

Mean Chronological Age (standard deviation) and Sex for the sCAS, SLI and TD Children

	sCAS	SLI	TD
Male	18	14	13
Female	2	6	7
Chronological Age (months)	48 (7.6)	60 (6.9)	61 (11.3)

Materials and Procedure

The questionnaire, designed for this study, (see Appendix A) asked about early development, including the presence/absence and age of onset of babble, how vocal the child was as an infant, language milestones, and associated developmental areas. These items were broadly consistent with areas cited in the literature as relevant to features of CAS (Davis & Velleman, 2000). Parents were encouraged to use any methods they could to help complete the questionnaire (e.g., talking to relatives, their child's infant health record book).

Data Analysis

Quantitative data from the questionnaire are reported. Where an age range was a possible response, the first month reported was taken as the reported age of emergence. Responses to items requiring a numerical value such as age or rating along an equal interval scale were treated as continuous data, and analysed via one way analysis of variance (ANOVA), with group (sCAS, SLI, TD) as the independent variable. Inspection of the data indicated that assumptions underlying the ANOVA were met, apart from one item (age smiled), which did not meet homogeneity of variance assumption. However, because ANOVA is robust to mild to moderate violations especially when groups are equal (Everitt, 1996), interpretation using ANOVA proceeded. Focussed comparisons were tested using Tukey's least significant difference (LSD) contrasts. Categorical data were analysed using chi-

square test of independence when assumptions were met or otherwise by the Fisher exact test (FET). Analyses were interpreted against an alpha level of .05.

Results

A summary of responses relating to the presence of behaviours and age of emergence are provided in Tables 3 and 4, respectively. The data reported below are organised according to items/behaviours, meaning that both frequency and age of emergence data for each group are reported together.

Vocalizations and Babbling

As shown in Table 3, more parents of the children with sCAS (55%) than both the SLI (25%) and TD (0%) group parents reported that their child had *not* made many sounds as an infant. The difference between the sCAS and TD samples was statistically significant, $x^2(1, N = 40) = 15.172$, $p < .001$. The difference between the sCAS and SLI groups was close to being statistically significant, $x^2 (1, N = 40) = 3.75$, $p = .053$.

In reporting the recalled volubility of their child as an infant on a scale of 1 to 5 (with 1 being "vocalized rarely" and 5 being "vocalized often"), there was a statistically significant difference between the groups, $F(2, 57) = 26.33$, $p < .001$. The sCAS group parents rated their child as having vocalized significantly less ($M = 2.3$, $SD = 1.1$) than the TD group ($M = 4.3$, $SD = 0.7$), $p < .001$, but not the SLI group ($M = 2.58$, $SD = 0.9$), $p = .402$.

The sCAS group differed significantly to the TD but not the SLI group in report of the presence of vowel noises in infancy, FET $p = .02$ and $p = .45$, respectively. Vowel noises were reported to be present for 100% of the TD children, compared to 70% in the sCAS group. The SLI and TD groups did not differ significantly on this item, FET $p = .231$. Mean reported age of emergence of these vocalizations was significantly different for the three groups, $F(2,43) = 6.024$, $p = .005$, with both clinical groups reporting later emergence (sCAS $M = 8.2$ months, SLI $M = 8.2$ months) than the TD group ($M = 4.9$ months), $p < .001$ for the difference between both clinical groups and the TD group.

Table 3

Frequency of Responses for Presence of Key Behaviours for the sCAS, SLI and TD Groups

	sCAS *n = 20*	SLI *n = 20*	TD *n = 20*
Make many sounds?			
Yes	9 (45%)	15 (75%)	20 (100%)
No	11 (55%)	5 (25%)	0
Make vowel noises?			
Yes	14 (70%)	17 (85%)	20 (100%)
No	3 (15%)	2 (10%)	0
Unsure	3 (15%)	1 (5%)	0
Babble (reduplicated)?			
Yes	12 (60%)	20 (100%)	20 (100%)
No	7 (35%)	0	0
Unsure	1 (5%)	0	0
Babble (variegated)?			
Yes	0	7 (35%)	13 (65%)
No	19 (95%)	11 (55%)	2 (10%)
Unsure	1 (5%)	2 (10%)	5 (25%)
Babble as much as other children			
More	0	0	8 (40%)
Same	2 (10%)	5 (25%)[a]	11 (55%)
Less	18 (90%)	14 (70%)[a]	1 (5%)
Feeding problems			
Yes	9 (45%)	9 (45*%)*	3 (15%)
No	11 (55%)	11 (55%)	17 (85%)
Dribbling issues			
Yes	9 (45%)	4 (20%)	2 (10%)
No	11 (55%)	16 (80%)	18 (90%)

[a] The percent for this item does not total 100 due to missing data for one respondent

There was a statistically significant difference between the sCAS group and the other groups in parental report of reduplicated babbling, FET $p = .003$ for both the SLI and TD groups. All children in the TD and SLI groups were reported to have babbled (reduplicated babble) in infancy, in contrast to 60% of the sCAS group. For those infants reported to have babbled (reduplicated), age of emergence was significantly later in both clinical groups (sCAS $M = 11.0$ months, SLI $M = 10.1$ months, TD $M = 7.2$ months), $F(2,46) = 10.141$, $p < .001$. Posthoc comparisons confirmed the difference lay between the sCAS and TD group, $p < .001$, and the SLI and TD group, $p = .001$.

When asked about how much their child had babbled as a baby, in comparison to other babies of the same age, the sCAS group parents were significantly more likely to report their child to have babbled *less* than other babies, in comparison to the TD group, (90% versus 5%), FET $p < .001$. However, the SLI group was also significantly more likely than the TD group to report that their child had babbled less (70% versus 5%), $p = .001$. There was no significant difference between the sCAS and SLI groups on this item, $p = .182$.

The frequency of parental report of the presence of variegated babbling was significantly different across the three groups, x^2 (2, $N= 60$) = 19.05, $p < .001$. None of the sCAS children were recalled as having produced variegated babble as an infant, while 65% of the TD group and 35% of the SLI group parents reported recalling the presence of variegated babbling. Post hoc comparisons using Fisher's Exact Test confirmed a significant difference between the sCAS and SLI groups, $p = .008$. There was no significant difference between the SLI and TD groups, $p = .113$. For those children who were recalled as having produced variegated babble, however, age of emergence was reported to be significantly later for the SLI group ($M = 12.2$ months), compared to the TD group ($M = 9.2$ months), $t(15) = 2.34$, $p = .033$.

Table 4

Mean (and standard deviations) for Age of Emergence (months) reported for the sCAS, SLI and TD Groups

	sCAS	SLI	TD
Vowel noises	8.2 (3.4)	8.2 (3.4)	4.9 (2.4)
Reduplicated babble	11.0 (2)	10.1 (3.1)	7.2 (1.9)
Variegated babble	-	12.2 (3.5)	9.2 (1.8)
Sat upright (unsupported)	7.4 (2.6)	7.2 (1.7)	5.6 (1.8)
Smiled (weeks)	8.6 (3.6)	16.8 (22.5)	6.9 (3.6)
Crawling	9.1 (2.4)	9.1 (1.9)	7.5 (1.6)
First steps (unaided)	13.6 (2.9)	12.5 (3.2)	11.7 (1.7)
First word	14.0 (6.7)	13.0 (4.7)	9.2 (2.5)
Two word combinations	33.3 (7.1)	27.0 (10.4)	14.6 (4.1)

Language and Other Developmental Milestones

Group data for language and other developmental milestones are displayed in Table 4. There was a significant difference in reported mean age of emergence for first words between the groups, $F(2,51) = 4.64$, $p = .014$. Reported age for the sCAS group was significantly later (M =14 months) than the TD group (M = 9 months), p = .005, but not the SLI group (M = 13 months), p = .555. The three groups again differed significantly on reported age of emergence of two word combinations, $F(2,47) = 22.23$, $p < .001$. This milestone was reported to emerge significantly later in the sCAS group (M = 33.3 months) when compared to both the TD group (M = 14 months), $p < .001$ and the SLI group (M = 27.0), p = .024.

There were no overall group differences in reported age of smiling, $F(2, 50) = 2.77$, $p = .072$, and of first steps, $F(2,57) = 2.44$, $p = .096$. Reported age of sitting upright and crawling showed significant group differences, $F(2, 54) = 4.85$, $p = .012$, and $F(2, 54) = 4.01$, $p = .024$, respectively. The sCAS group was reported as significantly later than the TD group for sitting, $p = .007$, and for crawling, $p = .017$. However, there were no significant differences between the sCAS group and the SLI group, $p = .790$ and $p = 1.000$, for sitting and crawling, respectively.

Feeding and Dribbling

Parent responses to questions about feeding and dribbling are also summarised in Table 3. No overall significant difference was found between the three groups for the rate of reported feeding issues, $x^2(2, N = 60) = 5.27$, $p = .072$. However, the same rate of feeding problems was reported in the CAS and SLI groups (both 45%), compared to only 15% of the TD group, and contrasts suggested the two clinical groups were significantly more likely to report feeding issues compared to the TD group, FET $p = .041$.

There was an overall significant difference between the three groups on the presence of dribbling issues, $x^2(2, N = 60) = 6.93$, $p = .031$. Post hoc comparisons revealed that the sCAS group (45%) was significantly more likely than the TD group (10%) to report such issues, $x^2(2, N = 40) = 6.144$, $p = .013$, whereas the SLI group (20%) was not significantly different to the TD group, FET $p = .331$. The two clinical groups were not significantly different on this item, $x^2(2, N = 40) = .784$, $p = .376$.

Correlations Between Variables

For the sCAS group, reported age of crawling and walking were significantly and positively correlated with that of two word combinations, $r = .49$, $p = .044$, and $r = .56$, $p = .021$, respectively. Further, the reported age of sitting upright was significantly correlated with that of first words, $r = .51$, $p = .033$. Reported age of crawling and walking were themselves correlated, $r = .82$, $p < .001$, and reported age of sitting upright was correlated with both age of crawling, $r = .75$, $p < .001$, and walking, $r = .58$, $p = .012$.

For the SLI group, reported age of first words and two word combinations were significantly correlated, $r = .61$, $p = .022$. Significant correlations were observed for reported age of sitting upright and crawling, $r = .49$, $p = .004$, sitting upright and walking, $r = .48$, $p = .039$, and crawling and walking, $r = .66$, $p = .002$. However, in contrast to the sCAS group, motor milestones were not significantly correlated with language milestones, $r = -.20$, $p = .460$ (crawling and two word combinations), $r = .28$, $p = .275$ (walking and two word combinations), and $r = .32$, $p = .222$ (sitting upright and first words).

Discussion

Study 1 sought to quantify parental report of early vocalizations in children with suspected childhood apraxia of speech (sCAS). The literature suggests a lack of consonant-vowel babble, or reduced amount and/or range of vocalizations may be an early feature reported in CAS (Maassen, 2002, Davis & Velleman, 2000). However, anecdotal reports have not previously been quantified, and differences in vocalizations and babbling may not be specific to CAS (Oller et al., 1999). Questionnaire responses on a range of early vocalization and developmental behaviours were compared for parents of children with a clinical diagnosis of sCAS, specific language impairment (SLI) and typically developing (TD) speech and language.

Differences in Vocalizations and Emerging Language

As expected, when compared to the TD group, the sCAS group parents were significantly more likely to report that their child had *not* made many sounds as a baby (55% versus 0%). Although, descriptively, more sCAS group parents reported that their child did not make many sounds as a baby compared to SLI group parents (25%), this difference was not confirmed statistically. This is in contrast to the observation that all the TD group parents reported that their child *had* made many sounds as a baby. On a scale of frequency of vocalisation, both the sCAS and SLI children were rated as having been 'quieter' infants. In contrast, the TD group were rated as having been significantly more vocal.

A striking consistency for the SLI and TD groups was that *all* parents reported that their child had babbled as an infant. In comparison, 35% of the sCAS group parents reported that their child had definitely *not* babbled. This difference was significant. Inspection of the response patterns revealed that those sCAS children who were reported not to have babbled were also those where the parent reported more negative responses overall for the other vocalization and babbling questions. The sCAS group parents recalled age of emergence of reduplicated babble to be significantly later than the TD group parents. Similarly, although all of the children with SLI were reported as having babbled as an infant, the mean age of emergence was significantly later (at nearly 11 months) than the mean of 7 months for the TD group. Oller, Eilers and Basinger (2001) found that parents reliably identify canonical babbling at the time of its occurrence. Although we cannot confirm the reliability of the parental responses because we do not have data on what the children *actually* did in infancy, these results suggest a pattern requiring further investigation.

In a study of over 3400 infants, Oller and colleagues (Oller et al., 1999) reported that infants with delayed canonical babbling had smaller expressive vocabularies at 18, 24, and 30 months. They suggested that the difficulty may originate in limited phonological production capabilities. Recall that the children with SLI in this study did not have concomitant phonological disorder. Given the assumed reliability of parent recall, the finding that all parents in the SLI and TD groups reported their child to have babbled as an infant may indicate that all of these children progressed through a canonical babbling stage, setting up the articulatory patterns used for later word production (Davis & MacNeilage, 1995). Some of the children with sCAS, reported not to have babbled at all in infancy, may have 'missed' this opportunity due to limited speech motor capabilities, and therefore been disadvantaged in terms of establishing a set of patterns to couple with lexical concepts for first word production (Maassen, 2002).

The present study also included items about 'variegated' babbling. A number of examples were provided; with the focus on the child having produced a non-meaningful vocalization where the consonant sound changed. Sixty five percent of the TD group reported recalling this type of babbling, in contrast to none of the sCAS parents. The SLI group parents were significantly more likely than the sCAS

group to report the presence of variegated babbling, with the SLI and TD groups not differing statistically. Again the SLI group were reported as having later emerging variegated babbling than the TD group.

First words were reported to emerge later in both the sCAS and SLI groups, consistent with developmental expectations and reported features of the two clinical groups (Oller et al., 1999, Davis & Velleman, 2000). The reported age of emergence of two-word combinations showed a widening gap, with TD children reported to reach the milestone on average at 15 months, followed by children with SLI (average 27 months), then children with sCAS (33 months). Given that children with sCAS are often reported to be resistant to traditional therapy approaches, and acquiring a substantial expressive vocabulary appears limited by speech output difficulties in the children (Maassen, 2002), it is not surprising that reported age of two-word combinations is one item that sets the sCAS group apart.

These results provide preliminary support for the notion of differences in the pre-linguistic vocalizations of children with sCAS (Maassen, 2002). The parent responses suggest that at least a portion of the children with sCAS were limited in their core repository of speech motor patterns during early development that could be drawn upon for later meaningful speech production. Developmental theories and models of speech production emphasize the importance of this early vocal experience and predict future production *and* perception problems in the absence of normal babbling (e.g., Westermann & Miranda, 2004). Furthermore, the results indicating the sCAS group to be significantly later in the emergence of two-word combinations may reflect the importance of early phonetic and phonological development for subsequent vocabulary acquisition (McCune & Vihman, 2001).

Differences in Motor Skills

Children with sCAS are often reported to have difficulties with a range of fine and gross motor skills (Davis et al., 1998, Davis & Velleman, 2000). This study included a limited number of questions relating to motor skills. The sCAS group was reported as having reached some gross motor milestones significantly later than both the SLI and TD children (sitting upright and crawling). Interestingly, reported age of both crawling and walking were significantly correlated with that of the emergence of

two-word combinations, and reported age of sitting upright was significantly correlated with that of first words. No such correlation with any of the language items and motor milestones was observed for the SLI group. This may support a core motor constraint in sCAS.

Other anecdotally reported issues in some children with sCAS include the presence of feeding and dribbling difficulties (Davis & Velleman, 2000). These usually relate to issues with food textures and/or coordination. Although the results of the present study relied on the parents' own interpretation of what may constitute an 'issue', they suggest that parents of both clinical groups report similar rates of feeding issues, significantly more than the TD parents. The results for the reported rates of dribbling issues are more difficult to interpret. The sCAS group reported more dribbling issues than the TD group. However, the sCAS group did not differ significantly from the SLI group on this item, and the SLI group did not differ significantly from the TD group.

Limitations and Conclusions

This study was not diagnostic in nature (cf. Shriberg et al., 2003). Until a set of pathobehavioural and/or genetic markers are identified for CAS, we cannot be certain that our group is representative of the larger CAS population. A framework based on commonly reported features of sCAS found in the literature was used retrospectively to describe these children. The children with sCAS displayed a number of features typical of CAS (see Table 1), and they represent cases identified by both their managing clinician and a Speech Pathologist experienced in complex differential diagnoses. Observation over time (after an extended period of diagnostic therapy) also confirmed the appropriateness of the sCAS clinical diagnosis.

Children with SLI were included to investigate whether differences in parent report of early vocalization could be associated with more general language difficulties. However, this study did not use a comparison group of children with phonological disorder, who are often reported to share many of the characteristics observed in CAS (McCabe et al., 1998). Therefore, it is not known to what extent the differences we observed between the sCAS and SLI groups reflect something specific to sCAS, or early vocalization behaviours in speech disordered children in general. It would

be of interest in future research to compare the parental report of early vocalizations in children with phonological disorder, including various subgroups (e.g. Dodd, 1995).

There are limitations associated with relying on retrospective recall. However, previous research has used questionnaires or parent interview to gain insight into parents' recollections of early development of other developmental disorders (e.g., Sivberg, 2003). Asking parents to recall detailed information about vocalizations, and using written examples to attempt to capture the intricacies of these vocalizations, was an ambitious exercise. In particular, given the lack of research confirming the reliability of parents' ability to identify variegated babbling at the time of occurrence, retrospective recall of this feature may be less reliable. The observation that many of the parents used aids to assist their recall, and the fact that they were able to state 'unsure' and 'can't recall' adds to the face validity of the results. In general, parents tended not to use these options when they were available, suggesting that the parents' recollections were reliable to some extent. Given the significant difference between groups in age of the children at the time of parent report, the sCAS and SLI groups were compared in terms of the number of 'unsure' responses. The groups were not different in this respect, t (4) = 0.71, p = .519.

Overall, the results of this preliminary study support the notion of differences in parental report of the early vocalizations of children with sCAS, when compared to TD children. However, on many items children with SLI were reported similarly to children with sCAS. The most striking differences between the two clinical groups related to parental report of the presence of babbling and the widening gap in expressive language ability reflected in the significantly later age of emergence of two-word combinations in the sCAS children. The reported behaviours recalled by parents in this study suggest some areas of difference that indicate the need for prospective, longitudinal observation of pre-linguistic vocalizations and speech-motor control in various 'at risk' groups of children (Oller, 1999).

Clinical Implications

Clinicians often inquire about early developmental milestones and behaviours, using a combination of written case history forms and face to face interview. Given the theoretical and practical significance of the prelinguistic stage of development, it is important that clinicians gather information about early vocal development. The present study used retrospective methodology as one method of collecting data. However, if the opportunity arises to collect this information prospectively (for example, with younger siblings of children already identified with sCAS), parent report offers a simple alternative to more in-depth protocols designed to directly assess vocal development (Nathani et al., 2006; Oller et al., 2001). While information on infant vocalization cannot be used diagnostically at present, the results suggest measures of pre-linguistic vocalization have the potential to increase our understanding of their role in normal and disordered speech development.

CHAPTER 3

STUDY 2. INVESTIGATING RETROSPECTIVE INFANT SPEECH BEHAVIOURS FOR CHILDREN WITH CAS FEATURES AT 3-4 YEARS

Overview

Study 1 examined prelinguistic vocal development in children with sCAS through comparison of parental report of such behaviours in children with the diagnosis, to children with SLI and those with typical speech and language development. Consistent with theoretical predictions based on the literature and clinical anecdotes, the sCAS children were reported to be significantly less vocal as infants, less likely to babble, later in the emergence of first words, and later in the emergence of two-word combinations compared to children with normal speech and language skills. However, children with SLI were reported similarly to the sCAS group on many items relating to prelinguistic development. Despite the areas of overlap, the sCAS group were reported to be significantly different to both comparison groups on items relating to babbling, age of emergence of two-word combinations, and age of some motor milestones. In addition, a significant correlation between motor and language milestones was observed for the sCAS but not the SLI group. The results suggested anomalies in pre-linguistic vocal development in children with sCAS and supported the need for further research into the developmental trajectory of speech and language development in this population.

Study 2 builds on the preliminary results from Study 1 in investigating prelinguistic vocal development in CAS. Reported in two phases, CAS features in a clinical sample of children were investigated, allowing their communication profiles to be explored and described in detail. The children had previously taken part in a screening program in infancy, and had gone on to require further speech pathology services in subsequent years. In the second phase, retrospective infant data available

for these same children were compared to that for a large group of children without identified communication impairments. This unique set of data allowed key hypotheses relating to early development in CAS to be explored further.

Study 2A: Profiling CAS features in a clinical sample

Introduction

As outlined in Chapter 1, a number of candidate features of CAS have been described in the literature. Few, however, have been operationally defined and described in such a way as to allow clear identification for research and clinical purposes. Researchers have acknowledged the need for detailed participant description in CAS studies, especially while a validated set of diagnostic criteria are lacking (American Speech-Language-Hearing Association, 2007). The main objective of Study 2 was to further investigate hypothesised anomalies in pre-linguistic vocal development, via analysis of infant data available for a unique group of children with CAS features. However, in keeping with recommendations for more detailed participant description, a first step was to operationally define and measure CAS features in a clinically-ascertained group of children.

The following features, introduced in Chapter 1, are commonly reported as being key characteristics of CAS and were explored for the purpose of detailed participant description in the present study:

Inconsistency. Inconsistent speech errors and/or variability in production are commonly reported features of CAS. Most clinical accounts and research studies cite inconsistency among their diagnostic inclusion criteria (Davis, Jakielski, & Marquardt, 1998; Groenen, Maassen, Crul, & Thoonen, 1996; McCabe, Rosenthal, & McLeod, 1998; Nijland et al., 2002). In contrast to many other features associated with CAS, this characteristic is usually assumed to be specific to children with apraxia. Children with phonologically-based speech sound disorders are typically reported to make consistent error patterns (cf. Dodd, 1995), often across whole classes of speech sounds. The feature of inconsistency is also generally accepted as

not being typical of the speech of dysarthric children (Shriberg, 2003). In contrast, the high unintelligibility often associated with CAS has been hypothesised to relate, in part, to the variability and thus unpredictability of speech errors made by children with the disorder (Maassen, 2002).

Despite the frequency with which inconsistency is reported as a unique feature of CAS, specific measures and criteria for establishing the presence of this characteristic are rarely specified in detail. Studies typically report the presence of inconsistent error patterns (e.g., Nijland et al. 2002) without further specifying the degree of inconsistency or method by which it is calculated. Groenen et al. (1996) used clinicians' judgements to establish the presence of inconsistency. Betz and Stoel-Gammon (2005) explored various methods for quantifying error consistency in children with speech disorders. Three alternative measures applied to the same set of target words highlighted the potential for variation in reporting such features. Despite the ambiguity in typical reports of this feature, Dodd and colleagues provide guidance for evaluating inconsistency in children with a range of speech disorders, including normative data (Dodd, 1995; Dodd, Hua, Crosbie, Holm, & Ozanne, 2002). In this method, used also in the present study, token to token variability is measured via production of the same set of words three separate times, controlling the potential confounding factors of phonetic context and length.

Prosodic anomalies. As introduced in Chapter 1, altered suprasegmental aspects of speech production is another feature frequently reported in investigations of CAS (American Speech-Language-Hearing Association, 2007). Children with the disorder are often reported to sound 'robotic' or have 'staccato' speech, with terms such as *monostress*, *monoloud* and *monopitch* used in clinical and research descriptions (Shriberg, Aram, & Kwiatkowski, 1997b; Shriberg, Aram, & Kwiatkowski, 1997c). The presence of lexical stress difficulties, particularly the presence of excessive-equal stress (where all or most syllables in a word or utterance receive prominent stress), has been identified as a potential differentially diagnostic feature of CAS. Such difficulties were the only differentiating feature of a subgroup of children suspected to have CAS in Shriberg and colleagues' studies (Shriberg et al., 1997c). Perceptually, Odell and Shriberg (2001) demonstrated that children with CAS produced a high proportion of utterances that were deemed to have

inappropriate stress, in particular, excessive-equal stress. However, metrical analyses have indicated that the pattern of stress errors in CAS children is similar to that observed in younger, typically developing children, in that weak syllables are *either* omitted or over-stressed (Velleman & Shriberg, 1999).

Moreover, although stress deficits, identified perceptually, are frequently reported in CAS participants, investigations using acoustic analyses have often failed to find anomalies in more objective acoustic correlates (Munson, Bjorum, & Windsor, 2003; Skinder, Strand, & Mignerey, 1999). Skinder et al. (1999) reported acoustic correlates of stress to be appropriate in their group of children with CAS, despite the participants being judged (perceptually) as less accurately producing stress patterns. Similarly, Munson, Bjorum and Windsor (2003) found no significant deficits in acoustic measures such as vowel durations, fundamental frequencies, vowel intensities and f0 peak timing for their participants with CAS, despite the children being perceived as producing inappropriate stress patterns. In contrast, Nijland et al. (2003), Skinder et al. (Skinder, Connaghan, Strand, & Betz, 2000) and Shriberg et al. (Shriberg, Green, Campbell, McSweeny, & Scheer, 2003) found stress deficits reflected in acoustic analyses focusing on duration and peak f0.

Consequently, despite the near consensus view of prosodic deficits being associated with CAS, the method of measuring such deficits is still being established. Although frequently reported, findings of prosodic disturbances have not been universal and are not consistently reflected in acoustic measures. Research has yet to investigate the role of factors such as age of participants and the nature and amount of therapy received as contributing factors in prosodic observations. Despite these limitations, syllable loss and lexical stress errors were reported on in the present description of study participants, following their prominence in studies investigating prosodic disturbances in CAS (Odell & Shriberg, 2001; Velleman & Shriberg, 1999).

High incidence of vowel errors. Difficulties with vowels is another commonly reported characteristic of CAS. In normal development, the acquisition of vowels usually occurs early and in a relatively short space of time (Ball & Gibbon, 2002; Selby, Robb, & Gilbert, 2000). In contrast, vowel-related deficits (either high incidence of vowel errors or restricted vowel phonemic inventory) are among one of

the most consistently reported characteristics of CAS (Davis, Jacks, & Marquardt, 2005; Davis & Velleman, 2000; Strand, 2003).

Despite the frequency with which vowel issues are reported as characteristic features of CAS, few researchers have specified their methods for identifying them. For example, vowel errors are one of 11 features included in a list frequently used for identifying CAS participants, of which eight are required for a diagnosis (Davis et al., 1998). The nature and degree of vowel errors is not specified, and may not necessarily be present in all children with suspected CAS. In typical descriptions of CAS participants, vowel errors are reported on articulation tests or conversational speech (e.g., Marion, Sussman, & Marquardt, 1993) but further detail (such as how many vowels are affected or percent vowels correct) is not provided. A method for quantifying the presence of vowel errors, including comparison with age-referenced normative data, is provided in the DEAP (Dodd et al., 2002), yet few researchers have utilised this assessment tool in CAS studies to date. The present research uses this tool to identify the presence of vowel errors.

Speech sequencing difficulties. An inability to easily sequence speech gestures is a commonly reported CAS feature, and one that reflects the often implied core deficit in speech motor programming and/or planning (American Speech-Language-Hearing Association, 2007). As well as being frequently reported, difficulties sequencing syllables is a characteristic that often persists in children with CAS, even when other aspects of speech production have improved (Lewis, Freebairn, Hansen, Iyengar, & Taylor, 2004). Researchers have described specific difficulty in tasks such as imitating a series of syllables (Marion et al., 1993) or difficulties sequencing phonemes and syllables, evident in productions of words and nonwords (e.g., Lewis et al., 2004; Nijland et al., 2002). Often, broad descriptions are provided, such as ‘difficulty in speech sequencing’ (Marquardt, Jacks, & Davis, 2004). In most accounts of this feature, children with CAS may have difficulty co-ordinating and producing sequences of syllables, especially where alterations in place of articulation are required. Infrequently, researchers have quantified this feature by way of performance on formal assessments of sequencing (Shriberg, Campbell et al., 2003), such as the Verbal Motor Production Assessment for Children (VMPAC, Hayden & Square, 1999). Most, however, have noted it as a participant feature without providing further detail.

Performance on diadochokinetic (DDK) tasks is often reported to be impaired for children with CAS (American Speech-Language-Hearing Association, 2007), providing additional evidence of difficulty in the production of sequences of syllables. Difficulties are especially evident for productions of alternating syllable sequences (Thoonen, Maassen, Wit, Gabreels, & Schreuder, 1996). Ekleman and Aram (1983), for example, reported their CAS participants to have 'marked inability/difficulty' repeating 'pataka'. Thoonen and colleagues (Thoonen, Maassen, Gabreels, & Schreuder, 1999; Thoonen et al., 1996) demonstrated difficulties on syllable repetition tasks for CAS children, with syllable repetition rates for single syllables differentiating them from children with dysarthria (the children with dysarthria produced slower productions), and rates for alternating syllables (e.g., pataka) differentiating them from children with typical development. There is much variation in the methods of presentation, scoring, and interpretation of DDK tasks. Williams and Stackhouse (2000), however, reported that in children aged 3 to 5 years, accuracy and consistency of production are more informative (than rate); the present study therefore focussed on these aspects of DDK.

As with other CAS-related features, syllable sequencing difficulties are still most commonly reported in clinical terms, without specificity or quantification of the nature of the difficulties. However, assessments which include syllable sequencing (e.g., the sequencing area of the VMPAC, diadochokinetic subtest of the DEAP) provide guidance on comparison to normal development. The present study used a combination of performance on the VMPAC as well as DDK performance (accuracy and consistency) to describe this feature in participants.

Increased errors as length and/or complexity increase. Almost certainly related to the speech sequencing difficulties described above, children with CAS are often reported to have increasing difficulties as length and complexity increase (American Speech-Language-Hearing Association, 2007). Davis and colleagues (Davis et al., 1998) included increased errors on longer units of speech output as one of their 11 features of CAS. The feature is thus often reported for participant selection in studies using the Davis et al. criteria (e.g., Skinder et al., 1999). "Increased errors on polysyllabic words" (Lewis et al., 2004, p. 124), and an "inability to produce complex phonemic sequences" (Nijland et al., 2002, p. 464) are

examples of descriptors associated with this CAS characteristic. However, details specifying how the feature is objectively identified are usually lacking. Research has yet to investigate the specific nature of the increasing difficulty. It is likely that length, phonetic complexity and phonotactic complexity all play a role. The present study used Roy and Chiat's (2004) preschool repetition task (because of its appropriateness for the age group studied and inclusion of varying numbers of syllables) to provide information on this feature for participant description.

Aim and Predictions

The purpose of Study 2A was to document and describe the communication profiles of the 'clinical sample', by operationally defining and quantifying key CAS features. As such, no specific hypotheses were developed. Because a number of measures were derived from non-standardised tasks, a group of age-matched typically developing children were tested on the same tasks in order to provide a normative reference of performance. The clinical sample children, some (but not all) of whom were identified by their managing clinicians as showing features consistent with a CAS diagnosis, were expected to demonstrate impaired performance on tasks reflecting CAS features. Given the observation of CAS-related features in children with a broad range of speech-sound disorders (McCabe et al., 1998), it was predicted that some of these features would also be present in many of the clinically-ascertained children. The number and severity of features was of interest in describing the clinical sample for later interpretation of infant profiles (Study 2B).

Method

Participants

Thirty children, 21 displaying typically developing speech and language skills and a clinical sample of nine children, aged 3 years 2 months to 4 years 9 months participated in Study 2A. Children met the following general inclusion criteria: no diagnosed or suspected intellectual impairment, pervasive developmental disorder, hearing impairment or significant medical conditions; normal nonverbal intelligence, and were monolingual speakers of English. Parents were provided with written and

verbal information about the study and gave written consent for their child to participate. Ethics clearance was obtained through the Curtin University of Technology and South Metropolitan Area Health Service Human Research Ethics Committees.

Clinical sample. Nine children (7 boys and 2 girls), ranging in age from 38 to 52 months, who had previously taken part in a community speech pathology screening program as infants (see Appendix C for a description of the program) and who were still in receipt of speech pathology services comprised the clinical sample. Speech pathology clinics in the Health Department of Western Australia and Language Development Centres (LDCs) in the Perth Metropolitan area were advised about the study via presentations at meetings, email requests, and telephone. Clinicians were requested to examine their caseloads for children who had previously been part of the program, who were now at least three years of age and were clients of the speech pathology service. Requests for participants were conducted over a period of 12 months.

Clinicians were aware that the study was particularly interested in children with CAS features, but that children with a range of speech and language issues were being recruited. Of the nine children recruited, three of these were identified by their managing clinician as having features consistent with CAS (participants 1, 2, and 3). Participant 2 had also undergone a second opinion assessment by a clinical specialist with significant experience in motor speech disorders. This assessment 'confirmed' the CAS diagnosis. Participants 4 to 9 were not identified by their referring clinicians as being suspected of having CAS. Participant 6, although previously taking part in the infant program, had only recently re-engaged with the speech pathology clinic and had not had a formal assessment by a speech pathologist. The speech and language skills of the clinical sample are described further in the results section in the context of profiling their communication skills.

Typically developing (TD) sample. The typically developing sample consisted of 10 boys and 11 girls with age-appropriate speech and language skills, recruited from two local mainstream kindergarten and 'pre-kindy' (i.e., 3 and 4 year-old) programs. Teachers were asked to distribute information packs to parents of children

who were developing appropriately for their age, and who did not have any developmental or medical issues. Language skills were screened using the linguistic concepts and recalling sentences in context subtests of the Clinical Evaluation of Language Fundamentals – Preschool (CELF-P, Wiig, Secord, & Semel, 1992). Phonological development was examined with the diagnostic screen of the Diagnostic Evaluation of Articulation and Phonology (DEAP, Dodd et al., 2002). The diagnostic screen is reported to have strong sensitivity, identifying 100% of true negatives, confirming the appropriateness of using it to confirm typical phonological development (Dodd et al., 2002). For inclusion into the TD sample, children were required to score within the normal range on the CELF-P subtests (i.e., standard scores above 7) and to have passed the diagnostic screen of the DEAP.

The performance scale of the Wechsler Preschool and Primary Scale of Intelligence, 3rd Edition (WPPSI-3, Wechsler, 2002) was used to screen nonverbal intelligence in both samples. Performance IQ (PIQ) was calculated using the Block Design and Object Assembly subtests (for children under 4 years), or the Block Design, Matrix Reasoning and Picture Concepts subtests (for children 4;0 and over). Chronological age, gender and PIQ for the two samples are displayed in Table 5. Independent groups t-tests adjusted for unequal sample sizes confirmed that the two samples did not differ significantly on chronological age or nonverbal intelligence, $t(28) = 0.92$, $p = .63$, and $t(28) = 0.53$, $p = .61$, respectively.

Procedure and Assessment Battery

Each child was tested in a quiet room with minimal distractions. TD children were assessed on location at the kindergartens over two sessions, a week apart. The clinical sample participants were assessed at the child's home or a nearby clinic, depending on parental preference. These children usually required three to four testing sessions to complete all the tasks. A Sony lapel condenser microphone and Sony Minidisc recorder (MZ-N710) were used to record the children's speech in stereo wave format with 16 bit digitisation and a sampling rate of 44100 Hz. In addition to assessing the children's nonverbal intelligence, the following battery of standardised and experimental assessments was administered in order to characterise and describe the participants' communication skills:

Table 5

Chronological Age, Gender and Performance IQ for the TD (n=21) and Clinical Samples (n=9)

	Chronological Age (months)	Performance IQ	Gender
TD Sample			11 M, 10 F
M	48	113	
SD	5.9	8.6	
Range	37-57	100-128	
Clinical Sample			
1	52	109	F
2	48	100	M
3	49	129	M
4	50	90	M
5	45	82[a]	M
6	45	90	M
7	47	124	M
8	40	115	F
9	38	141	M
M	46	108	
SD	4.6	8.6	
Range	38-52	82-141	

[a] Although this falls slightly below the normal range, given the size of the standard error measurement and observation of normal functioning in the kindergarten environment, it was decided to include this participant's data

CELF-P. Receptive and expressive language skills were assessed with the CELF-P (Wiig et al., 1992), a commonly used clinical assessment tool with sound psychometric properties, including strong concurrent validity and acceptable internal consistency, test-retest and inter-rater reliability, particularly for the ages targeted in the present study (Impara & Plake, 1998). All six subtests (three receptive and three expressive) were administered to the clinical sample, providing estimates of receptive and expressive language ability (the screening subtests were administered to the typically developing sample to confirm eligibility). Receptive and expressive language scores, expressed as standard scores, have a normative mean of 100 and standard deviation of 15.

DEAP. Articulation and phonological development of the clinical sample were assessed with the articulation assessment, phonological assessment and inconsistency assessment subtests of the DEAP (Dodd et al., 2002). This assessment tool has also been shown to have sound psychometric qualities, including strong test-retest and inter-rater reliability, and high content and concurrent validity. Children were required to name 30 pictures in the articulation assessment, covering most English consonants and vowels. The child's stimulability for phonemes not accurately produced in the naming section was also tested in this assessment. The phonology assessment required the child to name 50 pictures, covering all English consonants, vowels and diphthongs, and allowing phonological processes to be identified. In the inconsistency assessment, children were required to name 25 pictures on three occasions, allowing observation of (in)consistency of production of the same lexical items. Percent consonants correct (PCC), percent vowels correct (PVC), percent phonemes correct (PPC), and an inconsistency score were derived from the DEAP assessments, and compared to the norms provided.

Verbal Motor Production Assessment for Children (VMPAC). The focal oromotor control and sequencing areas of the VMPAC (VMPAC, Hayden & Square, 1999) were used to evaluate the clinical sample's speech motor abilities. Children were required to produce various speech and non-speech postures, in isolation (oromotor control) and in sequence (sequencing). Test-retest reliability for the VMPAC is reported to range from 0.56 to 0.88. Inter-rater reliability is stronger, with correlations ranging from 0.93 to 0.99. A recent review of tests designed for use in the assessment of children with CAS identified the common lack of tools to

reliably evaluate speech motor ability (McCauley & Strand, 2008). The VMPAC, however, was identified as the only available tool that was based on sound theory and also included normative data.

Diadochokinesis (DDK). Oromotor development and sequencing ability was also examined via a diadochokinesis (DDK) task. Participants were asked to produce rapid repetitions of single syllables (e.g. /pʌ/), and repetitions of alternating syllables (/pʌtʌkʌ), following live demonstration by the researcher. For each, the children were given an example of the syllable and asked to repeat it. They were then given an example of a repetitive string and were required to produce a similar string. If the child did not respond, they were given up to 3 more attempts. Following Williams and Stackhouse (2000), accuracy and consistency of each syllable (i.e., the child's accuracy in producing the syllable and consistency in multiple repetitions), ability to produce an alternating tri-syllabic sequence and consistency of multiple repetitions of the sequence were scored.

Preschool Repetition Test. A prosodically controlled word and nonword repetition task appropriate for use with young children (Roy & Chiat, 2004) was used to further investigate the children's speech production abilities. Procedures as outlined in Roy and Chiat (2004) were adhered to, with random presentation of each set and counter-balancing of words and non-words (18 of each, matched and balanced for phonemes, length and prosodic structure). Specifically, each child was introduced to a puppet and told that they were going to "help the puppet say some words/silly words". Two practice trials were given prior to the presentation of the block of items. To aid participation and for randomisation, each child selected a card (containing the 'word/nonword') out of a box, repeated the word after the examiner, and was then allowed to 'feed' it to the puppet. Each item was presented live to aid participation in this young cohort. Frequent encouragement was provided in the form of verbal praise and/or tangible reinforcers as needed.

Each item was transcribed (broad phonetic transcription) from a digitised recording and scored for overall accuracy, percentage of phonemes correct (PPC), syllable loss and stress errors. In contrast to methods of scoring accuracy where allowances are made for phonological processes produced by individual children (S.Chiat, personal

communication, October, 2006), a more conservative approach was employed in the present study. Items were scored as incorrect if any part of the word was produced incorrectly. This was because, in contrast to applications for children with typically developing speech or those using consistent phonological processes (S. Chiat, personal communication, October, 2006), some of the clinical sample children in the present study presented with largely inconsistent speech, making it impossible to determine occasions where a 'process' was being used. Instances of equal and excessive stress or misplaced stress (stress errors), perceptually-judged by the primary investigator, were noted. A second judge re-coded 10% of the sample for syllable loss and stress errors. Inter-rater reliability was found to be strong, Cohen's kappa = 0.94, $p < .001$. Syllable loss and stress errors were combined to reflect prosodic 'errors'. Results for each item were summed to produce an overall accuracy score (percentage), PPC, total syllable loss errors and total stress errors.

Profiling of CAS features

Using data from the standardised and experimental tasks, CAS features were examined and quantified for the clinical sample, with the following measures:

Inconsistency Score. Inconsistent production of the same word on different occasions (i.e., token to token variability) was noted. The inconsistency assessment of the DEAP, where the child is required to name a set of 25 pictures, three times, indicates occurrences where the same word is produced differently on different trials. An inconsistency score of 40% or more is considered outside the normal range on the DEAP, and was similarly employed to indicate presence of this feature (i.e., inconsistency) in the present study.

Prosodic Errors. Syllable loss and lexical stress errors, both hypothesised to contribute to the percept of prosodic anomalies in conversational speech (Velleman & Shriberg, 1999), were coded from the preschool repetition test. Lexical stress errors included instances of either misplaced stress (e.g., BAlloon) or equal-excessive stress (e.g., BA-LOON). Total number of prosodic errors (syllable loss plus lexical stress errors) was tallied for each participant. The feature of prosodic anomalies was considered to be present where a participant showed significantly more total prosodic errors, compared to the controls.

Percentage of vowels correct (PVC). Percentage of vowels correct, calculated from the phonology assessment of the DEAP, was used as a measure of vowel errors. PVC standard scores on the DEAP (M = 10, SD = 3) falling more than one standard deviation below the mean indicated the presence of the feature of 'high incidence of vowel errors' (Dodd et al., 2002).

Sequencing score. The sequencing score on the VMPAC was used as an indicator of sequencing ability (Shriberg, Campbell et al., 2003). This score (a percentage) represented the child's performance on various speech sequencing tasks. Using the normative information provided in the manual (Hayden & Square, 1999), scores below the normal range for the child's age were taken to indicate sequencing difficulties.

Performance on DDK. DDK performance was used to supplement the VMPAC sequencing scores in describing speech sequencing ability. Williams and Stackhouse (2000) have found accuracy and consistency to be important measures in younger children. It was predicted that clinical sample children with CAS features would have difficulty with the alternating syllable task (i.e., show low accuracy) and show reduced consistency in productions. Those that showed this difficulty (inaccuracy and inconsistency on the alternating syllable task) would be considered as having sequencing difficulties.

Percentage Phonemes Correct (PCC) regression slopes. Regression slopes for PPC across syllable length on the preschool repetition test were investigated to capture the notion of increasing errors as syllable length increases. It was assumed that a greater negative slope would be observed for children with CAS. However, because accuracy in repetition tasks also reduces over syllable length increases for children with other speech and language disorders (Chiat & Roy, 2007), specificity might be poor. Some CAS children might also produce a high level of errors across all syllable lengths. They may have less scope to show a larger slope value, because in a sense, they are closer to the floor level in the task. As such, regression slope as a function of intercept value (reflecting a proportionate measure) may be able to distinguish these children from TD and non-CAS children. Thus, a significantly larger negative regression slope relative to the intercept value was taken to reflect the feature of increasing errors as length increases.

Results

Speech and Language Assessments

Receptive and Expressive Language scores (CELF-P), percent consonants correct (PCC), percent vowels correct (PVC), inconsistency scores (DEAP), and focal oral motor control and sequencing ratings (VMPAC) for the clinical sample are displayed in Table 6. Based on these standardised assessments and consistent with the heterogeneity observed in clinical samples (Broomfield & Dodd, 2004), communication profiles varied considerably. Five children displayed receptive language skills below the normal range (three showing severe deficits, one with moderate and one with mild difficulties), and six presented with expressive language difficulties (five with severe and one with mild impairment). On the DEAP, PPC was below the normal range for all but three participants (7, 8 and 9). Percentage of vowels correct varied from extremely low (38% for participant 2) to well within the expected range for age (99% and 100% respectively for participants 7 and 9). Four children showed speech sequencing deficits on the VMPAC (participants 1, 2, 3 and 6), with two of these (participants 2 and 3) also displaying oromotor control deficits on this tool.

Participants 7, 8 and 9, whilst having been engaged in speech pathology services over a number of years for language delays, essentially displayed language and phonological skills within the normal range for their age on assessment. Participants 1, 2, 3 and 6 showed expressive language difficulties, impaired phonological skills including vowel errors, high degrees of inconsistency, and speech sequencing deficits. Participant 4, whilst also presenting with language and phonological issues, did not show inconsistency or sequencing difficulties. Participant 5, who had severe receptive and expressive language difficulties and a poor PCC, did not evidence difficulties in consistency, vowel production or sequencing.

With respect to CAS-related features derived from the standardised assessment results described above, inconsistency scores were over 40% (and thus considered outside the normal range) for participants 1, 2, 3 and 6; a high incidence of vowel errors (PVC standard score <7) was observed for participants 1, 2, 3, 4 and 6; and sequencing difficulties on the VMPAC were observed in participants 1, 2, 3 and 6.

Table 6

Assessment Scores from the CELF-P, DEAP and VMPAC for the Clinical Sample

	CELF-P		DEAP [c]			VMPAC	
ID	Rec [a]	Exp [b]	PCC	PVC	Incon [d]	Oro [e]	Seq [f]
1	102	79	43 (3)	94 (3)	76	WNL	mild
2	79	69	9 (3)	38 (3)	76	sev	sev
3	50	50	30 (3)	66 (3)	52	sev	sev
4	50	50	62 (3)	92 (3)	10	WNL	WNL
5	50	50	62 (4)	96 (7)	32	WNL	WNL
6	77	50	46 (3)	89 (3)	64	WNL	sev
7	127	108	94 (13)	99 (10)	12	WNL	WNL
8	91	88	68 (8)	92 (7)	28	WNL	WNL
9	100	94	89 (12)	100 (14)	8	WNL	WNL

[a] Receptive Language Score [b] Expressive Language Score [c] Standard scores (mean of 10 and standard deviation of 3) are shown in parentheses [d] Inconsistency score (%), scores over 40% are considered inconsistent (Dodd et al., 2002) [e] Rating from Focal Oromotor Control subtest [f] Rating from Sequencing subtest

DDK

Accuracy and consistency for the single syllable trains and alternating tri-syllabic sequence are shown in Table 7. Results are pooled and shown as percentages for the TD sample to enable an interpretive backdrop for the clinical sample children's results. For example, the percentage of TD children who accurately produced /pʌ/ is displayed (100%). As shown in the table, all of the 21 TD children were accurate

and consistent in their productions of the single syllables. All but one of the TD children were accurate when producing /pʌtʌkʌ /. Consistency varied more, with most but not all of the TD children producing consistent repetitions of the tri-syllabic sequence. Of those who were not 100% consistent, they invariably produced two or three consistent productions, with another few either at the beginning or end of the train containing some transposition.

Table 7

Accuracy and Consistency on Single and Tri-syllabic Sequences for the TD and Clinical Samples

	Accuracy				Consistency			
	/pʌ/	/tʌ/	/kʌ/	/pʌtʌkʌ/	/pʌ/	/tʌ/	/kʌ/	/pʌtʌkʌ/
TD[a]	100%	100%	100%	95%	100%	100%	100%	67%
Clinical[b]	77%	77%	67%	44%	100%	100%	89%	44%
1	no	yes	no	no	yes	yes	no	no
2	yes	no	no	no	yes	yes	yes	no
3	no	no	yes	no	yes	yes	yes	no
4	yes	yes	yes	yes	yes	yes	yes	yes
5	yes	yes	yes	no	yes	yes	yes	yes
6	yes	yes	no	no	yes	yes	yes	no
7	yes	yes	yes	yes	yes	yes	yes	yes
8	yes	yes	yes	yes	yes	yes	yes	no
9	yes	yes	yes	yes	yes	yes	yes	yes

Note. Utterances were considered inaccurate yet consistent in instances where the child was consistent in their production (e.g., /kʌ/ consistently produced as /tʌ/)

[a] Percentages represent the proportion of TD children who demonstrated each measure

[b] Results for the clinical sample are shown for each individual child. yes = demonstrated that feature (i.e., accurate / consistent)

For the clinical sample, Table 7 also shows whether individual children were accurate and consistent in their productions of the single and alternating syllables. As shown in the table, most of the clinical sample were accurate and consistent in their productions of the single syllables. The single syllables /pʌ/ and /tʌ/ were produced accurately by 77% of the children, and for those who were not accurate, they were nevertheless consistent in their productions (this occurred when, for example, a participant said /tʌ/ for /kʌ/, but was consistent in the use of this substitution pattern). Six out of the nine accurately produced /kʌ/, and eight were consistent with this syllable (regardless of accuracy). With the tri-syllabic sequence, participants 1, 2, 3, 5 and 6 were not able to produce the sequence at all. For these children, when multiple repetitions of the train were attempted, all but participant 5 were inconsistent. Note that participant 5 appeared to have difficulty understanding the task, and (possibly due to severely impaired receptive language skills and echolalia) copied the first part of the sequence only (i.e., would not wait for the end of the model). Participant 8 was able to produce the sequence accurately, but was inconsistent when producing multiple repetitions. Participants 4, 7 and 9 were both accurate and consistent. Considering both accuracy and consistency of the tri-syllabic sequence, participants 1, 2, 3 and 6 were both inaccurate and inconsistent in their productions. They were also the same participants with sequencing difficulties identified on the VMPAC.

Preschool Repetition Test (Roy & Chiat, 2004)

The children's performance on the repetition test are summarised in Table 8. Scores for the TD sample are summarised and presented as group data for comparison with the clinical sample. Consistent with previous findings (Chiat & Roy, 2007; Roy & Chiat, 2004), the TD children performed well on this task, with overall accuracy (i.e., percentage of items produced correctly) ranging from 70% to 97%. These children made few phoneme errors, and rarely lost syllables or made stress errors. In contrast, performance of the clinical sample varied considerably.

Table 8

Accuracy, Percent Phonemes Correct (PPC), and Prosodic Errors on the Preschool Repetition Test for the TD and Clinical Samples

			Prosodic Errors		
	Accuracy[b] %	PPC[c]	Syllable Loss[d]	Stress Errors	Total[e]
TD Sample[a]					
M	87	96.8	0.5	1.4	1.9
SD	7.2	2.1	1.0	1.4	2.0
Clinical Sample					
1	25*	70.0*	1	3	4
2	3*	25.9*	13*	11*	24*
3	3*	43.4*	7	7	14*
4	47*	77.1*	12*	3	15*
5	42*	81.3*	1	3	4
6	11*	70.5*	11*	2	13
7	75	91.0*	1	0	1
8	53*	84.3*	6	0	6
9	92	97.6	1	1	2

[a] *N*=21 [b] Percentage of items produced correctly [c]Percent Phonemes Correct

[d] total number of syllables lost, out of a total of 72 syllables [e] includes stress and syllable loss errors

* statistically significantly different (at $p = .05$) from the TD sample

Accuracy and PPC. Case by case analyses using Crawford and Howell's (1998) modified t-test procedure revealed the entire clinical sample, bar participants 7 and 9, to have significantly lower accuracy scores compared to the TD children. PPC scores were also significantly lower for each of the clinical sample participants except participant 9.

Syllable loss and stress errors. As shown in Table 8, the TD children lost very few syllables, and rarely produced stress errors. Due to large violations to assumptions underlying parametric analyses (i.e., highly skewed and non-normal distributions), boxplots were used to examine differences between the clinical sample children and the TD sample for these measures. Cases were considered 'extreme' where they were at least 3 times the interquartile range in distance from the 75th percentile. For syllable loss, this was the case for participants 2, 4 and 6. For stress errors, participant 2 met this criterion. However, when considering the total prosodic errors, participants 2, 3 and 4 were extreme in the number made, and thus were considered to be showing prosodic anomalies based on our criteria.

Table 9 displays PPC for each syllable length. Regression slopes were calculated for PPC as a function of syllable length. The mean slope for the TD sample was negative, indicating PPC generally decreased as syllable length increased. The clinical sample participants also showed this trend, but individual slopes were steeper. Analysis of regression slopes relative to the intercept value revealed significantly larger values for participants 2, 3 and 4, reflecting proportionately larger decreases in accuracy as syllable length increased; $t(20) = 2.28$, $p = .01$, $t(20) = 5.21$, $p < .01$, and $t(20) = 3.58$, $p < .01$, respectively. These participants were thus considered to show the feature of increasing errors as syllable length increased.

Table 9

Percent Phonemes Correct (PPC) for each Word Length on the Preschool Repetition Test

	Word length (# syllables)			Regression slope	Proportion[b]
	1	2	3		
TD Sample[a]					
M	97.7	98.2	95.6	-1.1	-.01
SD	2.8	2.3	3.9	2.7	.03
Clinical Sample					
1	82.5	58.5	73.0	-4.8	-.06
2	32.0	22.5	26.5	-2.8	-.08*
3	57.0	26.5	36.5	-10.3*	-.17*
4	96.5	77.5	70.0	-13.3*	-.12*
5	86.0	86.0	76.5	-4.8	-.05
6	82.5	55.0	78.0	-2.3	-.03
7	100.0	89.5	89.0	-5.5	-.05
8	93.0	80.0	83.0	-5.0	-.05
9	100.0	96.5	93.0	-3.5	-.03

[a] Mean PPC (and standard deviations) for the TD sample as a group are shown. Individual scores are provided for the clinical sample [b] Regression slope relative to intercept value

* statistically significantly different (at $p = .05$) from the TD sample

CAS Features

CAS-related features (as operationally defined for this study) observed in the clinical sample are displayed in Table 10. Of the nine children, four (participants 5, 7 8 and 9) do not display any of the features. The remaining five children demonstrated at least three CAS features. As shown by the tallying of number of features, participants 2 and 3 show the most number of features.

Table 10

Summary of CAS Features Observed in the Clinical Sample

	Participant								
Feature	1	2	3	4	5	6	7	8	9
Inconsistency[a]	✓	✓	✓	-	-	✓	-	-	-
Prosodic anomalies[b]	-	✓	✓	✓	-	-	-	-	-
High incidence of vowel errors[c]	✓	✓	✓	✓	-	✓	-	-	-
Speech sequencing difficulties[d]	✓	✓	✓	-	-	✓	-	-	-
Increased errors as length/complexity increase[e]	-	✓	✓	✓	-	-	-	-	-
Total (out of 5)	3	5	5	3	0	3	0	0	0

Features based on [a] inconsistency score (DEAP) [b] prosodic errors on PRT [c] PVC (DEAP) [d] VMPAC sequencing score/sequencing difficulties on DDK task [e] PCC regression slopes

The scores on the CAS-related measures for the clinical sample were also examined in terms of severity. Although the features represent those that are commonly reported to be specific to CAS, there is still no validated set of criteria to

differentially diagnose CAS. Importantly, it is not clear to what extent individual features (and in the way they have been measured in the present study) are *never present* in children with phonological disorders, or even children with language deficits. In fact, many features purported to be characteristic of CAS are present in children with general speech sound disorders (McCabe et al., 1998). Severity is one way to capture the idea of a continuum of apraxic symptoms (Hodge, 1994; Strand, 2002), and to document the nature of these features in children. The degree to which each child's score on the measures differed from the measure of central tendency was examined and is displayed in Table 11[1]. Where they were appropriate, t-scores were used, with z-scores calculated for measures which used standardised assessments (i.e., inconsistency and PVC). A negative sign indicates instances where performance was better than the TD sample (i.e., the opposite direction to what would be expected in the case of CAS).

Table 11

Severity of Apraxic Symptoms, as Measured by t- and z-scores for each CAS Feature

Participant	Incons	Prosodic	PVC	Sequencing	↑err/length	TOTAL
1	9.9	1.5	9.3	1.2	1.6	23.5
2	9.9	11.5	98.2	3.6	2.3	125.5
3	6.6	6.5	53.7	3.5	5.2	75.5
4	0.8	7.0	12.5	0.8	3.6	24.7
5	3.8	1.5	6.1	1.8	1.3	14.5
6	4.4	6.0	17.2	-0.2	0.7	28.1
7	1.0	0.0	1.4	-1.7	1.3	2.0
8	3.3	2.5	12.5	-1.7	1.3	17.9
9	0.5	0.5	-0.2	-1.6	0.7	-0.1

Note. Incons = Inconsistency, PVC = Percentage of vowels correct, Sequencing = sequencing errors, ↑err/length = increasing errors with length

[1] With reference to Table 11, it is noteworthly that some extreme scores were obtained. To ensure these did not unduly influence the children's overall severity ranking, the data was also analysed as ordinal data. Kendall's coefficient of concordance supported the overall ranking of the children and suggested good levels of agreement across the measures, W = .68, p = .001.

Tallying of scores indicated participant 2 to show the greatest severity of CAS symptoms, followed by participant 3. Participants 4, 6 and 1, who presented with 3 of the CAS features, showed scores that were less extreme than participants 2 and 3 in terms of difference from the typical sample. Considering severity as well as the number of CAS features, participants 2 and 3 show the largest number of features, and participant 2 stands out as being the most severely affected on these features.

Discussion

Study 2A aimed to provide a detailed description of the participants whose infant data were to be analysed in study 2B. In the absence of validated criteria for CAS diagnosis for research purposes, the need for such detailed participant information has been called for (American Speech-Language-Hearing Association, 2007). The speech and language abilities of nine children who had taken part in a community screening program as infants and were still presently receiving speech pathology services (the clinical sample) were examined in detail. A group of 21 children with typically developing speech and language skills were also assessed on experimental tasks to provide a normative reference sample. Performance on a number of standardised and experimental tasks allowed investigation of CAS features, operationally defined in the present study, in order to provide a more comprehensive participant description. As with other clinically-obtained samples, considerable heterogeneity was observed in the clinical sample, however some children displayed a considerable number of CAS features.

Language and Phonological Skills

Five of the clinical sample participants displayed receptive language impairments, with three of these showing severe difficulties in this area. Expressive language deficits were observed in six of the children. Three of the clinical sample children scored within the normal range on both receptive and expressive components of the CELF-P (participants 7, 8 and 9); these participants also showed no CAS features. Of those children with language difficulties in at least one area, a notable receptive-expressive gap (with stronger receptive skills) was evident for two participants (1

and 2). Participants 3, 4 and 5 demonstrated severe language impairments in both receptive and expressive domains.

All participants who demonstrated CAS features also had language difficulties. Participant 2, showing the greatest number and severity of CAS features, had a receptive-expressive gap, with mild receptive but severe expressive language impairment. This is consistent with typical accounts of children with CAS (Hall, 2003a). Participant 3, who showed all five CAS features but not to as great a severity, showed severe deficits in both receptive and expressive language areas. The presence of language difficulties in participants with CAS is consistent with previous reports (Ekelman & Aram, 1983; Lewis et al., 2004). Lewis and colleagues (Lewis et al., 2004), for example, documented language deficits in 8 out of their 10 CAS children tested at preschool age. Most of these children had difficulties in both receptive and expressive areas, and the difficulties persisted into school age for all but one child. Syntactic deficits were documented in a group of eight 4 to 11 year old children studied by Ekelman and Aram (1983). As outlined in the introductory chapter, although expressive language deficits are commonly reported for children with CAS, the presence of receptive language deficits in this population has not been thoroughly investigated.

Assessment of the children's phonology indicated that six of the nine clinical sample participants had difficulty with consonants (PCC standard score more than one standard deviation lower than the mean). Of these, all but one (participant 5) also showed vowel errors outside the expected range for their age. The presence of vowel errors was the most consistent CAS feature identified in the present sample. However, the actual percentage of vowels correct score was near or above 90% for most of these, reflecting the rarity of vowel errors in normal development. It is noteworthy that one participant (participant 2) showed a severely depressed PVC score. The variation in severity of vowel errors highlights the need for debate regarding the operational definition of this feature, and more detailed participant description in CAS studies.

Three children in the present study did not present with any phonological difficulties. Interestingly, they were also the same participants who showed normal language skills and did not demonstrate any CAS features. Associated areas of development, such as preliteracy skills, were not assessed however.

CAS Features

Four of the clinical sample children displayed none of the CAS features. The remaining five demonstrated 3 or more features. As displayed in Tables 10 and 11, participants 2 and 3 showed the highest number of features, with participant 2 also demonstrating the highest severity on these features. Four children (participants 1, 2, 3 and 6) displayed significant inconsistency and three (participants 2, 3 and 4) had significant prosodic anomalies. Vowel errors were present in five of the children (participants 1, 2, 3, 4 and 6), difficulties with speech sequencing were observed in four children (participants 1, 2, 3 and 6), and increased errors on longer items were demonstrated in three (participants 2, 3 and 4).

Analysis of CAS features, when operationally defined, suggested that many of the features were present in children for whom CAS was not suspected by their speech pathologists, and who did not present on the whole with a clinical profile suggestive of CAS. Although only three children (participants 1, 2 and 3) were identified by their managing clinician as having features consistent with CAS, five showed at least one CAS feature. This finding is consistent with McCabe et al. (1998), who found evidence of CAS features in the case note profiles of children with a range of speech sound disorders. It also suggests that such features should be investigated further in larger scale studies of both CAS and other speech sound disorders, and that the number and severity of such features may have important diagnostic implications.

The method by which CAS features were determined to be present requires consideration. Very few studies have detailed criteria used for determining the presence or absence of CAS features. Most describe a list of features but do not explicitly describe how each has been identified. In the present study, inconsistency was determined via the presence of token to token variability on a standardised tool

(DEAP). This method of quantifying inconsistency is consistent with one of the three formulae investigated by Betz and Stoel-Gammon (2005), and that recommended by Dodd and colleagues (Dodd et al., 2002). Similarly, the presence of a high incidence of vowel errors and speech sequencing difficulties were also determined via standardised assessment, allowing more objective comparison to age-norms.

In contrast, the presence of prosodic anomalies and increased errors as length/complexity increased were not determined via standardised assessments. Prosodic errors were captured via a combination of syllable loss and lexical stress errors, because both contribute to the percept of prosody (Velleman & Shriberg, 1999). However, some researchers have focussed on only one aspect as being indicative of prosodic disturbances. Shriberg and colleagues (Shriberg, Campbell et al., 2003), for example, investigated computed metrics relating specifically to lexical stress errors on trochaic words. In earlier studies (Shriberg et al., 1997c) showing 50% of CAS children to have inappropriate stress, coding of this feature was based on sentence level excess-equal stress, with the omission of weak syllables being a candidate explanatory factor in the percept, supporting the combined measure utilised here. Further research is needed to tease apart the nature of prosodic disturbances reported in children with CAS, with particular reference to syllable loss as a contributing factor to lexical and sentential stress.

The presence of increasing errors as length/complexity increases is another feature often mentioned but rarely defined in CAS research. Peter and Stoel-Gammon (2008) reported making a clinical judgement on the presence/absence of this (and other) features. Productions of simple versus complex word structures were compared to determine this feature, but detail objectifying the amount and nature of the comparisons were not included. Most other research provides less detail, making it difficult to compare results across studies. In the present research, performance on the Preschool Repetition Test (Roy & Chiat, 2004) was used to determine the presence or absence of this feature. Statistical comparison of PPC regression slopes relative to intercept values, designed to objectively assess the concept of increased errors on longer words whilst allowing for individual differences in accuracy at the one-syllable level, were informative for researching this feature. However, the procedure requires further investigation to confirm its use in quantifying this feature.

Issues such as the possible confounding influence of working memory, commonly reported in language disordered children in general (Weismer, Evans, & Hesketh, 1999), could make this measure problematic.

The approach of quantifying features of CAS, although vital given the need for detailed participant description, involved making multiple comparisons. As such, there is potential for compromising the Type 1 error rate. This issue is considered further in the general discussion, in the context of the entire thesis.

Summary

Using operationally-defined criteria for observing the presence/absence of commonly reported CAS features in a clinical sample, five children showed the presence of at least one feature. However, some children showed a greater number and severity of involvement of features, supporting either the presence of CAS in only these one or two children, or the notion of a continuum of praxis-type involvement in children with a range of speech and language impairment. Two children showed the most number of features (participants 2 and 3, with all five features). Taking into account severity of symptoms, one of these participants (2) showed particularly severe involvement on the characteristics, including an extremely high incidence of vowel errors, high rates of inconsistency, and severe speech sequencing difficulties. This participant was also the only one to show both high rates of syllable loss and lexical stress deficits, and clinically was observed to present as the 'clearest' case of CAS.

CAS features, defined according to one set of operationally descriptive criteria, were present in some children who had not been suspected of CAS. This finding is consistent with reports of wide-ranging criteria used by speech pathologists in establishing a diagnosis of CAS (Forrest, 2003), as well as that of reported CAS features in the general speech-impaired population (McCabe et al., 1998). The results of the present study highlight the need to use objective criteria in determining the presence of CAS features. Few studies have attempted to do this, despite frequent calls to include detailed participant information in CAS research, especially whilst the phenotype of the disorder is still under investigation (American Speech-

Language-Hearing Association, 2007). It has been suggested that features identified based on clinical experience may prove to be those that eventually meet psychometric requirements for inclusion as differentially diagnostic criteria (American Speech-Language-Hearing Association, 2007). However, until such criteria are established, future research should include detailed information on how such features are identified in participants. Importantly, the next phase of the present study examined retrospective infant data available for these same children, allowing investigation of hypothesised differences in prelinguistic vocal development.

Study 2B – Analysis of retrospective infant data

Introduction

According to developmental conceptualisations of CAS, the underlying core deficit involved in the disorder is one that would be evident prelinguistically (Maassen, 2002). A hypothesised core deficit originating in speech motor control within a developing system predicts an atypical pattern of early phonetic and language development in infancy (Maassen, 2002). A small number of studies provide support for this prediction, with case reports of delayed or decreased babbling in children with CAS (Velleman, 1994). In the absence of longitudinal studies following children with CAS from infancy (Zeigler & Maassen, 2004), retrospective research designs may be used to further explore these hypotheses (Sivberg, 2003).

Retrospective parental report (Study 1, Highman, Hennessey, Sherwood, & Leitão, 2008) provided preliminary support for group differences in overall rates of vocalisation and babbling, later emergence of two word combinations, and commonly constrained speech and motor development in sCAS children. However, the group pattern was not present for all children with the disorder, and direct observations of the children were not available. Phase one of the present study documented CAS features in a unique clinical sample of children with corresponding retrospective infant data available. Phase two reports on the nature of these infant data, comparing each child from the clinical sample to a larger sample of infant data for children without identified communication impairments.

Retrospective data from two comparison groups, collected when the infants were 8-9 months of age, were utilised. The false positives group comprised infants who, although initially had failed a communication screen, had subsequently demonstrated normal language development in a more in-depth follow up assessment conducted within a month of the screening. Data for these infants were thus considered to be the closest to typically developing that were available, and make an ideal comparison for the clinical sample participants' infant data. A second retrospective group (the true positives group) comprised data from infants who had failed the communication screen and also failed the more in-depth assessment of language development, and thus were considered to be 'at risk' of communication impairment at that stage. By way of their not being identified for participation in Study 2A, the large majority of this group are expected to have resolved their initial delays (Rescorla, 2002) and thus make an interesting comparison to those children who demonstrated CAS features in the preschool age.

The hypothesised speech motor control core deficit in CAS, affecting the infant's subsequent sensorimotor development and formation of linguistic representations, is expected to be evident prelinguistically in the form of inadequate syllabic articulatory gestures in infancy (Guenther, Hampson, & Johnson, 1998; Levelt, Roelofs, & Meyer, 1999). In the present study, it was expected that this would be reflected in limited or absent babbling at 9 months and selectively depressed expressive language scores on standardised assessments (as in infancy these capture information relating to prelingustic vocalisations as well as emerging word use). The standardised assessment of receptive language, as well as measures of general conceptual development, would be expected to be typically developing and not different to the false positives comparison group.

Hypotheses

Given the heterogeneity in the clinical sample and presence of CAS features in many of the cases, infant data were expected to show similar complexities. For the clinical sample children who displayed a high number and severity of CAS features, the following hypotheses were explored:

Participants with a high number and greater severity of CAS features at 3-4 years would show:

1. deficits in expressive language in infancy, as evidenced by scores on standardised language assessment more than one standard deviation below the mean
2. a relative expressive-receptive gap in infancy, as evidenced by a significant dissociation (Crawford, Garthwaite, & Gray, 2003) with a relative strength in receptive abilities
3. a lack of consonant-vowel babble at 9 months
4. reduced number of consonant sounds in infancy

With respect to the retrospective comparison groups, it was expected that the groups would differ from each other in terms of infant language scores, in line with their groupings, with the false positives showing stronger language skills. Case comparisons of the clinical sample participants to both of these groups were expected to reveal subtleties regarding developmental profiles in infancy. In particular, it was hypothesised that individuals with CAS profiles at preschool age would show differences in vocalisations and language ability consistent with the hypothesised core deficit in CAS, when compared to the false positives group. However, when comparing to the true positives group (who were also identified in infancy as 'at risk' of communication impairment), there was potential for the clinical sample participants to present similarly in infancy on gross measures of language development.

No specific hypotheses were developed for the clinical sample participants showing phonological and/or language issues. However, given the suggested close link between prelinguistic and later linguistic development in normal and disordered acquisition alike (Locke & Pearson, 1992; Oller, 2000; Oller, Eilers, Neal, & Schwartz, 1999; Stoel-Gammon, 1992), it was anticipated that some deficits in areas of development in infancy may be observed in these children also. These children were not expected to show specifically impaired speech motor development (evident in prelinguistic vocalisations) in the context of typical receptive language development, however.

Method

Participants

Clinical sample. The clinical sample participants from Study 2A, characterised in terms of their speech and language profiles and presence of CAS-related features, were treated as individual cases for analysis of the retrospective data. They represented a unique group of children who had data available from when they were infants, from their participation in a community speech pathology program. Relevant clinical information for each participant is summarised in Table 12, including a review of the number of CAS-related features they displayed in Study 2A.

Retrospective comparison groups. Retrospective data from 205 infants were available for use in the analyses of infant data. Like the clinical sample participants, the comparison group infants had participated in the community program and thus had retrospective data available. The following inclusionary criteria were met: no significant medical issues (e.g., Down Syndrome, cleft palate), term birth, singleton, exposed only to English, and were aged at least 3 years at the time the retrospective data were analysed. As infants, the participants had failed the screen, described below, and had either subsequently passed or failed the assessment – 'false positives' and 'true positives', respectively. Data for the false positives group were thus taken to represent a typically developing comparison group for the clinical sample. The true positives group represented children who, as infants, failed a communication assessment, and thus were at risk of communication impairments, similar to the clinical sample participants. However, in contrast to the clinical sample participants, these children were not subsequently identified with communication impairments.

Table 12

Characteristics of the Clinical Sample

ID	# CAS features (severity)	Language skills
1	3 (mild)	mildly impaired expressive language age-appropriate receptive language
2	5 (severe)	severely impaired expressive language mildly impaired receptive language
3	5 (moderate)	severely impaired expressive language severely impaired receptive language
4	3 (mild)	severely impaired expressive language severely impaired receptive language
5	0	severely impaired expressive language severely impaired receptive language
6	3 (mild)	severely impaired expressive language mildly impaired receptive language
7	0	age-appropriate expressive language age-appropriate receptive language
8	0	age-appropriate expressive language age-appropriate receptive language
9	0	age-appropriate expressive language age-appropriate receptive language

Note. Severity of CAS features based on data summarised in Table 11.

General information about the clinical sample and retrospective comparison groups is displayed in Table 13. As shown in the table, the three samples were similar in terms of age screened and assessed. The infants were screened, on average, at 8-9 months of age, and assessed a month later at 9-10 months. The false and true positives groups did not differ significantly on age screened, $t(203) = 0.64$, $p = .52$. Statistically, they differed on age assessed, $t(203) = 1.98$, $p = .049$, however in real terms this was an average of one week, the effect size was small ($d = .30$) and the ranges were similar.

Table 13

Age at Screen, Age at Assessment, and Gender for the Clinical Sample, False Positives and True Positives Groups

	Screen Age (weeks)	Ax[a] Age (weeks)	Gender (%)
Clinical Sample[b]			M: 7 (78%)
M	35.3	41.0	F: 2 (22%)
SD	2.3	3.4	
Range	30-38	37-48	
False Positives[c]			M: 29 (51%)
M	35.2	40.1	F: 28 (49%)
SD	1.3	2.8	
Range	32-38	36-47	
True Positives[d]			M: 77 (52%)
M	35.4	39.2	F: 71 (48%)
SD	1.5	2.6	
Range	30-39	36-48	

[a] Ax = assessment [b] $n = 9$ [c] $n = 57$ [d] $n = 148$

Retrospective Measures

Ages and Stages Questionnaire (ASQ, Bricker et al., 1999). The ASQs are a series of questionnaires developed to evaluate development across five areas (corresponding to the subscales): Communication, Gross Motor, Fine Motor, Problem Solving and Personal-Social. Parents are required to observe their child's behaviour in each of these areas, following set probes, and to indicate whether each behaviour is present (yes, sometimes, or not yet). The ASQ was administered by child health nurses, either face to face with the parent, or by parents completing the forms at home under the direction of the child health nurse. Summary scores for each developmental area are calculated and compared to the recommended cutoff scores. Psychometric properties of the ASQs are reported to be adequate, with internal consistency coefficient alphas for the questionnaire used in the present study ranging from .72 to .79, test-retest reliability and inter-observer reliability (percentage of agreement) both at 94%, concurrent validity (reported in terms of sensitivity and specificity in relation to the comparison instrument) of 72% and 86% respectively (Boyce & Poteat, 2005; Bricker et al., 1999).

WILSTAAR screen. Developed as a communication screening tool, the WILSTAAR screen consists of nine questions relating to receptive language/listening skills and expressive skills in the infant (Ward, 1992). Receptive items predominantly focus on auditory-perceptual skills such as whether the infant responds to someone calling his/her name, and notices and responds to familiar and unusual sounds (see Appendix D). The sole expressive item relates to the child's use of variegated babbling. Parents were asked the questions at the child's routine 8 month check up. Child health nurses documented parents' responses and the screens were then sent to local speech pathologists for scoring. The infant is considered to have failed the screen if any items are failed. Information derived from the screen included whether the child had passed or failed each component (i.e., receptive and expressive). The screen was reported to have strong concurrent and predictive validity by Ward (1992), but the screening and intervention program has subsequently been questioned by other researchers (St James-Roberts, 2004). Despite its limitations, the screen provided information on the infant's use of sounds and vocalisations, as well as listening skills/receptive language.

WILSTAAR record forms. A standard record form was also used to collect data on the infants at initial assessment. This included a standard set of questions relating to overall development, current communication skills, and the parent's use and style of language in the home. The record form includes two questions directly relevant to the current study, relating to the infant's use of sounds. These were: 'Does s/he make sounds much?', and 'What sounds does s/he make now?' Specific responses to each question were available for the clinical sample. For the retrospective comparison groups, responses to the second question were available in the database as a string variable, but no responses had been recorded for the first question.[2]

Receptive-Expressive Emergent Language assessment (REEL-2, Bzoch & League, 1991). The REEL-2 is a clinical assessment tool used to evaluate emergent receptive and expressive language ability in infants and toddlers. Administered by the clinician, it consists of a series of questions asked to the parent regarding aspects of receptive and expressive language development in their child. Comparison with the standardisation sample allows interpretation of the raw score and conversion to expressive, receptive and overall language quotients (EQ, RQ and LQ, respectively). Despite reporting internal consistency coefficients of above .92, and adequate test-retest reliability (.79, .76 and .80 for the three composite quotients), there has been some criticism of the robustness of the tool (Mitchell, 1985). An updated version has recently been released (Bzoch, League, & Brown, 2003) with improved standardisation procedures. However, as the 3rd edition was not available when the community program was implemented, only REEL-2 data were available for the present study.

Procedure

Infant Data. These data were collected during 2001-2002 for the community program, and were used retrospectively in this study. Infants and their parents attended their usual 8 month-old screening with their child health nurse (CHN). Immediately prior to or during this appointment, parents completed the ASQ. The WILSTAAR screen was administered by the CHN and sent to the speech pathologists for scoring. Infants who failed the screen were visited at home by a

[2] The investigator did not have access to individual record forms for the comparison groups, only the recorded data in a database

speech pathologist pair, with the REEL-2 and observations being conducted during this visit. If infants passed the language assessment, they were classed as a false positive; if they failed they were considered a true positive and were offered the speech pathology program (see Appendix C for details of the program). Data used for the present study, obtained with permission from the Child and Community Health Branch of the Department of Health, Western Australia, were collated in a database for further analysis. Note that ASQ data were not available for three of the clinical sample participants.[3]

Coding of reported vocalisations. For the purpose of the present study, the principal investigator transformed relevant measures contained in the database into formats suitable for analysis. To quantify the presence of canonical babbling and number of consonant sounds reported, raw data containing the responses to relevant questions from the WILSTAAR record forms were converted to numeric codes. The presence of well-formed syllables (from parent description) indicated the presence of canonical babbling (Oller, Eilers, & Basinger, 2001). Each supra-glottal consonant sound reported in the parent's list of sounds their infant was making were counted and tallied to form a total number of sounds reported.

Data Analyses

Data were screened for adherence to assumptions underlying the relevant analyses, and any violations are reported within the results of that particular analysis. Initial analyses on the two retrospective comparison groups were conducted to identify similarities and differences between the two groups. Each of the clinical sample participants' retrospective data were then systematically compared to the retrospective comparison groups', using the modified t-test procedure (Crawford, Garthwaite, Howell, & Gray, 2004) described in Chapter 1. The approach has been demonstrated to suitably control for the Type I error rate regardless of the control sample size, and is robust even when used with highly skewed data (Crawford & Garthwaite, 2005). Furthermore, in order to test for a dissociation or differential deficit in an individual, the Revised Standardised Difference Test (RSDT) method (Crawford & Garthwaite, 2005) was applied.

[3] This was because administration of the ASQ had not been introduced in the particular health service area at the time that these infants took part in the program – the primary investigator of the present study was not aware of this until the retrospective record forms were requested from health services.

Results

Overview

For each relevant measure, group results are provided for the retrospective comparison groups (false positives and true positives). Individual data for each of the clinical sample are also detailed, followed by the case comparisons investigating deficits and dissociations.

Ages and Stages Questionnaire (ASQ). Means and standard deviations for each subscale of the ASQ for the two retrospective comparison groups, and individual scores for the clinical sample are shown in Table 14. As shown in the table, subscale means were similar for the false positives and true positives groups, with no significant differences observed for communication, $t(173) = 0.84$, $p = .40$, gross motor, $t(173) = 0.20$, $p = .34$, fine motor, $t(173) = 0.60$, $p = .55$ and personal-social, $t(173) = 0.86$, $p = .39$ domains. Mean score for the problem solving subscale was significantly higher for the false positives group, $t(173) = 2.04$, $p = .04$.

Case comparisons of the clinical sample participants indicated that compared to the false positives group, participant 6 scored significantly lower on the communication subscale, $t(45) = 2.10$, $p = .02$. Scores for each of the remaining participants were not statistically significantly different on this domain, all p values > .10 (see Appendix E). Subscale scores for gross motor, fine motor, personal-social and problem solving were not statistically significantly different from the false positives group for any of the clinical sample, gross motor p values all > .05 ($p = .068$ for participant 7), fine motor p values all > .05, problem solving p values all > .20, and personal-social p values all > .20 (listed in Appendix E).

When compared to the true positives group, participant 6 again scored significantly lower in the communication domain, $t(130) = 1.99$, $p = .02$. No other participant scored significantly different to the false positives on this subscale, p values all > .10 (see Appendix E). Scores for the clinical participants on the remaining subscales were not statistically different to the false positive comparison group, gross motor p values all > .05, fine motor p values all > .05, problem solving p values all > .10, and personal-social p values all > .10 (listed in Appendix E).

Table 14

ASQ Scores for each Developmental Area for the False Positives, True Positives and Clinical Sample Participants

	Comm	GM	FM	Prob	Pers-Soc
False positives[a]					
M	50.6	50.8	57.0	55.8	54.7
SD	9.7	10.3	5.3	7.2	7.9
True positives[b]					
M	49.2	51.1	57.5	53.1	53.5
SD	9.6	9.8	4.6	7.7	7.5
Clinical sample[c]					
1	-	-	-	-	-
2	40	60	60	50	50
3	55	60	50	60	60
4	-	-	-	-	-
5	-	-	-	-	-
6	30	50	50	55	50
7	40	35	55	55	55
8	60	60	60	60	60
9	60	60	60	60	60

Note. ASQ data were not available on all participants as there were instances where ASQ collection had not been initiated in some health services. The scores represent values out of a possible total of 60. Comm = communication, GM = gross motor, FM – fine motor, Prob = problem solving, Pers-Soc = personal-social

[a] $n = 45$. [b] $n = 130$. [c] - = missing data.

WILSTAAR screen. By definition of their group membership, the infants in the retrospective comparison groups failed at least one section of the WILSTAAR screen.[4] Table 15 details the proportion of infants in each group failing the receptive and/or expressive components. As can be seen in the table, the most obvious difference between the groups relates to the proportion of infants failing the expressive component (35% of the false positives and 12% of the true positives, compared to nearly 89% of the clinical sample). Individual results for the clinical sample infants are displayed in Table 16. All but one (participant 3) of the clinical sample children failed the expressive component, and seven of the nine failed one or more receptive items. The two who didn't fail any receptive items were participants 2 and 3.

REEL-2. Mean receptive quotient (RQ), expressive quotient (EQ), and language quotient (LQ) for the two retrospective comparison groups are shown in Table 17. The homogeneity of variance assumption was violated for the EQ and LQ. Results were thus interpreted with the adjusted degrees of freedom (reported within the statistical sentence) where appropriate. Individual data for the clinical sample children are also displayed. Consistent with their groupings, the false positives group had significantly higher language scores on initial assessment than did the true positives group, $t(203) = 10.1$, $p < .01$, $t(74) = 13.0$, $p < .001$ and $t(74) = 12.7$, $p < .001$, for RQ, EQ, and LQ, respectively. Quotients were below the normal range (defined as more than one standard deviation from the mean and therefore below 85) for six, eight and nine of the clinical sample infants, for the receptive, expressive and overall language areas, respectively. Particularly low expressive quotients were observed for participants 4 and 6.

[4] Records for participant 3 revealed that, despite being included in the program, he did not fail any component of the screen. It is not clear why this was so. Potentially, the parent may have responded to the set questions but then revealed verbally that he was not producing variegated babbling. There was no record to confirm if this was the case however.

Table 15

Percentage of Infants Failing the Receptive and/or Expressive Component of the WILSTAAR Screen

		GROUP					
		False Positives		True Positives		Clinical	
		Pass	Fail	Pass	Fail	Pass	Fail
Receptive Items							
Notice sounds		96.5	3.5	94.6	5.4	88.9	11.1
Notice own name		93.0	7.0	88.5	11.5	55.6	44.4
Notice sounds as much as previously		98.2	1.8	95.9	4.1	100	0
Ignore interesting sounds		84.2	15.8	90.5	9.5	22.2	77.8
Turn a 2nd time to noise		93.0	7.0	91.9	8.1	88.9	11.1
Ever concerned hearing		89.5	10.5	87.2	12.8	77.8	22.2
	M	92.4	7.6	91.4	8.6	72.2	27.8
Expressive Item							
Variegated babble		62.4	35.1	87.3	12.7	11.1	88.9

[a] $n = 57$ [b] $n = 148$ [c] $n = 9$

Table 16

Individual Data for the Clinical Sample on the WILSTAAR Screen

	Participant								
	1	2	3	4	5	6	7	8	9
Receptive Items									
Notice sounds	P	P	P	P	P	P	**F**	P	P
Notice own name	P	P	P	P	**F**	**F**	P	**F**	**F**
Notice sounds as much as previously	P	P	P	P	P	P	P	P	P
Ignore interesting sounds	**F**	P	P	**F**	**F**	**F**	**F**	**F**	**F**
Turn a 2nd time to noise	P	P	P	P	P	P	**F**	P	P
Ever concerned re: hearing	P	P	P	P	**F**	P	**F**	P	P
Expressive Item									
Variegated babble	**F**	**F**	P	**F**	**F**	**F**	**F**	**F**	**F**

Table 17

Mean Receptive Quotient (RQ), Expressive Quotient (EQ) and Language Quotient (LQ) for the False Positives and True Positives Groups, and Individual Quotients for the Clinical Sample

	RQ	EQ	LQ
False positives [a]			
M	109.4	100.8	103.2
SD	21.1	17.1	15.9
True positives [b]			
M	80.8	69.0	74.5
SD	17.1	10.9	10.1
Clinical sample [c]			
1	78	78	78
2	89	56†	72
3	67 †	89	78
4	60 †	20 † *	40
5	60 †	50 † *	55
6	89	11 † *	44
7	67 †	67 †	67
8	89	67 †	78
9	82	82	82

[a] $n = 57$ [b] $n = 148$ [c] $n = 9$

† = statistically significantly different ($p = .05$) to the false positives groups. * = statistically significantly different ($p = .05$) to the true positives group

Using the false positives as the comparison group, single case comparisons suggested significantly lower receptive quotients for clinical sample participants 3, 4, 5 and 7, $t(57) = 1.99$, $p = .03$, $t(57) = 2.32$, $p = .01$, $t(57) = 2.32$, $p = .01$, and $t(57) = 1.99$, $p = .03$ respectively. All other clinical sample children scored receptive quotients that did not differ significantly to the false positives group, all p values >

.05 (see Appendix E). In contrast, expressive quotients were significantly lower for participants 2, 4, 5, 6, 7 and 8, $t(57) = 2.59$, $p = .01$, $t(57) = 4.70$, $p < .01$, $t(57) = 2.49$, $p = .01$, $t(57) = 5.21$, $p < .01$, $t(57) = 1.98$, $p = .03$, and $t(57) = 1.98$, $p = .03$, respectively.

When compared to the true positives group, receptive quotients of the clinical sample participants did not differ significantly, all p values > .05 (listed in Appendix E). Three children had expressive quotients significantly lower than the true positives group: participant 4, $t(148) = 4.48$, $p < .01$, participant 5, $t(148) = 1.74$, $p = .04$, and participant 6, $t(148) = 5.30$, $p < .01$. A fourth (participant 3) showed a significantly higher expressive quotient than the comparison group, $t(148) = 1.83$, $p = .04$. Expressive quotients for the remaining five clinical participants did not differ significantly when compared to the true positives group, all p values > .10 (Appendix E).

Presence of canonical babble and number of sounds reported. The percentage of infants in each group reported to be producing canonical babble at 9 months, as well as the number of consonant sounds reported, is displayed in Table 18. The table shows, descriptively, that a lower proportion of the clinical sample children were producing canonical babble. However, the average number of consonant sounds reported was similar for the three groups (with a lower range for clinical sample). Case comparisons of the number of sounds reported for individual clinical sample children did not evidence any significant differences, except for participant 6 whose report of no consonant sounds was significantly lower than that for both the false and true positives comparison groups, $t(57) = 2.46$, $p = .01$, and $t(148) = 2.02$, $p = .02$, respectively (see Appendix F for individual comparisons).

Table 18

Proportion of Infants Producing Canonical Babble, and Number of Consonant Sounds at 9 months

	Producing canonical babble	# consonant sounds		
	%	*M*	*SD*	*Range*
False Positives[a]	89.5	2.85	1.15	0-6
True Positives[b]	86.5	2.40	1.19	0-5
Clinical Sample[c]	77.8	2.3	1.32	0-4

[a] $n = 57$ [b] $n = 148$ [c] $n = 9$

The descriptions of vocalisations reported on the screening (8 months) and assessment (9 months) were explored further for the clinical sample participants. Table 19 displays this information. The description for the 8 month data came from the expressive item on the WILSTAAR screen – a yes/no question about variegated babble. Most, therefore, show only that a 'no' response was reported, indicating the child was not producing variegated babble. However, some child health nurses had also documented additional detail that revealed information about whether the infant was producing any sounds at all; this is reported in the table as it is rare and potentially informative. The 9 month descriptions came from the WILSTAAR record form where two questions were relevant: 'Does s/he make sounds much?', and 'What sounds does s/he make now?". Many responses to the first question were not recorded, or vague responses such as 'makes more now' were documented. Frequency comments were included for participants 2 and 6 – rarely/infrequently, and participants 5 and 9 – often/frequently.

Table 19

Description of Vocalisations for Clinical Sample Participants

Participant	Description of vocalisations
1	8 months: squeals but no individual sounds 9 months: ‘dada’, ‘nana’
2	8 months: no canonical syllables 9 months: ‘da’, ‘ga’, ‘hu’ – *all rarely*
3	8 months: producing variegated babble 9 months: ‘dada’, ‘mumu’, ‘baba’
4	8 months: no variegated babble 9 months: ‘bubub’, ‘dadad’
5	8 months: no variegated babble 9 months: ‘mumum’, ‘dadad’, ‘nanana’, ‘bubub’, ‘aa’ *all produced often*
6	8 months: no variegated babble 9 months: cooing only, *doesn’t make sounds much*
7	8 months: producing canonical babble 9 months: ‘dad’, ‘muma’, ‘nana’, ‘bub’
8	8 months: no variegated babble 9 months: ‘mumma’, vowel sounds
9	8 months: no variegated babble 9 months: ‘mumma’, ‘dadda’, ‘nanna’ *produced often*

Note. Descriptions of vocalisations are orthographical representations

Dissociations. Applying the RSDT (Crawford & Garthwaite, 2005) procedure, dissociations between receptive and expressive language abilities were explored in the clinical sample participants. Table 20 summarises the results of the individual comparisons with the false positives group, and also illustrates where a significant dissociation was detected. Participants 2 and 6 showed a pattern of classical dissociation, whereby expressive quotients were significantly lower than the false positives group 'norms', receptive quotients did not differ statistically, and the discrepancy scores were significantly larger than the comparison group, $t(56) = 1.76$, $p = .04$, for participant 2, and $t(56) = 4.56$, $p < .01$, for participant 6. Participant 4, however, showed a pattern of 'strong' dissociation (Crawford & Garthwaite, 2005), whereby receptive *and* expressive scores were significantly lower than the comparison sample, but the discrepancy between scores was also significantly larger, $t(56) = 2.54$, $p = .01$ (with expressive markedly lower than receptive).

Comparison of the REEL-2 scores of the clinical sample participants to the true positives group are displayed in Table 21. Compared to this similarly 'at risk' retrospective comparison group, participants 4 and 6 showed a classical dissociation, with receptive skills not significantly different but expressive skills that were significantly lower than the comparison sample, and a significant discrepancy, $t(147) = 2.40$, $p < .01$, and $t(147) = 4.25$, $p < .01$, respectively. A significant dissociation was not observed for participant 5, $t(147) = 0.39$, $p = .71$. With an expressive quotient significantly higher than the comparison group, participant 3 showed a classical dissociation in the opposite direction, $t(147) = 1.94$, $p = .03$.

Table 20

Summary of Deficit and Dissociations in Receptive and Expressive Language for the Clinical Sample compared to the False Positives Group

ID	RQ	EQ	Dissociation? (type [a])
1	ns [b]	ns	✘
2	ns	significantly lower	✓ (classical)
3	significantly lower	ns	✘
4	significantly lower	significantly lower	✓ (strong)
5	significantly lower	significantly lower	✘
6	ns	significantly lower	✓ (classical)
7	significantly lower	significantly lower	✘
8	ns	significantly lower	✘
9	ns	ns	✘

[a] *Classical* dissociation occurs when one area is significantly lower, there is no significant difference in the other area, *and* the difference is significantly greater than the distribution of differences for the comparison sample. *Strong* dissociation occurs when both areas are significantly lower than the comparison sample but there is also a significant difference between the two areas within the individual (Crawford & Garthwaite, 2005) [b] ns = non significant

Table 21

Summary of Deficit and Dissociations in Receptive and Expressive Language for the Clinical Sample compared to the True Positives Group

ID	RQ	EQ	Dissociation? (type)
1	ns [b]	ns	✗
2	ns	ns	✗
3	ns	significantly higher	✓
4	ns	significantly lower	✓ classical
5	ns	significantly lower	✗
6	ns	significantly lower	✓ classical
7	ns	ns	✗
8	ns	ns	✗
9	ns	ns	✗

[a] *Classical* dissociation occurs when one area is significantly lower, there is no significant difference in the other area, *and* the difference is significantly greater than the distribution of differences for the comparison sample. *Strong* dissociation occurs when both areas are significantly lower than the comparison sample but there is also a significant difference between the two areas within the individual (Crawford & Garthwaite, 2005). [b] ns = non significant

Discussion

The prelinguistic communication and developmental abilities of nine clinically-ascertained children were investigated in Study 2B. A retrospective data design was applied, with the aim to explore the core deficit underlying CAS by focussing on infant profiles, where the confounding influence of development itself may be minimised (Bishop, 1997; Karmiloff-Smith, 1998). The children, described in detail

in Study 2A, presented with a range of speech and language profiles at 3 to 4 years of age, including varying numbers and severity of CAS features. Children demonstrating a high degree of CAS features were of particular interest in evaluating the hypotheses regarding a core deficit in speech motor control for this disorder. As with variability in their 3 to 4 year old skills, the clinical sample showed varying communication profiles in infancy. Single case methodology was applied to compare each clinical sample child with comparable infant data for two retrospective comparison groups - those that had initially failed a language screen but subsequently passed a more comprehensive language assessment (false positives) and those that failed both the screening and assessment (true positives).

Consistent with their groupings, the false positives group showed significantly stronger receptive and expressive language abilities than the true positives. As their language scores were within the normal range, they represent the closest to typically developing infants that we have data for, so made an ideal comparison with the clinical sample. The true positives group also provided an important comparison as they represent children who, as infants, were showing delayed language precursers, but were not identified as speech/language impaired preschoolers.

On measures of general development (i.e., the ASQ), the comparison groups differed only in the problem solving domain, with higher scores for the false positives group. Given that the inclusion criteria ensured that none of the infants had overall global developmental delays, the difference may represent a qualitative one, rather than suggesting a general cognitive disparity for the two comparison groups. Indeed, the problem solving scale has a number of items with a fine motor skill prerequisite. For example, items include the ability to pass a toy back and forth between hands, banging a toy on a surface, and banging toys together. Thus, the relative difference in scores for this subscale may be more suggestive of subtle differences in this domain (specifically, hand-banging perhaps, given the overall fine motor scores do not differ otherwise).

Participants with a High Degree of CAS Features

Ranked according to the number of CAS-related features shown in Study 2A, the clinical sample participant with the highest number and greatest severity of features (participant 2) demonstrated significantly poorer expressive skills and a significant dissociation in receptive-expressive abilities in infancy when compared to typically developing infants (the false positives group). This pattern, coupled with information regarding his prelinguistic vocalisations, is consistent with the deficit in speech motor control hypothesised by a number of researchers as being explanatory in CAS.

A core deficit in speech motor control, interpreted in the context of developmental models of speech production, predicts a relatively isolated deficit prelinguistically, affecting vocalisations but not conceptual development. Data relating to this child's prelinguistic vocal development indicated that he was not producing any canonical syllables at 8 months, and was rarely using any by 9 months, consistent with the notion of restricted syllabic articulatory gestures. This deficit was observed in the context of intact conceptual development, as indicated by scores in the normal range for receptive language, problem solving and personal-social domains. According to developmental models of speech production (Levelt et al., 1999), an absence of such articulatory gestures would negatively affect the development of the protosyllabary – limiting subsequent vocabulary acquisition and associated linguistic development. Records for this child confirm the rarity of canonical babbles in the prelinguistic period, and later restricted vocabulary and language development.

These results represent unique data revealing the profile in infancy of a child with a subsequent clinical diagnosis of CAS. They are consistent with the few descriptions of early development of children with CAS present in the literature (Velleman, 1994), and theoretical models of vocal development (Guenther, Ghosh, & Tourville, 2006; Levelt et al., 1999; Westermann & Miranda, 2004). Of the two children with CAS described by Velleman (1994), one did not produce any babbling at the age normally expected and showed late emergence of first words. The frequency of babbling was reduced in the other child. Additional information relating to receptive language and overall developmental abilities were not provided in these case reports, however.

The pattern described above was not observed for participant 3 of the present study, who also showed a high number of CAS features in Study 2A. This child displayed infant data in contrast to what would be expected if a diagnosis of CAS was appropriate and a core deficit in speech motor control underlies the disorder. Receptive scores were significantly lower than the comparison groups', and expressive skills were higher. Moreover, description of his prelinguistic vocalisations suggested the presence of age-appropriate vocalisations including canonical babble. This suggests that either hypotheses relating to the core deficit in CAS as being evident prelinguistically to have not been supported in this case, or that the child does not have CAS. It is the case that clinically, this child did not present as a 'clear case' of CAS[5], suggesting that the measures utilised in Study 2A may not have adequately captured the characteristics of importance in a clinical diagnosis. Alternatively, and in keeping with the variability observed in Study 1, it is possible that while a core speech motor control deficit, evident prelinguistically and in the context of intact abilities in the conceptual domain may account for some cases of CAS, it may not explain *every* case. Dyspraxic features may emerge in conjunction with linguistic development for other reasons; for example, if a core deficit in the organisation of hierarchical units was present (Velleman & Strand, 1994).

The three clinical sample participants who presented with some but not all of the five CAS features in study 2A demonstrated varying profiles in infancy. Participant 6 presented similarly to participant 2: significantly lower expressive scores and dissociated receptive and expressive skills, plus an absence of canonical babble and limited vocalisations at 9 months. This child's expressive scores in infancy were not only poorer than the false positives group, but also the similarly at risk true positives. Participant 4 presented similarly in terms of depressed expressive language ability in infancy, but he also demonstrated receptive skill delays. In contrast, participant 1, who presented at preschool age with 3 of the CAS features described in study 2A, demonstrated infant data that did not 'stand out' when compared to the other retrospective groups. Receptive and expressive language scores were not different to either the false positives or the true positives group, and while she was not producing variegated babble by the 8 month screen, she was producing two canonical syllable

[5] Based solely on the primary investigator's clinical judgement – a clinical 'gestalt' or impression without objective quantification

types by 9 months (no comment on frequency was made, however). These varying profiles present a less-than-clear picture of CAS in infancy. However, until such time as a validated set of criteria for research purposes is established for diagnosing CAS, it is difficult to ascertain how much of the variability observed in this study relates to the heterogeneity in the participants at preschool age.

It is interesting to note, also, that the pattern of dissociation observed for participant 2 when compared to the false positives group was not observed when the true positives were used as the comparison group. Recall that this group, like the clinical sample participants, had been identified as 'at risk' of communication impairment in infancy by way of failing both a screening tool and a standardised assessment of language development. The analyses using this group for comparison suggest that, on standardised assessments of language ability in infancy, the nature of later skills or deficits may not be immediately apparent. That is, standardised assessments may provide only 'gross' information relating to language ability and risk status for communication impairments. This is consistent with research highlighting the potential for large changes in profiles on standardised assessments over time (Darrah, Hodge, Magill-Evans, & Kembhavi, 2003), and clinical cautions to avoid applying labels such as CAS prematurely (Davis & Velleman, 2000).

The Remaining Clinical Participants

Clinical sample children who at 3 to 4 years of age presented with receptive and/or expressive language deficits, and even those with apparently 'resolved' difficulties, but no CAS features, also showed lowered scores on communication assessments in infancy. This finding is consistent with research identifying the predictive but variable nature of communication abilities in infancy for later language performance (Reilly et al., 2007; Zubrick, Taylor, Rice, & Slegers, 2007). It is also consistent with research supporting the importance of prelinguistic vocalisations in subsequent lexical and phonological growth (Oller et al., 1999; Whitehurst, Smith, Fischel, Arnold, & Lonigan, 1991). The profiles of individual children are of interest: participant 5, who presented with SLI at preschool age, demonstrated significantly lowered receptive *and* expressive language skills in infancy, no dissociation in these domains, and was documented to be babbling frequently at 8 to 9 months. This may

be instructive with respect to the underlying nature of SLI and how it differs to that involved in CAS.

Participants who essentially demonstrated age-appropriate speech and language skills when assessed at the preschool age revealed that they presented similarly to the 'false positives' group in general, with lowered expressive language scores but age-appropriate vocal development by 9 months. These results may suggest that these children were similar to the late talkers group that go on to be 'late bloomers' – that is, they catch up their initial delays by preschool age (Rescorla, 2002; Rescorla, Ross, & McClure, 2007). Further research is needed to expand on these exploratory results.

Limitations

A number of limitations in the present study should be noted. Firstly, the retrospective data presented here were collected for purposes other than for the present study. As such, there were instances of missing data for the ASQ, limiting the scope of comparisons for the children. Also an artefact of utilising existing data, some of the measures themselves may not be specific enough to adequately explore the speech motor system. For example, although data relating to the parents' description of the sounds made were available, it was not clear whether parents had listed as many sounds as possible (i.e., *all* the sounds made by the infant), or whether they had given some *examples* of sounds made. Data relating to the frequency of production would also have been valuable. Frequency data were not available, so it was not clear in most cases just how vocal the infant was.

It was also not feasible to confirm the communication status of the (de-identified) children from the retrospective comparison groups. It is possible that some of these children, whilst not being identified for participation in Study 2A, did have speech and/or language deficits, in the case, for instance, that they may have moved inter-state or actively dis-engaged from local speech pathology services. However, it is likely that the large majority of these children did go on to have normal speech and language development (Rescorla, 2002).

Importantly, the infants had all taken part in a community screening program (see Appendix C). Those that had failed the screen and the assessment (i.e., the true positives group and the clinical sample) had gone on to receive a brief, parent-based intervention program in an attempt to facilitate language development. Clinical sample children, because they had been subsequently identified with speech and/or language deficits past the program duration, had then received varying types and amounts of clinic-based therapy. This is a major contributing confound to the present study's results, and although unavoidable given the use of retrospective data, should be carefully considered when interpreting the results. It is not clear, for example, whether differences in the children's current profiles related to the type and amount of therapy they had received, rather than differences that may have been present in infancy.

Conclusion

Irrespective of the differences observed in infant profiles, and the limitations associated with using pre-existing data, the presence of a profile consistent with the hypotheses in the most 'clear' CAS case does provide preliminary support for the notion of a core deficit in speech motor control. The data provided a rare opportunity to examine the prelinguistic profile of children with CAS features. A dissociated pattern of development, with selectively impaired speech motor control in the presence of intact receptive language and conceptual development supports the notion of limited articulatory gestures as being involved in (at least some) cases of CAS. Further research is needed to extend the investigation of prelinguistic vocalisations in CAS. In particular, longitudinal investigations that allow speech and language trajectories to be observed over time (and not just retrospectively) are sorely needed. Study 3, described in the following chapter, utilises such an approach.

CHAPTER 4

STUDY 3. LONGITUDINAL INVESTIGATION OF CAS FEATURES IN AN AT-RISK INFANT SAMPLE

Introduction

Studies 1 and 2 provided preliminary support for the notion of a core deficit in speech motor control in CAS, evident in the early vocalisation and communication profiles of children with the disorder. However, Study 1 was based on parent report, relying heavily on the recall abilities of the parents. Infant data available for Study 2 showed variable profiles for the children showing the most number of CAS features at 3-4 years of age, with the child identified with the greatest number and severity of features showing the predicted profile at 9 months, but the picture for other children with CAS features being less clear. Moreover, as the infant data were collected for a purpose other than the study in question, conclusions were restricted by the nature of data available.

Given the dynamic and interactive nature of development, and difficulty disentangling core deficits from subsequent deficits, longitudinal investigations may provide the best opportunity to document the natural course of developmental disorders such as CAS (Bishop, 1997; Maassen, 2002; Zeigler & Maassen, 2004). Such paradigms may also contribute to the identification of early features for identifying infants at increased risk. The need for longitudinal studies of CAS commencing in infancy has been identified by a number of researchers (American Speech-Language-Hearing Association, 2007; Zeigler & Maassen, 2004). However, because it is not possible to diagnose CAS in infants and toddlers (Davis & Velleman, 2000), there is a lack of longitudinal studies focussing on the disorder, especially from an early age. A number of large scale more general longitudinal studies of speech and language development have recently been reported (e.g., Reilly et al.,

2007; Zubrick, Taylor, Rice, & Slegers, 2007). These studies seek to identify early predictors of later speech and language impairment. However, none have reported findings specifically relating to CAS.

As outlined in the introductory chapter, there is evidence to support the notion of familial aggregation in CAS (Lewis, Freebairn, Hansen, Taylor et al., 2004; Thoonen, Maassen, Gabreels, Schreuder, & de Swart, 1997), and thus the method of identifying infants for longitudinal study via family history of the disorder. Such paradigms have been used in the study of other complex developmental disorders such as autism spectrum disorders (ASD) (Iverson & Wozniak, 2007; Landa & Garrett-Mayer, 2006) and dyslexia (Koster et al., 2005). Landa and Garret-Meyer (2006), for example, studied the early language abilities of infant siblings of children with autism. Iverson and Wozniak (2007) similarly targeted infant siblings in their investigation of early vocal-motor development in ASD. In such studies, overall group patterns of performance are described, as they are informative about the possibility of a broader phenotype even when some siblings do not go on to receive an ASD diagnosis or the study timeframe does not allow diagnosis to be confirmed (Iverson & Wozniak, 2007). Other studies have, after investigating siblings longitudinally, subsequently reported case studies of children who received a confirmed diagnosis a number of years later (e.g., Bryson et al., 2007). Studies aiming to identify early precursors of dyslexia have also employed methods whereby infants to be investigated are identified via their positive family history of the disorder (Koster et al., 2005; Lyytinen et al., 2001). Koster and colleagues (Koster et al., 2005), for example, studied lexical acquisition in toddlers with at least one parent and one first-degree relative showing a dyslexic profile and compared them to toddlers with no such family history. Group differences were reported prior to the establishment of whether the children went on to receive a diagnosis of dyslexia. It is preliminary research such as this that paves the way for further, more focussed research looking at confirmed cases of the disorder in question.

The present study employed a similar paradigm, whereby infants with a family history of CAS were recruited for participation in longitudinal investigation. Analyses of the vocalisations and developmental profiles of these children were conducted in order to further explore the core deficit in CAS. As with Studies 1 and 2, the present

study investigated the hypothesis of a core speech motor control deficit in CAS. According to this theory, if impaired speech motor control underlies CAS, it can be expected to be evident in prelinguistic vocalisations (Maassen, 2002). The aim of the present study was to extend the findings of Studies 1 and 2, via longitudinal investigation of an at-risk sample of children from infancy to 2 years of age. As well as providing the opportunity for more direct investigation of early vocalisation and language development, it allowed investigation of perceptual and acoustic aspects of vocalisation. Predicted patterns of development investigated in the present study are described further below.

Auditory-Perceptual skills. The hypothesis of an initial core deficit in speech motor skills, interpreted in the context of Levelt's modified developmental model of speech production (Levelt, Roelofs, & Meyer, 1999), does not predict initial deficits in auditory-perceptual skills in early infancy. Some researchers have reported deficits in fine-grained perceptual skills in school age children with CAS (Groenen, Maassen, Crul, & Thoonen, 1996; Maassen, Groenen, & Crul, 2003). Such deficits may be accounted for by interpreting CAS in a developing system, where abnormal or absent babbling may lead to subsequent differences in both production *and* perception (Westermann & Miranda, 2004), due to the role of phonetic skill in establishing later representations used for perception. However, initially in infancy, auditory-perceptual skills are presumed intact.

Motor development. An isolated speech motor control deficit does not directly predict delayed or disordered general motor development. However, such deficits are frequently reported in children with CAS. As described in Chapter 1, research suggests a close relationship between canonical babbling and repetitive motor movements such as hand banging (Ejiri & Masataka, 2001), with commonly-timed neural growth in the respective brain areas (Locke & Pearson, 1992). In a dynamic system it may be possible that a constraint in one area (e.g., speech motor control, affecting canonical babbling) may negatively affect another closely related area (e.g., hand banging), or vice versa (Mitchell, 1995). Thus delays in fine motor development may not be unexpected in infants at risk of CAS.

Cognitive and conceptual skills. Given the proposed initial independence of the conceptual and speech motor systems in early development (Levelt et al., 1999), an isolated core deficit in speech motor control would be expected to be found in the presence of intact conceptual development. Notwithstanding considerations of the existence of CAS in other complex neurobehavioural disorders (American Speech-Language-Hearing Association, 2007), normal conceptual and cognitive development would thus be expected in its idiopathic form. This would be reflected in measures of communicative intent and conceptualisation. The development of intentionality and use of gestures, for example, would be expected to be typically-developing.

Receptive and expressive language skills. As with auditory-perceptual skills and conceptual development, receptive language would be expected to be initially unaffected in CAS under a core speech motor control deficit account. A relative strength in receptive language would be expected to be most evident very early on in development. Over an extended period of time, receptive skills may be compromised secondary to any emerging perceptual deficits (Westermann & Miranda, 2004) and/or the impact of limited expressive language on opportunities for receptive language development. However, initially, receptive language would be expected to be intact, reflected in age-appropriate receptive vocabulary and comprehension abilities.

Expressively, the proposed account of CAS predicts specific and persistent sequelae for vocabulary acquisition and syntactic development. As described earlier, in typical development the initially independent conceptual and speech motor systems are coupled when the child first produces real words (Levelt et al., 1999). If a restricted set of articulatory gestures exists, the emergence of first words may be delayed or limited in terms of the number and rate new words are produced. Although lexical concepts would continue to be acquired, deficient speech motor abilities would restrict the normal rapid expansion of expressive vocabulary. A high degree of homophony would be expected in these words (Davis & Velleman, 2000). As early syntactic development (particularly the emergence of two-word combinations) is thought to be contingent on the acquisition of a critical mass of vocabulary items (Moyle, Ellis Weismer, Evans, & Lindstrom, 2007), deficits in this area of expressive language would be expected. Expressive delays are expected even prelinguistically, because of the way ‘expressive language’ is evaluated in infancy – vocal development typically features within assessment tools for this area.

Speech motor control. As outlined in the introductory chapter, atypical vocalisation development is predicted if a speech motor control deficit is involved in CAS. Central to the hypothesis herein and consistent with features proposed to be associated with CAS (Davis & Velleman, 2000), an affected infant may be expected to have delayed, reduced or absent babbling, reduced frequency of canonical babbles, limited consonant and vowel inventory, limited phonotactic variation, and acoustic patterns consistent with impaired speech motor control. Deficits in speech motor control are often exposed in acoustic analyses of speech production (Kent, 2000). In the case of CAS, if the presumption of the core deficit responsible for the disorder as being present from birth is accurate, evidence of it may be reflected in acoustic measures of the infant's initial syllabic gestures.

Syllable duration and formant frequency measures may reveal irregularities in the earliest instance of speech motor control. Syllable duration, for example, may be longer if overall within-syllable articulation rates are slower or there is less coarticulation (Bahr, 2005). Nijland and colleagues (Nijland, Maassen, & van der Meulen, 2003) reported longer segment durations for children with CAS. Bahr (2005) also found CAS children to display significantly longer word durations, compared to both children with phonological disorder and those with typically developing speech skills. Coarticulation data however, have shown inconsistent results. Some studies have found coarticulation to be stronger in children with CAS (Nijland, Maassen, van der Meulen et al., 2003), whereas other studies have found it to be more variable and idiosyncratic in others (Nijland et al., 2002). Examination of duration in infant canonical syllables, irrespective of the direction of prediction for CAS, would provide information on the nature of these initial syllabic articulatory gestures.

Analyses of formant and fundamental frequencies may reveal further information about speech motor control (Kent, 1976). Mean fundamental frequency of phonation is typically stable until 9 months of age, before it decreases until 3 years of age (Kent, 1976; Voperian & Kent, 2007). It has been suggested that fundamental frequency measures may reflect neurological maturity (Bosma, Truby & Lind, 1965, as cited in Kent, 1976). Fundamental frequency, and its perceptual correlate, pitch, play an important role in signalling adult-like phonation (Oller, 2000).

Measures of the first two formants (F1 and F2) in vowel production are typically reflective of tongue height and advancement, respectively (Voperian & Kent, 2007), and therefore may be sensitive to developmental changes in speech motor control and use of the vowel space. Children with CAS are frequently reported to have limited vowel inventories and show a tendency to neutralise vowels (Davis, Jacks, & Marquardt, 2005), features often hypothesised to relate to impairments in speech motor control (Bahr, 2005). Velleman and colleagues reported higher F2 values in children with CAS, compared to children with phonological impairment (Velleman, Huntley, & Lasker, 1991, as cited in Bahr, 2005), and although Bahr did not see a similar trend, the overall results were hypothesised to reflect a limited use of the vowel space. Typical methodology for measuring vowel space involves comparing F1 and F2 values for 3 or 4 target vowels, allowing the area traversed by the articulators to be depicted. A reduced vowel space and more centralised vowels would be reflected in a smaller planar area. Although it is not possible to control the type of vowels produced by infants to ensure a range of targets are attempted, less variable or more restricted F1 and F2 values would still be expected if vowels were restricted. In infants, average frequencies for Formants 1 (F1) and 2 (F2) have been found to be relatively stable from 4 months of age until around the second birthday (Robb, Chen & Gilbert, 1997, as cited in Voperian & Kent, 2007), despite rapid increases in vocal tract length and subsequent non-linear changes at later ages (Voperian & Kent, 2007). Although variability in formant frequencies typically reduces with age, some research has suggested this progresses faster for F1 (where variability is minimal by 3 years of age) than F2 (Nittrouer, 1993).

Consistent with theories of articulatory phonology (Browman & Goldstein, 1992) and explanations of the movements underlying babbling (MacNeilage & Davis, 1990), tighter coupling between the articulators may be found in the case of impaired or delayed speech motor control. As has been suggested for other areas of motor development (Hay, 1984), the movements underlying the syllabic articulatory gestures of babbling are proposed to be initially ballistic, with individual articulators showing gradually increased independence over time (Browman & Goldstein, 1992). Impaired motoric skill in this area may result in restricted use of the vowel space, consistent with reports of vowel neutralisation in children with CAS (Davis et al., 2005).

Dissociation between conceptual and speech motor control abilities. Finally, if Levelt and colleagues' (Levelt et al., 1999) model of early development is accurate, an infant with an isolated core deficit in speech motor control would be expected to show a significant dissociation between measures of conceptual and speech motor control development (Maassen, 2002). Conceptualiser skills would be predicted to be intact in such an infant, with significantly impaired speech motor abilities, and a significant dissociation between the two areas. This would be expected to be evident pre-linguistically.

Hypotheses

The present study aimed to investigate longitudinally the vocalisation, language and general development of infants at increased risk of CAS. As an initial grouping, infant siblings of children with CAS were compared to infants with no such family history (and thus no putative genetic risk). Infant siblings of children with CAS may show features consistent with a broad phenotype of a 'verbal trait deficit' (Lewis, Freebairn, Hansen, Taylor et al., 2004); thus, hypotheses relating to group profiles were:

Infant siblings would show, relative to the comparison group infants:

1. Lower expressive language scores
2. Lower scores on speech sound development
3. Lower scores on fine motor development

Furthermore, those infant siblings showing evidence of a communication deficit (at 2 years of age) would be considered at even greater risk of CAS, or a more general speech/language delay (Lewis, Freebairn, Hansen, Taylor et al., 2004). Their data would be inspected for evidence of CAS-related features. Hypotheses relating to infants showing such features were that they would show:

a. A lack of canonical babbling at 9 months
b. A persistently restricted phonetic inventory
c. Reduced rate of pre-linguistic canonical vocalisations

d. Acoustic correlates of a deficit in speech motor control: that is, longer syllable durations, atypical fundamental frequency, restricted use of the vowel space (reflected in measures of F1 and F2)
e. A significant dissociation in speech motor and conceptual development, with conceptual abilities intact

It is acknowledged that due to the absence of specific and validated diagnostic criteria for CAS in this age group, variables used to classify infants at further increased risk of CAS are (necessarily) circularly-linked to the hypotheses. However, investigating hypotheses c, d and e in such infants would provide further evidence for or against the proposed core deficit in CAS.

Method

Overview

Study 3 involved longitudinal data collection on infants with a family history of CAS and infants with no such familial risk, followed by detailed analysis of vocalisation data for infants of interest following the longitudinal observation period. Infants who had an older sibling with a clinical diagnosis of CAS were recruited, along with a comparison group of infants with no family history of speech, language or literacy difficulties. The infants were assessed and tracked longitudinally over a 15 month time frame (from 9 to 24 months of age). Data at 2 years of age identified two infants whose communication skills were not developing appropriately for their age. Their profiles over time were examined for evidence of any CAS-related features, and their vocalisation data were investigated in more detail and compared to the typically developing comparison group infants.

Participants

Sixteen infants and their primary caregivers (all mothers in this study) took part in Study 3. Recruitment facilitators were advised about the study and were requested to distribute information and consent forms to parents of infants who met the criteria outlined below. All infants were from monolingual English home environments, and

did not have any identified medical, cognitive or physical disability. Socio-economic status, estimated by postcode data, was predominantly middle-class. All infants in the sibling group were referred for an audiological assessment to confirm normal peripheral hearing acuity and middle ear function, to remove this as a possible confounding factor.

Siblings group. The siblings group consisted of eight infants (four boys and four girls), all younger siblings of children with a clinical diagnosis of CAS. Speech Pathologists in the Perth metropolitan area were made aware of the study via an electronically distributed flyer, as well as direct requests at meetings and/or via telephone. They were requested to identify in the first instance, children on their caseload who they believed met the clinical criteria for CAS. No specific guidance on which particular features were diagnostic of CAS were provided as the primary goal was not to evaluate epidemiological issues surrounding CAS (e.g., how many children with a confirmed diagnosis of CAS have an infant sibling with concerning features) but rather to recruit as many potential infant siblings who may be at greater risk of CAS as possible. The speech pathologists distributed information about the study to families who also had a baby (biologically related to the child with CAS features) who was under the age of 9 months at that time, and to families who were expecting a new baby. Two exceptions to the target age were made: siblings (SIBS) 3 and 5 were 10 months and 12 months respectively when the study commenced, but were included in light of the small numbers that were expected and rarity of infant data. Recruitment commenced early in the PhD process (out of necessity given the relatively low incidence of CAS), and some families 'registered' interest in taking part in the study early on, being contacted as their infant approached the age to commence participation. Data were first collected on the infants when they were 9 months of age. Chronological age (weeks) at each data collection stage is displayed in Table 22.

Comparison group. The comparison group consisted of eight infants (four boys and four girls) with no reported family history of speech, language or literacy difficulties. Child Health Nurses were asked to identify infants who, according to their observations, were developing appropriately, had no significant health, medical or developmental issues, and who did not have a family history of speech or language difficulties (based on parent report). Parents were advised that they could participate in a study relating to infant vocalisations and were given an information sheet and

consent form if they were interested. Chronological age at each data collection point is displayed in Table 22. Independent samples t-tests confirmed no significant differences in chronological age between the two groups, $t(13) = 0.24$, $p = .82$, $t(14) = 0.37$, $p = .72$, $t(14) = 0.40$, $p = .69$, $t(14) = 1.95$, $p = .07$, $t(10) = 0.61$, $p = .56$, for the 9, 12, 15, 18 and 24 month collection points, respectively.

Table 22

Mean Chronological Age (weeks) of the Sibling and Comparison Group Infants at each Data Collection Point. Standard deviations are shown in parentheses

	Data Collection Time point				
	9 month	12 month	15 month	18 month	24 month
Sibling	39.5 (3.5)	52.8 (0.8)	66.0 (1.3)	79.4 (1.4)	105.9 (3.8)
Comparison	39.9 (0.8)	52.7 (0.5)	65.8 (1.2)	78.0 (1.4)	104.8 (0.8)

Materials

Audio equipment. A Sony lapel condenser microphone and Sony Minidisc recorder (MZ-N710) were used to record the infants' vocalisations in stereo wave format with 16 bit digitisation and a sampling rate of 44100 Hz. The lapel microphone was clipped to the infant's bib or clothing, approximately 5cm from the mouth, and was connected to the minidisc recorder via an extended cord. A small velcro 'dot' was used to secure the cord over the infant's shoulder, so that the cord ran behind the infant, minimising the likelihood that it would be distracting. During the 12 and 18 month sessions, many of the infants were particularly aware of and interested in the microphone, and attempted (at times successfully) to pull it off their clothing. On these occasions, the mother and investigator attempted to distract the

infant via offering toys, food or noise-makers while the microphone was re-attached. Occasionally, the microphone had to be re-positioned to the infant's shoulder where it was not as easily detected by the infant.

WILSTAAR screen (Ward, 1992). The WILSTAAR screen, previously described in Study 2B (Chapter 3) was also used in the present study.

Ages and Stages Questionnaire (ASQ, Bricker et al., 1999). The ASQ questionnaire, described in Study 2B, was also utilised in the present study. Psychometric details of the ASQ and the WILSTAAR screen were discussed in Chapter 3.

Communication and Symbolic Behaviour Scales (CSBS) – Developmental Profile (Wetherby & Prizant, 2002). The CSBS is an assessment package for investigating communication abilities in infants and toddlers. It comprises the Infant-Toddler Checklist, Caregiver Questionnaire, and Behaviour Sample. All three tools were used in the data collection process for this study, although the Behaviour Sample was used only as a context to collect a vocalisation sample at 12 and 18 months. Each component of the CSBS has demonstrated sound psychometric properties, including strong internal consistency, test-retest stability, and inter-rater reliability, and sound content, face, construct and criterion validity (Wetherby, Allen, Cleary, Kublin, & Golstein, 2002; Wetherby & Prizant, 2002). Internal consistency for the checklist and caregiver questionnaire, measured with Cronbach's coefficient alpha, is strong, with coefficients ranging from .95 to .97. Test-retest stability coefficients for all standard scores are significant and large, ranging from .65 to .93. Strong correlations between the components, and evidence of good predictive validity are also documented (Wetherby & Prizant, 2002)

The Infant-Toddler Checklist, designed as a first-level screening tool, consists of 24 questions covering the seven areas of emotion and eye gaze, communication, gestures, sounds, words, understanding and object use. Parents are asked to indicate the response (from a choice of 3 to 5 depending on the item) that most accurately describes their child's current skills/behaviour. Raw scores calculated for the seven areas described above (clusters) are used to generate three composite scores: social composite, speech composite, and symbolic composite, with corresponding standard scores and percentile ranks. The social composite is derived from the emotion and eye gaze, communication, and gestures areas; the speech composite from the sounds

and words areas; and the symbolic composite from the understanding and object use areas. Composite scores have a mean of 10 and standard deviation of 3. 'Cut-off' levels for concern are set at 1.5 standard deviations below the mean, equivalent to a standard score below 7.

The Caregiver Questionnaire consists of 41 items within the same seven areas as the Infant-Toddler Checklist, plus four open-ended questions. As with the checklist, the items ask the parent to rate the presence and frequency of occurrence of a range of communication behaviours for their child. However, the caregiver questionnaire, comprising many more items, is more comprehensive than the checklist, and allows standard scores and percentile ranks to be calculated for the seven cluster areas (emotion and eye gaze, communication, gestures, sounds, words, understanding and object use), as well as the three composite scores (Social, Speech and Symbolic). Both cluster and composite scores have means of 10 and standard deviations of 3, and the recommended cut-off scores are as per the checklist. Examples of items from each of the cluster areas are provided in Table 23. Although the checklist and caregiver questionnaire target the same areas, with the caregiver questionnaire being more comprehensive, both were included in the initial assessment. This was to provide comprehensive data that were appropriate to use for each of the ages (in the case of using the caregiver questionnaire), but also in acknowledgement that information relating to which infants passed the more streamlined screen would have practical/clinical implications.

In the CSBS behaviour sample, a standard set of toys and communicative temptations are used to collect a communication sample. Following a short warm up, the infant is seated in a high chair, with the parent and examiner either side. Communicative temptations, book sharing, symbolic play, language comprehension and constructive play probes are then administered. The communicative temptations section involves the systematic presentation of a wind-up toy, balloon, bubbles, and clear jar containing desired food items (in this study sultanas were used), following a standard set of procedures. The behaviour sample is suitable for use on infants from 12 months of age.

Table 23

Example Items from each Area of the Communication and Symbolic Behaviour Scales (CSBS) Caregiver Questionnaire (Wetherby & Prizant, 2002)

Cluster area	Example Question
Emotion and Eye Gaze	When your child is playing with a toy, does he/she look at you and then back at the toy?
Communication	Does your child try to get your attention when you are busy doing something, such as when you are talking with an adult or preparing a meal?
Gestures	Which of the following gestures have you seen your child use? (list of 10 provided)
Sounds	Children use sounds to communicate in vocal play before they use sounds in words. Does your child use a variety of different consonant sounds, such as “ba”, “ga”, “ta”, and “da”, either in vocal play or in words?
Words	Does your child use words to communicate (if so, which of the following….?)
Understanding	Does your child respond when you call his/her name (for example, by looking/turning head)?
Object Use	Does your child build or arrange toy objects (for example, build a tower of blocks, stack rings, put puzzle pieces together)?

Receptive-Expressive Emergent Language Test, third edition (REEL-3, Bzoch, League, & Brown, 2003). The third edition of the REEL was administered in Study 3, due to its improved psychometric properties (compared to earlier versions). The REEL-3 is a clinician-administered test of emerging language in children from birth to three years of age. Parents are asked a standard set of questions on receptive and

expressive language, with entry, basal and ceiling criteria well defined in the manual. Raw scores for the two scales: receptive and expressive language, are calculated and converted to standard scores (referred to as ability scores) and percentile ranks. The REEL-3 has sound psychometric properties, with good internal consistency (e.g., coefficients of .92 and .93 for the receptive and expressive subtests respectively), and strong test-retest reliability and inter-rater reliability (e.g., mean Cohen's kappas of .99 for both subtests, Hurford & Stutman, 2004). Validity (content, criterion-related, and construct) were found to be similarly acceptable (Hurford & Stutman, 2004).

Language Development Survey (LDS, Rescorla, 1989). Originally designed as a screening tool for identifying delayed expressive language in toddlers, the LDS has been shown to be an efficient yet reliable parent-report measure of expressive vocabulary and word combination usage (Rescorla & Alley, 2001; Rescorla, Ratner, Jusczyk, & Jusczyk, 2005). The LDS consists of a checklist of 310 words commonly found in children's first vocabularies; the parent is required to indicate which of these their child currently uses spontaneously. In addition, the parent indicates whether or not the child combines two or more words, and documents examples of the three longest utterances typical of the child. This simple tool, suitable for ages 18 to 35 months, has been demonstrated to have strong psychometric properties, for example acceptable test-retest reliability, strong sensitivity and specificity and good predictive validity (Rescorla, 1989; Rescorla et al., 2005)

MacArthur-Bates Communication Development Inventories (CDI, Fenson et al., 1993). The Words and Sentences version of the CDI is a parent-completed tool designed to measure expressive vocabulary, morphological and syntactic development in children aged 16 to 30 months. Part I comprises a checklist of 680 words organised into semantic categories (the parent identifies those which their child currently produces), as well as questions about the child's use of various language forms. The second section assesses production of selected morphemes (e.g., regular plural '*s'*), irregular plural nouns and past tense verbs, and complexity of multi-word forms. The tool has been utilised extensively in research (e.g., Reilly et al., 2007) and has demonstrated excellent psychometric properties (Fenson et al., 1993). Percentile scores are reported for the CDI measures.

Procedure

Parents were provided with written and verbal information about the study and gave written consent for their child to participate. Ethics clearance was obtained from the Curtin University of Technology and South Metropolitan Area Health Service human research ethics committees. Parents who consented to participate were contacted by phone by the principal investigator and basic screening information was obtained to confirm eligibility. This included checking for family history of speech, language or literacy difficulties and administering the WILSTAAR screen.

Data were collected when the infants were 9, 12, 15, 18 and 24 months (within two weeks of reaching each age[1]). For face to face data collection sessions, infants were visited (by the investigator) in their homes with their primary caregiver present. Appointments were scheduled around the infant's sleep and feeding times to ensure maximum participation. At times, older siblings were present during data collection sessions. Face to face data collection sessions typically lasted approximately one hour. A summary of the measures used at each time-point are displayed in Table 24. Procedures specific to each time-point are described further below.

9 months. A short (approximately 15 minutes) 'warm up' was conducted, where the infant was familiarised with the investigator and the equipment, while the examiner spoke to the parent. During this time, the examiner clarified and checked responses on the Infant-Toddler Checklist, Caregiver Questionnaire and ASQ, which were typically completed by parents the day before or on the day of the session.

The microphone was then attached to the infant's bib or clothing. To obtain a vocalisation sample, parents were advised to interact with their baby as they normally would, using toys available and familiar to them, for a duration of approximately 20 minutes. During this time, the investigator avoided interacting with the infant, and only monitored the equipment. If the infant became upset or distressed, the parent was encouraged to attend to their baby's needs before recommencing the play session. Parents and infants typically played with blocks, books, toy vehicles or soft

[1] This target was not met on two occasions for the 24 month old data (questionnaires not completed by parents within the target timeframe)

toys. The representativeness of the vocalisation sample was established via parental report[2]. The REEL-3 was then administered, following the standard procedures. Following the data collection session, assessments were scored for later analysis.

12 months. The session again commenced with a short 'warm up' period, where the infant was re-familiarised with the investigator and equipment. The ASQ and CSBS caregiver questionnaire were discussed during this time. The video equipment was then set up, and the infant was settled into a high chair, with the parent and investigator positioned seated on either side (the investigator to the child's left). The microphone was attached to the infant's bib or clothing while the parent distracted him/her. The behaviour sample items were then presented as per the CSBS protocol. The behaviour sample was used as a context to record a vocalisation sample (the behaviour sample is not appropriate for use at the 9 month age). The REEL-3 was subsequently administered, following standard procedures.

15 months. For this timepoint, data were collected via post and telephone. The CSBS Caregiver Questionnaire was posted to the parents and scored on their return. The REEL-3 was administered over the phone with the primary caregiver. No vocalisation sample was obtained at this age.

18 months. The data collection session for 18 months chronological age was identical to that for the 12 month sample, with the CSBS Behaviour Sample and REEL-3 being administered, and Caregiver Questionnaire and ASQ discussed. In addition, the Language Development Survey was also included, to capture an estimate of emerging vocabulary.

24 months. Data for 24 months were collected via the CSBS Caregiver Questionnaire and MacArthur-Bates CDI. The LDS was not re-administered as the CDI obtained similar, but more extensive data on expressive vocabulary and emerging syntactic development for this age.

[2] Note that parents universally reported that the sample was 'typical' of their child's vocalisations, but that this did not necessarily equate to it being the 'best' the infant could do – parents reported that the amount of vocalisations produced changed over the day, and varied day to day. Only in instances where equipment failed or there were exceptional circumstances (e.g., an infant being unsettled the whole session) was another session arranged

Table 24

Summary of Tools Used at Each Data Collection Time-point

Assessment	Data Collection Time-point (age in months)				
	9	12	15	18	24
WILSTAAR screen	✓				
CSBS Infant-Toddler checklist	✓				
ASQ	✓	✓		✓	
Vocalisation sample	✓	✓		✓	
REEL-3	✓	✓	✓	✓	
CSBS Caregiver Questionnaire	✓	✓	✓	✓	✓
LDS				✓	
MCDI					✓

Note. WILSTAAR = Ward Infant Language Screening Test Assessment and Remediation, CSBS = Communication and Symbolic Behaviour Scales, ASQ = Ages and Stages Questionnaire, REEL-3 = Receptive-Expressive Emergent Language Test, 3rd Edition, LDS = Language Development Survey, MCDI = Macarthur-Bates Communicative Development Inventory

Identification of Infants with Early CAS Features

Standardised data were examined for communication 'status' at 2 years of age. Using criteria established in the research literature and also utilised in clinical practice, infants were identified as either having 'communication skills within normal limits for age' or 'communication skills not within normal limits for age'. Specifically, a CDI expressive vocabulary score and/or sentence complexity score of less than the 10th percentile indicates delayed/restricted expressive language development (Fenson et al., 1993). Descriptively, this manifests in a raw expressive vocabulary score of less than 50 words and/or the absence of two (or more) word combinations. In

addition, if any subtests of the CSBS Caregiver Questionnaire were below the recommended cut-off, communication skills were deemed to be restricted for age.

Provided one or more infant/s presented at 24 months with communication skills below that expected for age, data were planned to be investigated further for potential features consistent with increased risk of CAS. Features proposed by Davis and Velleman (2000) for the infant-toddler age group included a receptive-expressive gap, systematic gaps in phonetic repertoire, the absence of consonant-vowel babble, and the use of gestures and/or signs (Davis & Velleman, 2000). It is acknowledged that there may be considerable overlap in features of CAS and other severe speech-sound disorders at this age, but given the absence of longitudinal studies detailing the presentation of CAS over time (Zeigler & Maassen, 2004), identification of toddler/s with increased risk profiles is still of high importance. More detailed analyses of vocalisation samples were planned for such infant/s, in comparison to the typically developing infants.

Vocalisation Samples

Vocalisation samples for siblings of interest and the comparison infants were digitised and prepared for perceptual and acoustic analysis. Each sample was imported into PRAAT (sampling rate of 44100 Hz). The investigator listened to each sample and identified each infant 'utterance', using both the visual display (time-amplitude waveform; wideband spectrogram) and acoustic-perceptual cues. Following procedures described by Stark, Bernstein and Demorest (1993) an utterance was defined as a single vocalisation, or a series of vocalisations separated by all others by 2 seconds. Individual vocalisations within the utterances were then categorised according to the Stark Assessment of Early Vocal Development – Revised (SAEVD-R, Nathani, Ertmer, & Stark, 2006). Spectrographic displays were used to assist the categorisation, where needed. The vocalisation categories and descriptions in the complete coding system are presented in Table 25. Following suggestions by Nathani, Ertmer and Stark (2006), vocalisations were grouped into the three over-arching categories: pre-canonical vocalisations, canonical vocalisations, and advanced forms. This approach has previously been utilised in investigations of vocal development in cochlear implant wearers (Ertmer & Mellon, 2001; Ertmer et al., 2002).

Pre-canonical vocalisations. Vocalisations coded at this level were those that lacked the well-formedness typical of canonical syllables. That is, they lacked combined consonant and vowels produced with a rapid transition. Table 25 outlines the vocalisation types coded within this level, which includes those at the 'reflexive', 'control of phonation' and 'expansion' stages in the SAEVD-R. Examples include quasi-resonant nuclei, low-pitched grunt like sounds, and fully resonant nuclei.

Canonical vocalisations. The production of well-formed, adult-like syllables is the hallmark of this level. Vocalisations included at this level included: vocalisations perceived to comprise a clear consonant-vowel, either in isolation (CV), as a disyllable (CVCV), more than two syllables produced in the one vocalisation (canonical babbling, CB), or those types further outlined in Table 25.

Advanced forms. Syllables with more complex phonotactic structure were coded at this level (see Table 25). This included: single syllables with VC structure or containing a consonant cluster, diphthongs (characterised by formant transitions less than 200 msec), and jargon strings – vocalisations containing multiple syllables with varying intonation patterns.

Infant vocalisations that were inaudible or unable to be coded due to the presence of background noise (such as toys or the mother's voice) were discarded. Instances where the vocalisation could not be readily-coded were re-analysed by two additional investigators, and resolved via consensus opinion. Total vocalisations in each level were calculated, as well as proportions relative to the total number of vocalisations, and rate of vocalisation. A second speech pathologist experienced in infant vocalisations re-coded 10% of the vocalisations. Inter-rater reliability was found to be high, Cohen's kappa = 0.86, $p < .001$.

Table 25

Vocalisation Types at the Pre-canonical, Canonical and Advanced levels (Nathani et al., 2006)

Level	Vocalisation Types	Description/Examples
Pre-canonical	Vegetative Sounds	Burp, cough, sneeze
	Quasi-resonant nuclei (Q)	Faint, low pitched grunt-like sounds with muffled resonance. Characterised by lack of energy above 2000 Hz
	Fully-resonant nuclei (F)	Vowel-like sounds longer than Qs, with energy across wide range of frequencies
	Isolated closants or consonants	Raspberry, trill, click
	Chuckle	Brief chuckles or sustained laughter
	Isolated vowel (V)	Transcribable vowel; longer and more resonant that Qs and Fs
	Vowel glide (Vg)	Vocant with change in vowel quality (no audible gap or closure, but transitions greater than 200ms)
	Ingressive (IN)	Single long (>200ms) ingressive sound or series of short ingressive sounds
	Squeal (SQ)	High pitched, in isolation or as a series
	Marginal babbling (MB)	Series of closant and vocant segments or series of Vgs. Formant transitions >120ms

Continued over

Table 25 (continued)

Level	Vocalisation Types	Description/Examples
Canonical	Consonant-vowel (CV)	Consonant-vowel syllables
	Canonical babbling (CB)	Reduplicated or non-reduplicated
	Whispered (WH)	MB, CB or CV produced without voice
	Disyllables (CVCV)	Two adjacent CV syllables
	CV-C	Consonant-vowel following by isolated consonant (after a gap)
Advanced	Complex syllables	VC, CCV, CCVC
		Complex disyllables (VCV, VCVC)
		Multisyllabic strings
	Jargon	Series of syllables with at least two different Cs and Vs with changing stress and/or intonation pattern
	Diphthong	Formant transitions <200ms and total duration <500ms

Acoustic Analyses

Following coding of vocalisations, a number of acoustic measures were made on pre-linguistic canonical syllables. The production of canonical syllables has been shown to be an important predictor of later speech/language development (Oller, Eilers, Neal, & Schwartz, 1999). Canonical syllables also represent the first syllabic articulatory gestures containing a consonant (Oller, 2000) that may form part of the developing protosyllabary. Not only is the presence and frequency of canonical syllables of great interest in regard to CAS, but it was hypothesised that acoustic differences reflecting a core motoric impairment in the production of these usually

'well formed' syllables may theoretically be present in infants with significant CAS risk factors. Acoustic measures therefore investigated are outlined below. Canonical vocalisations that were produced in the presence of invasive background noise (e.g., the mother's voice or toys) or that were whispered were unable to be analysed acoustically.

Duration. The duration of each canonical syllable was measured in milliseconds from the onset of the infant vocalisation to the offset. The vocalic portion of the vocalisation, indicated by the commencement of the formant structure, was then identified for subsequent measurements. Typically, canonical syllable duration in infants ranges from between 100 to 500ms in length (Rvachew, Creighton, Feldman, & Sauve, 2002).

Fundamental frequency (F_o). Measurement of the mean and standard deviation of F_o was obtained from PRAAT. For each canonical syllable, after selecting the vocalic portion, PRAAT was used to obtain automatic measurement of F_o. Default settings were used, except that minimum F_o was adjusted to 150Hz to suit the higher fundamental frequency of infants, as recommended by the PRAAT manual (Boersma & Weeink, 2002).

F_1 and F_2. Formants 1 and 2 were measured by identifying the mid-point of the steady state vowel and calculating the average across three consecutive formant frequency estimates. Consistent with recommendations for analysis of infant vocalisations (Boersma & Weeink, 2002), adjustments were made to the maximum formant parameter (corresponding to the 5^{th} formant) to ensure formant estimates overlaid on the spectrogram tracked the first and second formant band accurately. Spectral slices using FFT analysis were used to assist the location of these formants.

Fifteen percent of the syllables were reanalysed to ascertain intra-rater reliability. Mean absolute difference values were 3.2ms and 6.2ms for total duration and vocalic duration, respectively, 2.1Hz, 9.6Hz, and 74.4 Hz for mean F_o, F1 and F2, respectively. These values were comparable with the values reported in the literature (Bahr, 2005).

Dissociation Between Conceptualiser and Speech Motor Control

Finally, in order to examine the proposed independence of conceptual and speech motor areas of development in infancy (Levelt et al., 1999), the procedures described by Crawford and colleagues (Crawford, Garthwaite, & Gray, 2003) for testing for a dissociation were applied. The social composite of the CSBS caregiver questionnaire was used as a measure of conceptualiser development. This composite reflects development in the area of communicative intent and conceptual development (including emotion and eye-gaze, and gesture use). The sounds subtest from the same tool was used as a standardised measure of speech motor output, reflecting the infant's production of syllabic articulatory gestures.

Results

Overview

Longitudinal data from the standardised assessments will first be presented for all 16 infants. Both group and individual data will be described. Overall group differences between the siblings and comparison groups may be informative relative to the notion of aggregation of a broader phenotype in CAS (Lewis, Freebairn, Hansen, Taylor et al., 2004). Based on their presentation at 24 months of age and the pattern of characteristics over time, more detailed vocalisation and acoustic data are reported on two infants in the siblings group, and compared to data for the eight comparison infants.

Data were screened for adherence to assumptions underlying the relevant parametric analyses. Violations to the homogeneity of variance assumption were observed for a number of subtests on the CSBS caregiver questionnaire. The timepoint at which this was observed varied for each subtest, but typically was only observed at one session for each (e.g., gesture use at 18 months). Although group numbers were not large, they were equal, and because ANOVA is usually robust to moderate violations in assumptions when group sizes are equal, analysis using this method proceeded (Tabachnick & Fidell, 2001).

Group Comparisons Over Time

Mean standard scores for the siblings and comparison group on the ASQ, CSBS Caregiver Questionnaire, and REEL-3 subtests at each timepoint are displayed in Tables 26, 27 and 28, respectively. Instances of missing data (6.25% of the total data set) were replaced with the participant's mean for the relevant subtest in order to maintain equal group sizes. Two way mixed ANOVAs (between groups variable is group and the repeated measures variable is timepoint) were conducted to examine general differences between the groups over time. The dependent variable is subtest scores for each of the standardised assessments. Effect size is indicated by partial eta squared (η^2_{partial}) values.

Ages and stages questionnaire (ASQ). Mean scores on each subscale for the siblings and comparison groups are displayed in Table 26. The siblings as a group displayed significantly lower scores than the comparison group on the Communication, Fine Motor, and Problem Solving areas, $F(1, 14) = 11.1$, $p = .01$, $\eta^2_{\text{partial}} = .44$; $F(1, 14) = 16.4$, $p < .01$, $\eta^2_{\text{partial}} = .54$; and $F(1, 14) = 5.6$, $p = .03$, $\eta^2_{\text{partial}} = .29$, respectively. The groups were not significantly different on the Gross Motor and Personal-Social areas; $F(1, 14) = 0.5$, $p = .49$, and $F(1, 14) = 4.5$, $p = .052$, $\eta^2_{\text{partial}} = .24$, respectively. No timepoint by group interaction effect was present for any of the ASQ subtests (see Appendix G for details). A main effect of timepoint was found for the Gross Motor, $F(2, 28) = 5.4$, $p = .01$, $\eta^2_{\text{partial}} = .28$, and Personal Social, $F(2, 28) = 3.5$, $p = .04$, $\eta^2_{\text{partial}} = .20$, areas. Posthoc contrasts, using the Bonferroni-adjusted p value, showed a difference in Gross Motor scores between the 12 and 18 months' marginal means, $p = .03$ (higher scores at 18 months). For the Personal Social scores, pair-wise posthoc contrasts were not statistically significant ($p = .08$ for the descriptively higher 9 month compared to 12 month contrast).

Table 26

Mean ASQ scores (standard deviations in parentheses) for the Comparison and Siblings Group at 9, 12 and 18 months

ASQ area	Timepoint (months)		
	9	12	18
Communication			
Comparison	55.6 (9.3)	45.6 (10.5)	51.9 (8.8)
Siblings	45.0 (6.8)	40.0 (7.6)	38.1 (12.5)
Gross Motor			
Comparison	55.6 (7.3)	40.6 (23.1)	53.8 (8.8)
Siblings	50.0 (11.0)	50.6 (10.2)	59.4 (1.8)
Fine Motor			
Comparison	56.9 (4.6)	58.8 (2.3)	59.4 (1.8)
Siblings	52.8 (7.3)	48.8 (5.2)	53.1 (8.0)
Problem Solving			
Comparison	56.9 (3.7)	53.8 (8.3)	51.3 (7.9)
Siblings	49.4 (11.2)	48.1 (10.3)	44.4 (6.8)
Personal-Social			
Comparison	56.9 (3.7)	51.3 (5.9)	51.9 (5.9)
Siblings	50.0 (12.2)	43.8 (9.5)	46.2 (7.4)

Note. ASQ scores represent values out of a possible total of 60.

CSBS Caregiver Questionnaire. Examination of the CSBS Caregiver questionnaire subtests (displayed in Table 27) revealed a significant main effect of group on both the Sounds, $F(1, 14) = 19.6$, $p < .01$, $\eta^2_{partial} = .58$, and the Object Use, $F(1, 14) = 26.3$, $p < .01$, $\eta^2_{partial} = .65$, subtests, with the siblings scoring lower on both. The groups did not differ significantly on the remaining subtests of the CSBS: Emotion

and Eye Gaze, $F(1, 14) = 0.17$, $p = .69$; Communication, $F(1, 14) < 0.01$, $p = .95$; Gesture, $F(1, 14) = 2.44$, $p = .14$, $\eta^2_{partial} = .15$; Words, $F(1, 14) = 4.29$, $p = .06$, $\eta^2_{partial} = .24$; and Understanding, $F(1, 14) = 0.51$, $p = .49$. Again, there were no significant group by timepoint interaction effects for any of the CSBS measures (listed in Appendix G). Statistically significant main effects of timepoint were observed for five of the seven subtests (expression and eye gaze, communication, gesture, sound, word and understanding). For these, posthoc analyses revealed the significant differences to lie between the 9 and 24 month (expression and eye gaze); 9 and 18, 9 and 24, 12 and 18, and 12 and 24 month (gesture); 9 and 24, 12 and 24, and 15 and 24 month (sounds); and 15 and 24 month (word) timepoints (details in Appendix H). A statistically significant linear trend was observed for the expression and eye gaze, $F(1, 14) = 8.76$, $p = .01$, communication, $F(1, 14) = 5.21$, $p = .04$, gesture, $F(1, 14) = 57.18$, $p > .01$, sounds, $F(1, 14) = 25.24$, $p > .01$, words, $F(1, 14) = 9.82$, $p = .01$, and understanding, $F(1, 14) = 19.69$, $p > .01$, subtests.

Receptive-Expressive Emergent Language (REEL-3). Mean receptive and expressive language ability scores for the two groups are displayed in Table 28. A statistically significant group difference on receptive ability scores (with lower scores for the siblings) was indicated, $F(1, 14) = 7.3$, $p = .02$, $\eta^2_{partial} = .34$. Expressive scores were also significantly lower for the siblings as a group, $F(1, 14) = 17.1$, $p < .01$, $\eta^2_{partial} = .55$. Both quotients varied significantly over time; $F(3, 42) = 6.1$, $p < .01$, $\eta^2_{partial} = .30$ (receptive), and $F(3, 42) = 5.2$, $p < .01$, $\eta^2_{partial} = .27$ (expressive), with linear trends observed for both language areas (receptive: $F(1, 14) = 14.53$, $p = .002$; expressive: $F(1, 14) = 6.58$, $p = .02$). Posthoc analyses revealed that the receptive ability score varied significantly between 12 and 18 months (scores higher at 18 months), $p = .01$. Pair-wise contrasts of expressive ability score did not reach significance with post hoc analysis. Although numerically the increase in receptive scores at 18 months was less for the siblings, there was no significant group by timepoint interaction for either receptive or expressive language ability scores, $F(3, 42) = 1.6$, $p = .21$, $\eta^2_{partial} = .10$, and $F(3, 42) = 0.1$, $p = .96$, respectively.

Table 27

Mean CSBS Scores (standard deviations in parentheses) for the Comparison and Siblings Group at 9, 12. 15, 18 and 24 months

Subtest	Timepoint (months)				
	9	12	15	18	24
Expression & eye gaze					
Comparison	9.8 (3.7)	13.8 (2.6)	13.6 (2.0)	12.6 (2.6)	13.4 (2.0)
Siblings	10.4 (3.7)	12.3 (2.7)	12.5 (2.9)	12.6 (2.6)	13.5 (2.8)
Communication					
Comparison	12.0 (2.5)	12.6 (2.5)	12.6 (3.3)	13.5 (2.7)	13.8 (3.7)
Siblings	12.4 (3.5)	10.5 (3.7)	13.6 (2.3)	13.9 (3.6)	14.5 (3.9)
Gesture					
Comparison	11.9 (2.2)	10.8 (1.9)	13.9 (4.2)	17.0 (0)	16.1 (2.2)
Siblings	9.9 (3.8)	9.9 (2.9)	12.5 (4.2)	13.1 (4.7)	16.0 (2.8)
Sounds					
Comparison	10.9 (1.5)	11.4 (1.8)	11.2 (1.2)	13.4 (2.5)	15.7 (2.4)
Siblings	8.4 (3.1)	8.5 (2.4)	9.0 (2.4)	8.6 (1.7)	12.0 (4.5)
Words					
Comparison	10.8 (2.1)	11.1 (2.6)	9.8 (2.7)	12.3 (1.8)	15.2 (2.7)
Siblings	10.0 (1.2)	9.5 (2.1)	8.8 (3.5)	9.6 (1.9)	11.7 (4.9)
Understanding					
Comparison	9.5 (2.9)	11.4 (1.4)	9.9 (1.4)	12.6 (2.2)	14.6 (3.4)
Siblings	11.5 (2.1)	11.8 (2.3)	11.1 (2.6)	11.9 (2.9)	15.0 (3.2)
Object Use					
Comparison	10.9 (1.8)	11.8 (2.3)	11.4 (1.1)	11.6 (2.1)	13.8 (2.7)
Siblings	8.4 (3.5)	8.5 (3.6)	9.6 (2.9)	10.4 (2.7)	9.9 (1.3)

Table 28

Mean REEL-3 Receptive and Expressive Language Ability Scores (standard deviations in parentheses) for the Comparison and Siblings Groups at 9, 12, 15 and 18 months

Subtest	Timepoint (months)							
	9		12		15		18	
Receptive								
Comparison	96.8	(8.0)	97.8	(11.0)	103.6	(9.7)	109.8	(6.7)
Siblings	91.6	(9.6)	91.2	(5.6)	96.3	(7.7)	95.3	(8.2)
Expressive								
Comparison	95.0	(3.3)	93.5	(9.2)	99.6	(9.7)	105.9	(11.7)
Siblings	77.0	(10.1)	77.3	(10.0)	85.5	(14.0)	91.0	(20.8)

Data specific to each of the timepoints are discussed further below, with further detail on individual performance.

9 months

WILSTAAR screen. All eight infant siblings failed the WILSTAAR screen, with all failing the expressive component only. In contrast, all of the comparison group infants passed this screen.

Ages and Stages Questionnaire. Scores in each developmental area for the infants in both groups are presented in Table 29. Inspection of individual scores revealed that all individual comparison infants scored within the typically-developing range for each sub-area. In contrast, one infant sibling (SIB2) scored below the recommended cut-off on the communication area, and another (SIB3) scored below the cut-off on the problem solving and personal-social areas.

Table 29

ASQ Area Scores and REEL-3 Receptive and Expressive Ability Scores for the Comparison (C) and Siblings (SIB) at 9 months

	ASQ					REEL-3	
	Comm	GM	FM	Prob	Pers-Soc	Rec	Exp
C1	60	55	60	55	55	95	94
C2	55	60	60	60	60	88	94
C3	60	60	55	60	60	85	89
C4	60	50	60	60	60	92	98
C5	40	60	50	55	55	105	93
C6	55	60	60	50	50	106	95
C7	60	60	50	55	60	103	98
C8	55	40	60	60	55	100	99
SIB1	55	60	60	55	60	**82**	90
SIB2	**35**	60	50	55	50	95	**65**
SIB3	40	40	40	**25**	**25**	**75**	**77**
SIB4	45	40	60	60	50	85	**75**
SIB5[a]	-	-	-	-	-	-	-
SIB6	55	35	60	55	60	100	**83**
SIB7	50	60	55	55	60	98	**83**
SIB8	50	45	50	45	55	100	**83**

Note. Ages and stages questionnaire (ASQ) scores below the recommended cut-off are shown in bold. Rec = Receptive; Exp = Expressive; Comm = Communication; GM = Gross Motor; FM = Fine Motor; Prob = Problem Solving; Pers-Soc = Personal-Social.

[a] SIB 5 did not commence the study until 12 months.

REEL-3. Receptive and expressive ability scores are also shown in Table 29. All comparison infants achieved receptive and expressive ability scores within the normal range at 9 months. In contrast, six siblings (SIBS 2, 3, 4, 6, 7 and 8) had expressive language ability scores lower than the range considered to be typical (defined as >1 SD from the mean). Two siblings displayed receptive scores below the typically-developing range (SIBS 1 and 3).

CSBS Infant-Toddler Checklist. Individual standard scores for the three composite areas on the Infant-Toddler Checklist are presented in Table 30. The recommended cut-off for 'concern' on this tool is a standard score below 7 (1.5 SDs below the mean). The siblings, as a group, scored significantly lower than the comparison group on the speech composite, $t(13) = 4.2$, $p < .01$. The groups did not differ significantly on the social and symbolic composites, $t(13) = 0.43$, $p = .68$, $t(13) = 0.19$, $p = .85$, respectively. Inspection of the individual scores shows that three siblings (SIBS 2, 3 and 4) scored below the recommended cut-off on the Speech composite. One comparison infant (C2) scored below the cut-off on the social composite at this age.

CSBS Caregiver Questionnaire. Figure 3 displays individual data for the CSBS Caregiver Questionnaire at 9 months for the comparison and siblings groups. Standard scores for the individual subtests (referred to as 'clusters'), as well as the three composite areas (social, speech and symbolic) are shown. Composite scores, which are derived from the individual subtests, are shown at the right end of the graphs. Line graphs (allowing illustration of which children, and in which areas, fell below cut-off scores) are used to assist the reader in following an individual child as well as to visually compare the two groups. Instances where scores were below the recommended cut-off for this tool are illustrated by their falling on or below the bold horizontal line. Inspection of individual data revealed that none of the comparison infants scored below the normal range on composite scores, but that one (C2) was below the expected range on the emotion and eye gaze cluster. Of the siblings, four (SIBS 1, 2, 3 and 4) scored below the cut-off on one or more of the clusters, with three of these (SIBS 2, 3 and 4) also scoring below on one or more of the composites.

Table 30

CSBS Infant-Toddler Checklist Standard Scores for the Comparison (C) and Siblings (SIB) Groups at 9 months

	Social Composite	Speech Composite	Symbolic Composite
C1	11	8	7
C2	**6**	11	10
C3	13	11	12
C4	13	14	13
C5	14	13	13
C6	9	9	12
C7	13	15	13
C8	11	9	12
M	11.3	11.3	11.5
SD	2.7	2.6	2.1
SIB1	11	8	12
SIB2	16	**5**	13
SIB3	7	**4**	12
SIB4	7	**6**	7
SIB6	11	8	12
SIB7	14	8	12
SIB8	8	7	14
M	10.6	6.6	11.7
SD	3.5	1.6	2.2

Note. Standard scores of 6 and below (shown in bold) are considered in the 'concern' range on this tool.

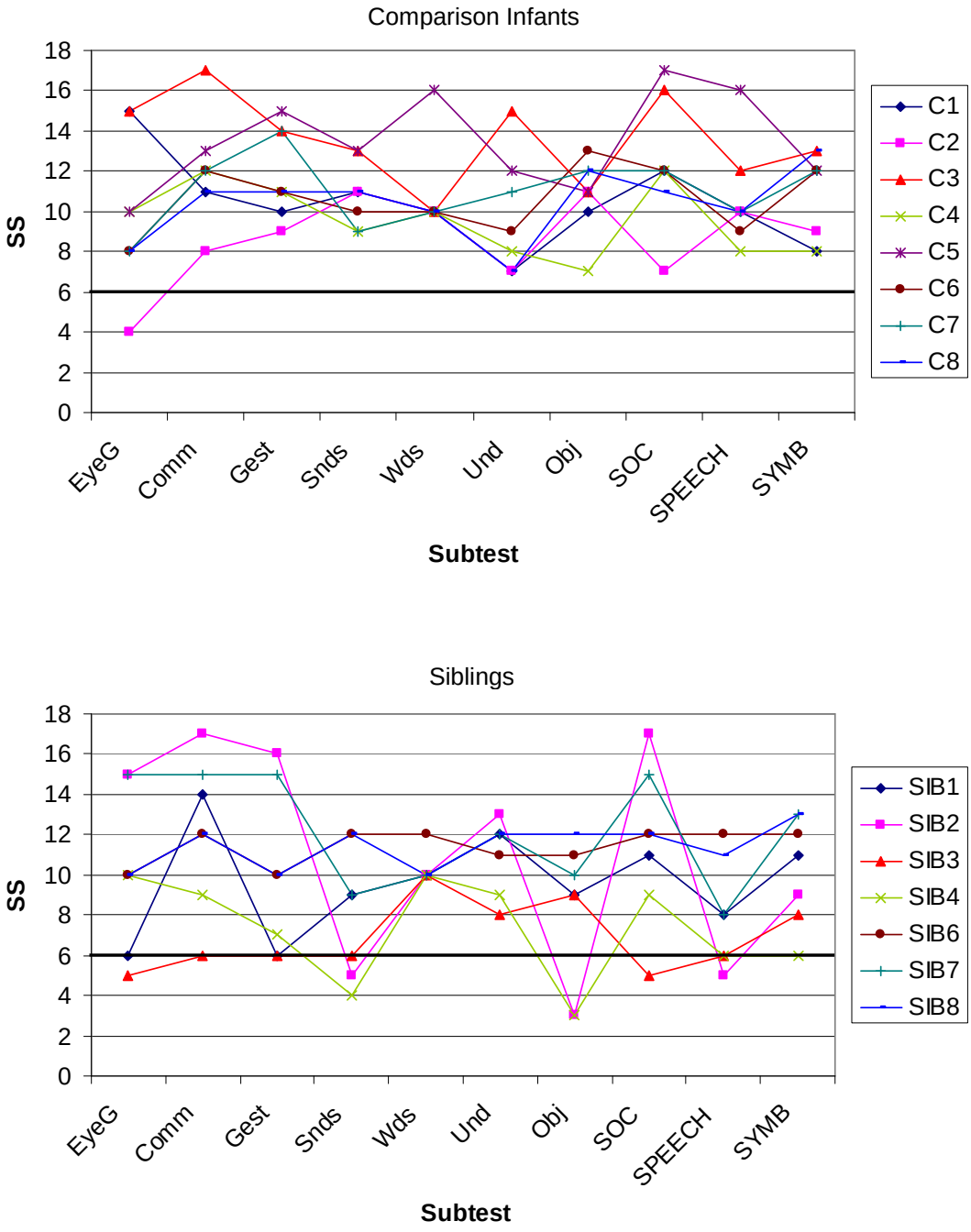


Figure 3. CSBS Caregiver Questionnaire standard scores for the comparison infants (top) and siblings (bottom) at 9 months (Note that standard scores of 6 and below, indicated by falling on or below the dark line, are considered in the 'concern' range for this tool. EyeG = Expression and Eye Gaze cluster; Comm = Communication cluster; Gest = Gesture cluster; Snds = Sounds cluster; Wds = Words cluster; Und = Understanding cluster; Obj = Object Use cluster; SOC = Social composite; SPEECH = Speech composite; SYMB = Symbolic composite).

12 months

Ages and Stages Questionnaire. Scores on each developmental area of the ASQ 12 month for the comparison and siblings groups are presented in Table 31. At 12 months, two of the comparison group infants (C5 and C8) were below the normal range on the Gross Motor subtest of the ASQ, while none of the siblings scored below the cut-offs in any of the developmental areas.

Table 31

ASQ Scores for each Developmental Area, and Receptive and Expressive Ability Scores on the REEL-3, at 12 months

	ASQ					REEL-3	
	Comm	GM	FM	Prob	Pers-Soc	Rec	Exp
C1	45	50	60	60	50	98	95
C2	45	50	60	60	50	98	95
C3	20	30	60	50	45	73	85
C4	50	60	60	60	60	98	84
C5	50	**15**	60	60	60	108	110
C6	50	50	60	55	50	108	89
C7	50	60	55	-	45	103	104
C8	50	**0**	60	55	60	97	89
SIB1	40	60	55	40	55	95	73
SIB2	45	30	50	60	45	90	70
SIB3	45	45	50	35	35	82	75
SIB4	50	50	45	60	45	-	-
SIB5	30	60	40	50	35	98	60
SIB6	30	50	45	35	30	85	85
SIB7	35	60	50	55	50	97	92
SIB8	45	50	55	50	55	92	84

Note. ASQ scores below the recommended cut-off are shown in bold.

Rec = Receptive; Exp = Expressive; Comm = Communication; GM = Gross Motor; FM = Fine Motor; Prob = Problem Solving; Pers-Soc = Personal-Social. Rec = Receptive; Exp = Expressive.

REEL-3. Table 31 also displays receptive and expressive ability scores at 12 months on the REEL-3. One comparison infant (C4) scored below the expected range on the expressive component, and one (C3) was below on the receptive component. In contrast, five siblings had expressive scores below the average range (SIBS 1, 2, 3, 5 and 8), and one (SIB3) was also below the average range on receptive ability score.

CSBS Caregiver Questionnaire. Individual and group summary standard scores for the CSBS Caregiver Questionnaire subtests and composite areas are displayed in Figure 4. As can be seen in the figure, all comparison infants scored within the normal range on all subtests of the CSBS Caregiver Questionnaire. In contrast, four siblings displayed one or more scores below the expected range: SIB1 (Object Use cluster), SIB2 (Sounds cluster and Speech composite), SIB3 (Communication, Sounds and Object Use clusters, and Speech composite) and SIB5 (Gesture and Object Use clusters).

15 months

REEL-3. Table 32 shows receptive and expressive ability scores for individual infants on the REEL-3 at 15 months. Inspection of individual scores revealed that whilst none of the comparison group scored below the normal range on either area, three siblings (SIBS 2, 3 and 6) had expressive scores below the average range. Receptive scores were within normal limits.

CSBS Caregiver Questionnaire. Standard scores for the cluster and composite areas for the two groups at 15 months are displayed in Figure 5. Inspection of each participant's scores indicated scores below the cut-off for one of the comparison group infants (C2, on the Words cluster). In contrast, two siblings had scores below the cut-off in a composite score (SIBS 1 and 2, Speech composite) and one or more cluster scores – SIB1 on the Words cluster, and SIB2 on both Sounds and Words. Another two of the infant siblings scored below the cut-off for one individual cluster (see Figure 5; SIB3: Object Use; SIB6: Gesture).

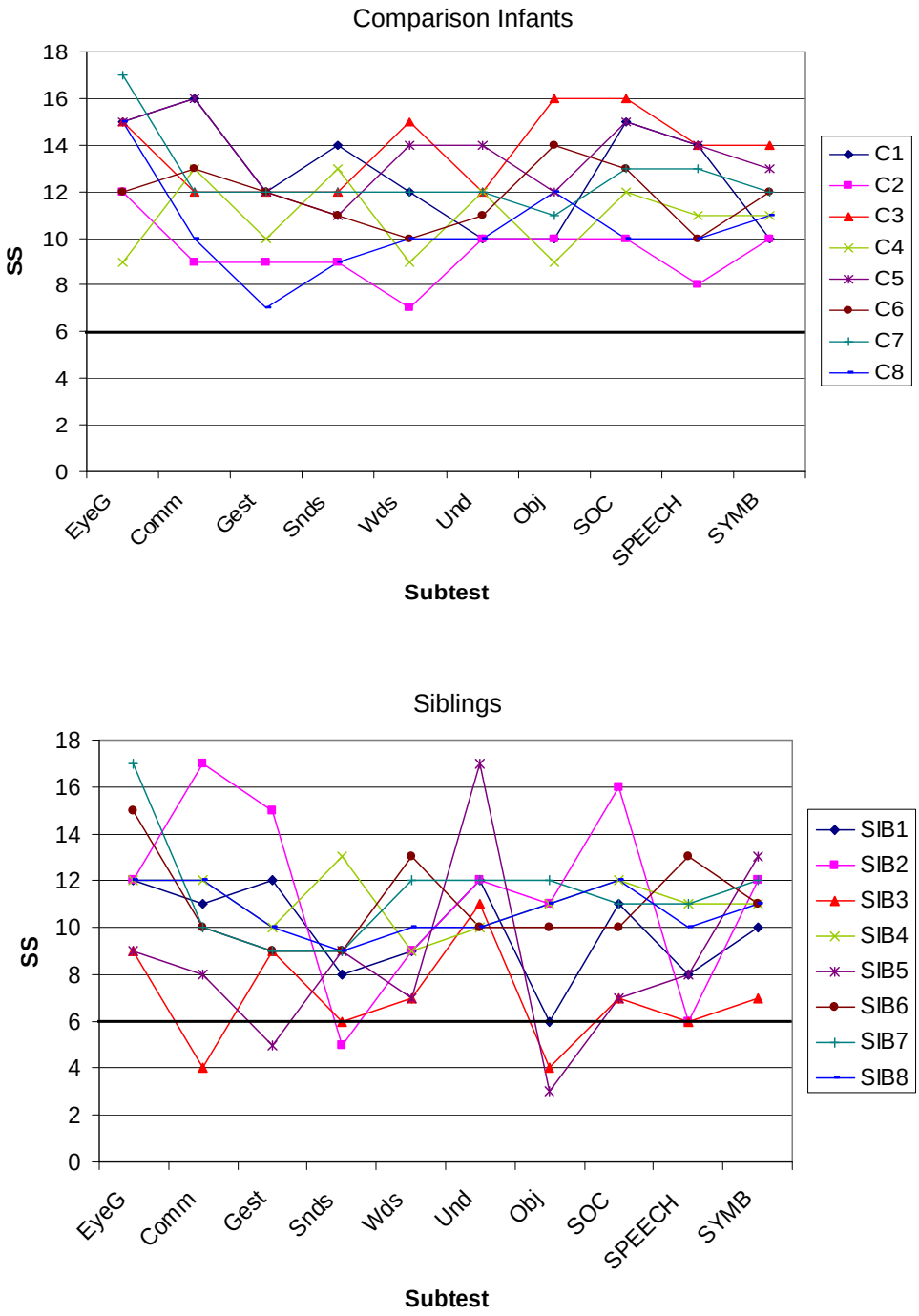


Figure 4. CSBS Caregiver Questionnaire standard scores for the comparison infants (top) and siblings (bottom) at 12 months (Note that standard scores of 6 and below, indicated by falling on or below the dark line, are considered in the 'concern' range for this tool. EyeG = Expression and Eye Gaze cluster; Comm = Communication cluster; Gest = Gesture cluster; Snds = Sounds cluster; Wds = Words cluster; Und = Understanding cluster; Obj = Object Use cluster; SOC = Social composite; SPEECH = Speech composite; SYMB = Symbolic composite).

Table 32

Receptive and Expressive Ability Scores on the REEL-3 at 15 months for the Comparison (C) and Siblings (SIB) Groups

	Receptive	Expressive
C1	98	94
C2	95	85
C3	97	88
C4	108	103
C5	125	113
C6	105	108
C7	-	-
C8	98	102
SIB1	-[a]	-
SIB2	97	60
SIB3	92	80
SIB4	95	85
SIB5	115	95
SIB6	92	82
SIB7	93	108
SIB8	93	93

[a] - = missing data: for reasons beyond the investigator's control, REEL-3 data was not able to be obtained at this target age.

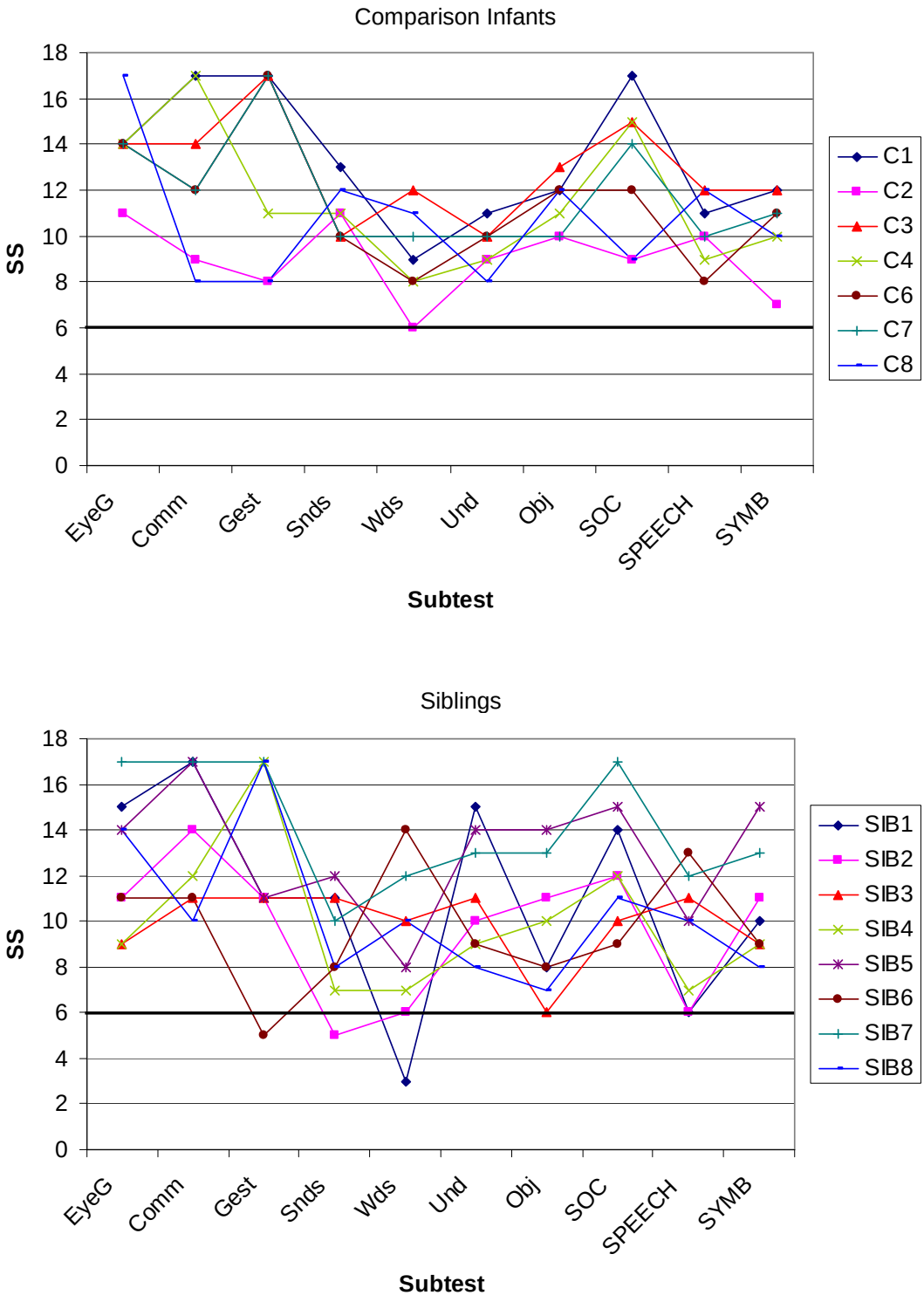


Figure 5. CSBS Caregiver Questionnaire standard scores for the comparison infants (top) and siblings (bottom) at 15 months (Note that standard scores of 6 and below, indicated by falling on or below the dark line, are considered in the 'concern' range for this tool. EyeG = Expression and Eye Gaze cluster; Comm = Communication cluster; Gest = Gesture cluster; Snds = Sounds cluster; Wds = Words cluster; Und = Understanding cluster; Obj = Object Use cluster; SOC = Social composite; SPEECH = Speech composite; SYMB = Symbolic composite).

18 months.

Ages and Stages Questionnaire. Individual scores for each ASQ developmental area are shown in Table 33. All comparison group infants' scores were within the normal range on all subtest areas of the ASQ at 18 months. Four siblings (SIBS 1, 2, 3 and 5) scored below the cut-off on the communication subtest; all other subtest scores were within the normal range.

Table 33

ASQ Scores for each Developmental Area, and Receptive and Expressive Ability Scores on the REEL-3 at 18 months

	ASQ					REEL-3		LDS
	Comm	GM	FM	Prob	Pers-Soc	Rec	Exp	
C1	55	60	60	60	60	115	100	50
C2	40	40	60	40	50	103	109	27
C3	55	60	60	60	55	103	80	9
C4	60	55	60	50	50	120	119	118
C5	60	55	60	55	55	110	113	37
C6	60	60	60	50	50	110	108	44
C7	40	60	55	55	55	102	109	47
C8	45	40	60	40	40	115	109	67
SIB1	**20**	55	55	40	55	103	79	10
SIB2	**30**	60	55	50	40	105	75	13
SIB3	**35**	60	50	55	40	87	83	11
SIB4	40	60	60	50	45	92	79	11
SIB5	**30**	60	55	40	45	98	138	13
SIB6	60	60	55	35	40	82	80	23
SIB7	40	60	60	40	60	102	95	24
SIB8	50	60	35	45	45	93	99	24

Note. ASQ scores below the recommended cut-off are shown in bold.

Rec = Receptive; Exp = Expressive; Comm = Communication; GM = Gross Motor; FM = Fine Motor; Prob = Problem Solving; Pers-Soc = Personal-Social. Rec = Receptive; Exp = Expressive. LDS = Language Development Survey and represents expressive vocabulary.

REEL-3. Table 33 also displays receptive and expressive language ability scores for the participants. One comparison infant scored below the expected range on the expressive component; all other comparison infants scored within the normal range on both the receptive and expressive subtests. In contrast, expressive language ability scores fell below the cut-off for five siblings (SIBS 1, 2, 3, 4, and 6), with SIB6 also below the normal range on receptive language ability score.

Language Development Survey (LDS, Rescorla, 1989). Individual and group expressive vocabulary scores (number of words) are also shown in Table 33. Mean number of words was significantly lower for the infant siblings (*M = 16.1, SD = 6.3)* than the comparison group (*M = 49.9, SD = 32.3), t*(14) = 2.89, *p* = .01. Although there was large variability in expressive vocabulary at this age, it is interesting to note that all siblings had less than 25 words, in contrast to the comparison group in which all but one toddler (C3) had vocabularies over 25 words (with most *well* over this number).

CSBS Caregiver Questionnaire. As displayed in Figure 6, all comparison group infants scored within the expected range on all areas of the CSBS Caregiver Questionnaire. Two siblings had cluster standard scores below the cut-off (SIB2 on the Sounds cluster; SIB6 on the Gesture and Object Use clusters).

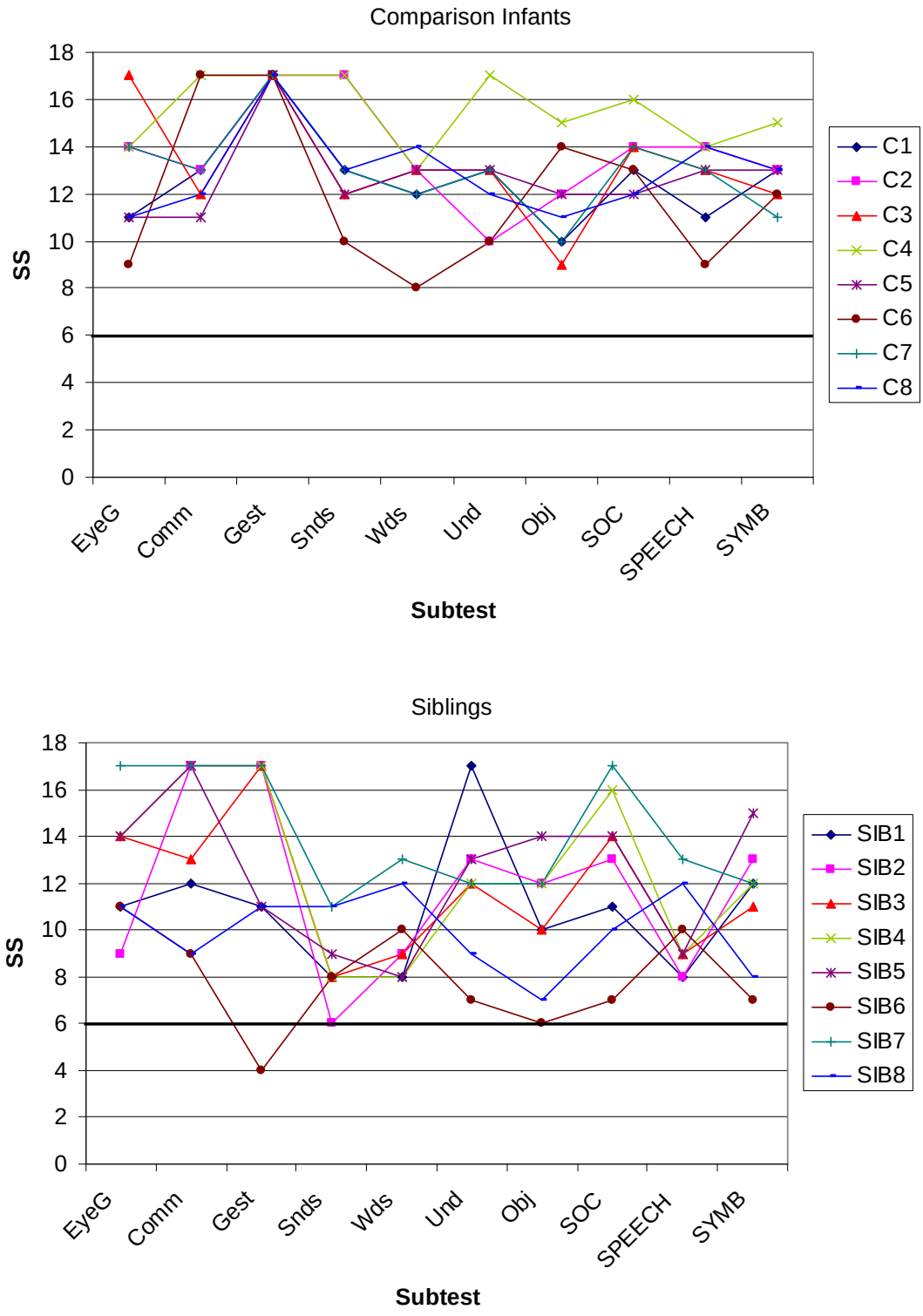


Figure 6. CSBS Caregiver Questionnaire standard scores for the comparison infants (top) and siblings (bottom) at 18 months (Note that standard scores of 6 and below, indicated by falling on or below the dark line, are considered in the 'concern' range for this tool. EyeG = Expression and Eye Gaze cluster; Comm = Communication cluster; Gest = Gesture cluster; Snds = Sounds cluster; Wds = Words cluster; Und = Understanding cluster; Obj = Object Use cluster; SOC = Social composite; SPEECH = Speech composite; SYMB = Symbolic composite).

24 months.

CSBS Caregiver Questionnaire. Figure 7 displays CSBS caregiver questionnaire standard scores for the siblings and comparison group infants, respectively. Data were not available for two of the comparison infants. Inspection of individual profiles revealed all comparison infants to have scored above the cut-offs on all subtests. Of the siblings, two infants scored below the recommended cut-offs on one or more subtests. SIB1 scored below on the words cluster, and the speech composite, but within the normal range on all other areas. SIB2 scored below on both the sounds *and* words clusters, and the speech composite. All other siblings' scores were within the normal range.

MacArthur-Bates Communicative Development Inventories (MCDI, Fenson et al., 1993). Table 34 displays raw vocabulary scores, corresponding percentiles and sentence complexity percentile scores for the sibling and comparison groups. Mean expressive vocabulary for the comparison infants was 450 words (range 367 to 548). In contrast, mean expressive vocabulary for the siblings was 191 words (range 17 to 428 words), significantly lower than the comparison group, $t(11) = 3.6$, $p = .01$. Similarly, sentence complexity was significantly lower for the siblings, $t(11) = 2.9$, $p = .02$. Descriptively, all comparison infants were using greater than 50 single words and were combining words. In contrast, two infant siblings had not reached this important milestone (SIBS 1 and 2)

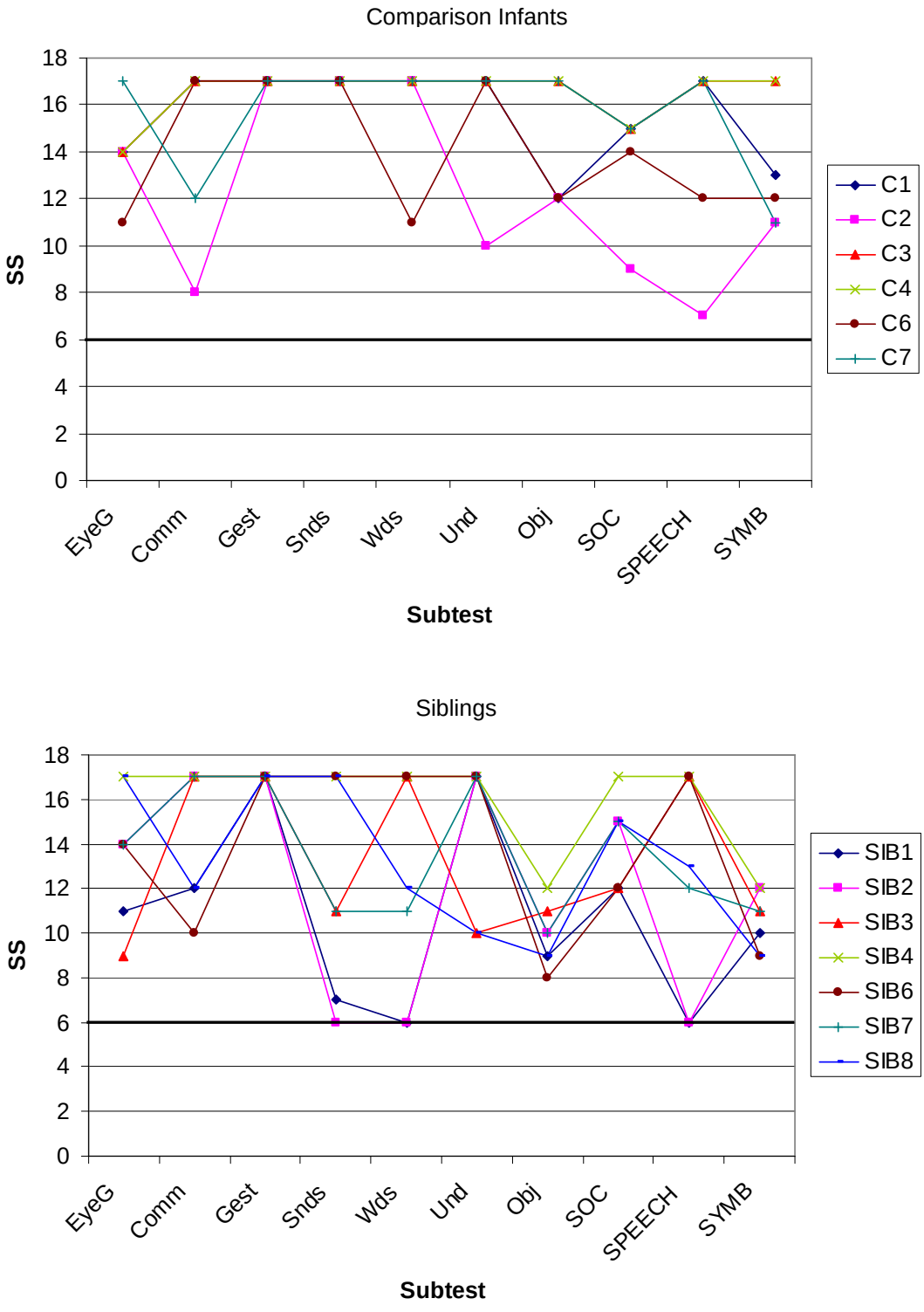


Figure 7. CSBS Caregiver Questionnaire standard scores for the comparison infants (top) and siblings (bottom) at 24 months (Note that standard scores of 6 and below, indicated by falling on or below the dark line, are considered in the 'concern' range for this tool. EyeG = Expression and Eye Gaze cluster; Comm = Communication cluster; Gest = Gesture cluster; Snds = Sounds cluster; Wds = Words cluster; Und = Understanding cluster; Obj = Object Use cluster; SOC = Social composite; SPEECH = Speech composite; SYMB = Symbolic composite).

Table 34

MCDI Raw Vocabulary Scores, Vocabulary Percentile and Sentence Complexity Percentile Scores for the Comparison (C) and Siblings (SIB) Group Infants at 24 months

	Raw Vocabulary Score	Vocab %ile	Sent Complexity %ile
C1	548	90	95
C2	376	70	70
C3	553	93	80
C4	457	83	80
C5	-	-	-
C6	392	60	55
C7	367	55	68
C8	-	-	-
M	*450*	*75*	*75*
SD	*85*	*16*	*14*
SIB1	17	<5	<10
SIB2	32	<5	<10
SIB3	189	30	25
SIB4	273	26	25
SIB5	-	-	-
SIB6	428	60	65
SIB7	83	9	20
SIB8	316	61	92
M	*191*	*28*	*35*
SD	*156*	*24*	*31*

Note. "-" = missing data

Identification of Siblings at 'Increased Risk'

At 24 months, the communication skills of two siblings were not developing appropriately for their age, based on the CDI and CSBS results. Expressive vocabulary scores were less than the 5th percentile for both, and were well below the recognised 50 single word and use of two-word combinations criteria used in research and clinical settings alike (Reilly et al., 2007; Rescorla, 1989; Rescorla et al., 2005) – 17 words for SIB1 and 32 for SIB2, with neither using any two-word combinations. Both were also below the recommended cut-off on the CSBS caregiver questionnaire Speech composite. SIB1 scored below the expected range on the Words cluster, but within the normal range on all other cluster and composite scores. In contrast, scores for SIB2 fell below the cut-off for both the Sounds and Words clusters.

Whilst profiles varied over the timeframe studied, all other siblings were within the normal range on all assessments by 2 years of age. Table 35 compares the areas that were below the normal range at each age interval for SIBS 1 and 2. As can be seen from the table, the two siblings, although both presenting with restricted expressive language development at 2 years of age, present with varied profiles longitudinally from 9 to 24 months. In particular, SIB2 consistently scored below the normal range on the CSBS Sounds cluster, a measure of the presence, type and frequency of syllable production. In contrast, SIB1's performance on this cluster was within the normal range at each age sampled.

Differences between the profiles of the two siblings are most evident at 9 months, where SIB2 shows strengths in expression and eye-gaze, gesture, and receptive language, but deficits in sounds and object use. In contrast, SIB1 presented with strengths in sounds, object use and the measure of expressive language at this age, in the presence of deficits in expression and eye-gaze, gesture and receptive language.

Table 35

Comparison of SIBS 1 and 2 on Communication Assessments at each age

Age (months)	Tool	Subtest	SIB1	SIB2
9	ASQ	Communication	WNL	↓
	CSBS ITC	Speech composite	WNL	↓
	CSBS CQ	Expression-Eye Gaze cluster	↓	WNL
		Gesture cluster	↓	WNL
		Sounds cluster	WNL	↓
		Object Use cluster	WNL	↓
		Speech composite	WNL	↓
	REEL-3	Receptive language	↓	WNL
		Expressive language	WNL	↓
12	CSBS CQ	Sounds cluster	WNL	↓
		Speech composite	WNL	↓
	REEL-3	Expressive language	↓	↓
15	CSBS CQ	Sounds cluster	WNL	↓
		Words cluster	↓	↓
		Speech composite	↓	↓
	REEL-3	Expressive language	-	↓
18	ASQ	Communication	↓	↓
	CSBS CQ	Sounds cluster	WNL	↓
	REEL-3	Expressive language	↓	↓
24	CSBS CQ	Sounds cluster	WNL	↓
		Words cluster	↓	↓
		Speech composite	↓	↓
	MCDI	Expressive vocabulary	↓	↓
		Sentence complexity	↓	↓

Note. CSBS ITC = CSBS Infant-Toddler Checklist; CSBS CQ = CSBS Caregiver Questionnaire; MCDI = MacAruthur Communicative Development Inventories. WNL = Within normal limits, "↓" indicates a score falling below the accepted cut-off for the tool, - = missing data

Table 36 illustrates the presence of CAS-related features described by Davis and Velleman (2000) for the infant-toddler age for the two siblings. Of the features that can be assessed via the assessment tools described, SIB2 shows the presence of all of these features. Sibling 2 thus presented with a pattern suggestive of increased risk of CAS. As it is not possible to diagnose CAS at this young age, he can only be considered at increased risk. The mother of this sibling described his early development and current presentation as being very similar to the older sibling with CAS, but very different to another older brother who did not have CAS. In contrast, SIB1, whilst presenting with delayed language development at 24 months, did not present with any of the CAS-related features. More detailed analysis of vocalisations was therefore undertaken on these two cases as well as those of the larger comparison group sample.

Table 36

Presence of CAS-related Characteristics (Davis & Velleman, 2000) for Siblings 1 and 2

Feature	Measure	SIB 1	SIB2
Gaps in phonetic repertoire	CSBS Sounds subtest	✗	✓
Lack of consonant-vowel babble	CSBS Sounds subtest	✗	✓
Developed use of gestures/signs	CSBS Gesture subtest & parent report	✗	✓
Late motor milestones	CSBS Object use subtest	✗	✓

Note. ✓ = feature present ✗ = feature not present

Vocalisation Data

Vocalisation samples collected at 9 months were analysed for SIB1 and SIB2, and the eight comparison infants. A total of 1220 vocalisations were coded by the investigator. Table 37 displays the total number of vocalisations and rate of vocalisation at each age. Statistical comparisons were made using the modified t-test

procedure described by Crawford and colleagues (Crawford, Garthwaite, Howell, & Gray, 2004), appropriate for use in comparing single cases to small comparison groups.

Table 37

Total Number of Vocalisations and Rate of Vocalisations at 9 months

	#[a] vocalisations	Rate (vocalisation/minute)
C1	84	2.9
C2	88	2.3
C3	145	6.6
C4	165	6.3
C5	266	11.5
C6	174	7.5
C7	83	2.4
C8	145	3.9
M	*143.8*	*5.4*
SD	*61.7*	*3.2*
SIB1	14	0.9
SIB2	56	2.8

[a] number of vocalisations

Total number and rate of vocalisations. The comparison infants produced an average of 144 vocalisations during the 20-30 minute vocalisation sample, or 5.4 per minute. There was considerable variation, with vocalisation rates ranging from 2.3 to 11.5 vocalisations per minute. Although SIB1's rate was descriptively lower (0.9 vocalisations per minute) than the range seen in the comparison group, it was not statistically lower, $t(7) = 1.4$, $p = .11$. SIB2's rate of vocalisations (2.8) was not different to the comparison infants', and fell within the range observed for the comparison group.

Type of vocalisations. Table 38 displays the breakdown of vocalisation types (Nathani et al., 2006) for the infants at 9 months. As expected at this age, the majority of vocalisations were pre-canonical in the typically developing infants. However, all had entered the 'canonical stage', producing a range of canonical syllables and even some advanced forms. Approximately 74% of vocalisations were pre-canonical, 18% canonical and 8% advanced forms, with the proportion of canonical vocalisations ranging from 6 to 28% in the comparison group. In contrast, 100% of both SIB1 and SIB2's vocalisations were pre-canonical, significantly greater than the comparison group, $t(7) = 2.4$, $p = .02$ (the proportion of canonical vocalisations was also significantly lower, $t(7) = 2.2$, $p = .03$).

Table 38

Breakdown of Vocalisation Types (percentages shown in parentheses) used by the Infants at 9 months

	Pre-Canonical	Canonical	Advanced
C1	75 (89%)	5 (6%)	4 (5%)
C2	55 (62%)	19 (21%)	15 (17%)
C3	121 (83%)	18 (12%)	6 (4%)
C4	126 (76%)	36 (22%)	3 (2%)
C5	173 (65%)	56 (21%)	37 (14%)
C6	116 (67%)	48 (28%)	10 (6%)
C7	67 (81%)	9 (11%)	7 (8%)
C8	115 (79%)	19 (13%)	11 (8%)
M	106 (75%)	26 (17%)	12 (8%)
SD	38.5	18.4	11.0
SIB1	14 (100%)	0 (0%)	0 (0%)
SIB2	56 (100%)	0 (0%)	0 (0%)

Acoustic Analyses

Detailed acoustic analyses were conducted on pre-linguistic canonical syllables. As no canonical syllables were produced by SIBs 1 and 2 at 9 months, their 12 and 18 month vocalisation samples were also coded, and canonical syllables identified for acoustic analysis. The proportion of each vocalisation type are shown in Table 39. Even at 12 months, the proportion of canonical vocalisations for SIB2 (less than 2%) is significantly lower than the comparison group infants' at 9 months of age, $t(7) = 2.0$, $p = .045$. There was no significant difference evident for the same comparison for SIB1, however, $t(7) = 0.41$, $p = .08$. Comparing these data (i.e., from 12 months, to that of the comparison infants who were producing canonical syllables at 9 months) presented a potential confound whereby biological/physical changes in the oral cavity size that would impact on some acoustic measures. However, in order to compare similarly prelinguistic canonical syllables, this approach was necessary.

Table 39

Number of Pre-canonical, Canonical and Advanced Vocalisations used by Siblings 1 and 2 at 12 and 18 months of age (percentages are shown in parentheses)

	Pre-Canonical	Canonical	Advanced
12 months			
SIB1	84 (84%)	15 (15%)	1 (1%)
SIB2	180 (98%)	3 (1.6%)	1 (0.5%)
18 months			
SIB1	67 (73%)	24 (26%)	1 (1%)
SIB2	258 (95.5%)	9 (0.3%)	3 (0.1%)

Table 40 displays the range of canonical syllabic articulatory gestures produced by the typically developing infants and SIBs 1 and 2. The transcriptions are presented as a context for interpreting the acoustic analyses (to follow), and show the range of syllabic gestures explored by the infants studied. As shown in the table, there was individual variation in the range of syllables produced by the typically developing infants. The most common consonants included bilabial, alveolar and velar stops, and nasals. Most of the comparison infants produced velars and/or fricatives, also. The vowels produced by the infants showed individual variation, with most comparison infants producing vowels with varying tongue position (cf. C3, however). SIB1 and SIB2's initial syllabic articulatory gestures also consisted of stops and nasals, with no velars or fricatives evident. SIB2 produced two types of vowels in the canonical syllables – the centralised schwa, and mid-front /ɛ/.

Duration. Mean total duration of canonical syllables, and mean duration of the vocalic portion are shown in Table 41. Canonical syllables produced by the typically developing comparison group were on average 267 milliseconds (ms) in total duration, with the vocalic portion 232 ms. Total duration for SIB1 was not significantly different to the comparison infants, $t(7) = 0.25$, $p = .40$, although descriptively it was outside the range observed for the comparison infants (i.e., longer). Duration of the vocalic portion, however, was significantly longer, $t(7) = 3.28$, $p = .01$, possibly related to the relative frequency of nasal onsets (i.e., [m] and [n], compared to the siblings who also produced a range of stops; see Table 40). There were no significant differences in these measures when SIB2 was compared to the typically developing infants, $t(7) = 0.96$, $p = .19$ and $t(7) = 1.59$, $p = .08$, for total duration and duration of vocalic portion, respectively.

Table 40

Canonical Syllabic Gestures used by the Typically Developing (TD) Infants and Siblings (SIBS) 1 and 2 at the Earliest Recording of Canonical Syllables (9 months for the TD infants, and 12 months for SIBS1 and 2)

Infant	Canonical syllables produced during vocalisation sample
C1	[dɛ] [dæ] [nɛ] [gɪ]
C2	[di] [də] [du] [bə] [bɒ] [mə] [gə] [gɛ] [k^hæ]
C3	[dæ] [dɛ] [mæ]
C4	[dæ] [dɛ] [dʌ] [də] [di] [tɛ] [bʌ] [bæ] [mə] [næ] [kə]
C5	[dæ] [gæ] [da] [dɛ] [tʌ] [ʧæ] [gə] [bə] [tə] [nə] [sə] [dʌ] [bɛ]
C6	[dæ] [dɛ] [tʌ] [tæ] [tɛ] [bə] [bæ] [gə] [gɛ]
C7	[di] [nə] [næ] [nʌ] [gæ]
C8	[dɒ] [da] [t^hæ] [bæ] [və]
SIB1	[dæ] [bɜ] [bə] [nə] [nʌ] [mæ] [mə]
SIB2	[dɛ] [bə] [mə]

Note. Canonical syllables are shown as these were the focus of acoustic analyses

Table 41

Mean Duration, Fo Mean and Standard Deviation Values for Canonical Syllables

	n	Duration (ms)	Duration Vocalic (ms)	Fo Mean (Hz)	Fo SD (Hz)
C1	2	331.5	264.0	284.5	15.7
C2	17	292.4	254.4	306.6	17.9
C3	12	244.5	235.5	320.0	23.2
C4	19	228.0	196.1	318.1	20.8
C5	26	325.0	288.8	342.9	27.7
C6	31	260.8	239.6	318.9	19.3
C7	6	205.5	192.7	272.3	18.5
C8	7	250.6	183.6	296.4	18.6
M	*15*	*267.3*	*231.8*	*307.5*	*20.2*
SD	*10.1*	*45.2*	*37.8*	*22.5*	*3.7*
SIB1	28	377.6	370.5	259.5	27.0
SIB2	10	313.1	295.4	364.3	65.2

Fundamental frequency (Fo). Table 41 also displays Fo mean and standard deviations of canonical syllables for the infants studied. Mean Fo for SIB1 (259.5 Hz) was significantly lower than the comparison group, $t(7) = 2.01$, $p = .04$. However, variation (standard deviation) for the same infant was not significantly different to the comparison group, $t(7) = 1.70$, $p = .07$. In contrast, SIB2's mean Fo (364.3 Hz) was significantly higher than the comparison group ($M = 307.5$, $SD = 22.5$), $t(7) = 2.38$, $p = .02$. In addition, the variation was significantly greater for SIB2, $t(7) = 11.32$, $p < .001$.

F1 and F2. Mean and standard deviations for the first two formants are shown in Table 42. Mean F1 for the comparison group was 891.6 Hz (*SD* = 122.4); F2 was 2739.7 (*SD* = 321.8). For SIB1, F1 fell within the range and was not significantly different to that of the comparison infants, $t(7) = 0.53$, $p = .31$, but F2 was significantly lower, $t(7) = 2.78$, $p = .01$. F1 for SIB2 was not significantly different to the comparison group, $t(7) = 0.56$, $p = .30$. Second formant values however, were also significantly lower for SIB2, $t(7) = 1.89$, $p = .05$. Coefficient of variation for both F1 and F2 are also displayed in Table 42. Case comparisons indicated that SIB1 did not show any statistically significant differences to the comparison group on this measure for either F1 or F2, $t(7) = 0.68$, $p = .52$, and $t(7) = 0.79$, $p = .46$, respectively. In contrast, SIB2's coefficient of variation of F1 was approaching statistical significance (descriptively higher and beyond the range of the comparison infants'), $t(7) = 2.31$, $p = .054$, while there was no statistical difference in the same measure for F2, $t(7) = 0.84$, $p = .43$.

Scatterplots displaying the relationship between F1 and F2 for prelinguistic canonical syllables produced by each of the comparison infants and the two siblings under investigation are displayed in Figure 8. The scatterplots are presented on a single page to facilitate visual comparison of the individual relationships between F1 and F2. Figure 9 shows the combined data for the comparison infants and SIB1 and SIB2, presented on the same axes. The scatterplots reveal that while there was variability with respect to the number of canonical syllables produced, the typically developing infants appear to be utilising a larger vowel space. As described above, F2 can be seen as lower in the siblings studied, compared to the typically developing infants. As can be seen in both Figures 8 and 9, a particularly strong relationship between F1 and F2 was evident for SIB2.

Table 42

Mean, Standard Deviation, Coefficient of Variation and Correlation Coefficients for F1 and F2 for Canonical Syllables Produced by SIBS 1 and 2 and the Comparison Infants

	F1			F2			$r_{F1:F2}$[a]
	Mean	SD	CoVar	Mean	SD	CoVar	
C1	772.5	224	0.29	2931.5	255	0.09	-[b]
C2	707.2	121	0.17	2538.6	563	0.22	-.41
C3	930.4	212	0.23	3287.1	627	0.19	-.01
C4	905.3	303	0.33	2858.6	554	0.19	-.59**
C5	1044.4	213	0.20	2830.7	380	0.13	-.08
C6	1028.5	224	0.22	2693.8	383	0.14	.53**
C7	950.2	136	0.14	2585.7	505	0.20	.17
C8	794.2	157	0.20	2192.0	183	0.08	-.36
m	*891.6*	198.8	0.22	*2739.7*	431.3	0.16	-.11
sd	*122.4*	58.9	0.06	*321.8*	157.6	0.15	.38
SIB1	823.1	221	0.27	1789.6	357	0.20	.49**
SIB2	964.5	362	0.38	2093.8	425	0.20	.90**

[a] Pearson correlation coefficient for F1 and F2 [b] insufficient number of syllables to run correlation

**significant at $p = .01$ level (none were significant at only .05)

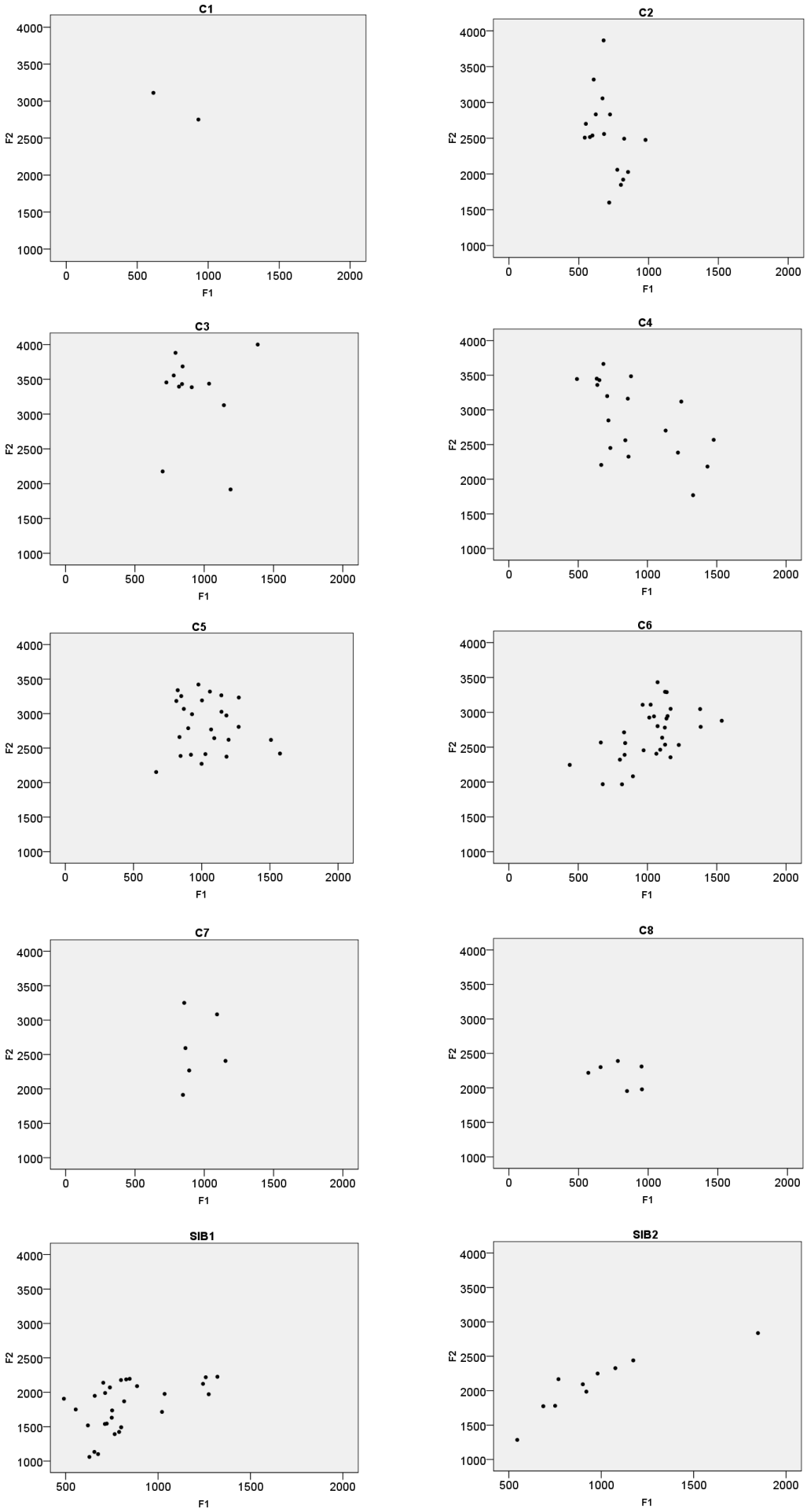


Figure 8. *Scatterplots showing F1 and F2 for canonical syllables for the comparison infants (C) and siblings 1 and 2*

Correlation coefficients for F1 and F2 (displayed in Table 42) explored for the infants revealed a significant negative correlation for C4, and a significant positive correlation for C6. The two formants were also strongly positively correlated for SIB1, r = .49, p = .01, and SIB2, r = .90, p < .01. Analyses (z test of two independent correlations) comparing the positive correlations (Meng, Rosenthal, & Rubin, 1992) revealed a significantly stronger correlation for SIB2, compared to both C6 and SIB1, z_{diff} = 2.09, p = .04, and z_{diff} = 2.19, p = .03, respectively.

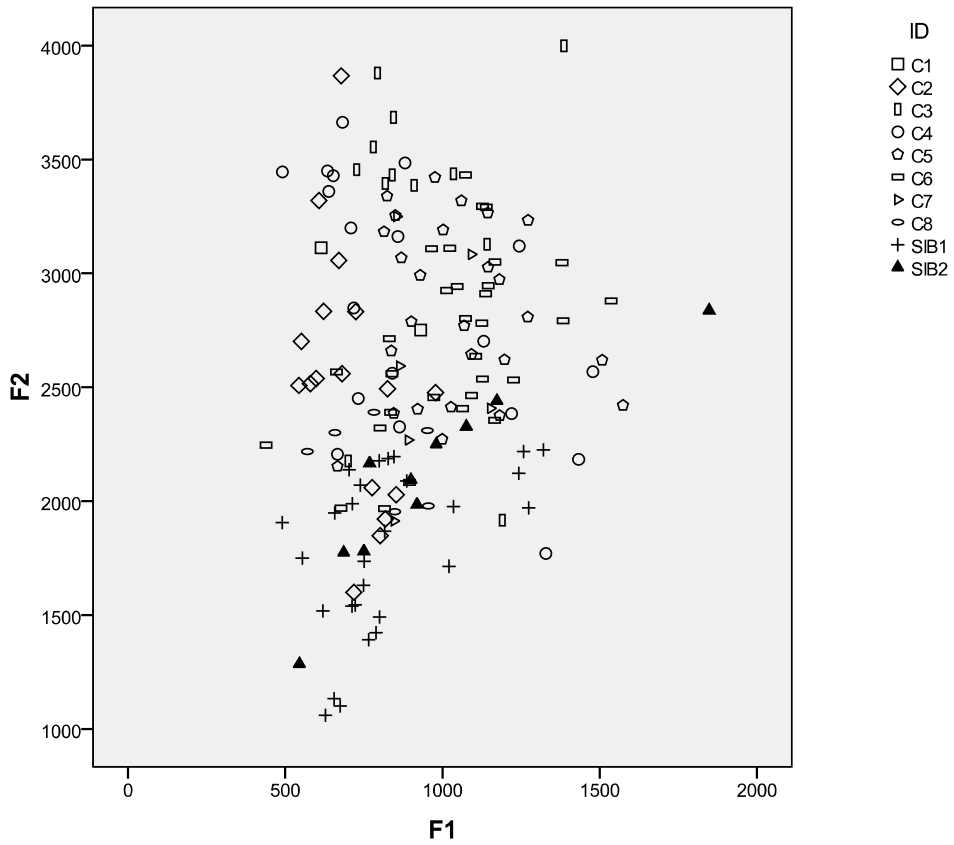


Figure 9. Scatterplot displaying F1 and F2 combinations for the comparison infants (open/unfilled shapes) and siblings 1 (cross) and 2 (filled triangle).

Dissociation of Conceptualiser and Speech Motor Areas

Applying the Revised Standardised Difference Test procedure (Crawford & Garthwait, 2005; Crawford et al., 2003), a significant dissociation in measures of conceptualiser and speech motor ability was observed for SIB2, $t(7) = 4.31$, $p < .001$. The pattern of performance, with no significant difference to the comparison infants

on the social composite of the CSBS caregiver questionnaire, but a significant deficit on the sounds subtest, *and* a significantly larger discrepancy than the comparison sample distribution, represents a classical dissociation (Crawford & Garthwaite, 2005). In contrast, no such dissociation was observed for SIB1, $t(7) = 0.67$, $p = .52$.

Discussion

The communication skills of 16 infants, half of whom had a family history of CAS, were investigated longitudinally in Study 3. General development, and speech and language skills were tracked from 9 to 24 months of age in the two groups of infants: siblings of children with CAS, and a comparison group with no such family history. Group comparisons were made in the first instance, and were informative in relation to notions of a broader phenotype (Lewis, Freebairn, Hansen, Taylor et al., 2004). Individual profiles were subsequently inspected for communication status at 2 years, and CAS-features over time. At 2 years of age, two of the infant siblings (and none of the comparison group) had not met expected speech and language milestones. Investigation of the pattern of skills suggested one of the siblings to present with features highly suggestive of possible CAS; the other did not present with such features. More in-depth analyses of these and the comparison group infants' vocalisations were examined and are reported below, following a discussion of overall trends in the groups' developmental profiles.

Developmental Profiles of Infants with a Family History of CAS.

Comparison of group profiles revealed that, as predicted, the siblings demonstrated lower expressive language scores, lower scores on fine motor development, and lower scores on speech sound development than the comparison infants. These group differences did not interact with the sampling timepoint, suggesting a general persistence of such deficits and their presence from the earliest timepoint sampled. On one of the two measures of receptive language development (REEL-3 receptive ability score), the siblings scored significantly lower than the comparison infants. However, the groups did not show a statistically significant difference on the understanding subtest of the CSBS, and inspection of individual siblings' scores showed that the large majority were within normal limits on the REEL-3 receptive

ability score at each timepoint, suggesting mixed results regarding receptive language ability in the siblings. The siblings also showed lower scores on the Problem solving subscale of the ASQ, although there was only one instance of a clinically-important depression in scores (for SIB3 at 9 months only).

Investigation of group and individual profiles over time highlighted the variability and dynamic nature of development. The significant main effects observed for timepoint on a number of the measures in the present study, with a trend of generally increasing scores with age, are also indicative of variability in this developmental period. Although standard scores were used (and thus there is no clear reason why scores would show a general increase over time), it is possible that they are reflective of the tendency for typically developing children to 'catch up' in any initially-delayed areas in early development (Horner, 1988). There were instances where infants in the comparison group (who at 2 years of age showed communication development within normal limits) scored below cut-offs on individual assessment tools. This is consistent with research demonstrating instability in serial assessments of typically developing infants across this age group (Darrah, Hodge, Magill-Evans, & Kembhavi, 2003). However, occurrences of below-typical scores were rare and transient, with all comparison infants showing normal language development by 2 years of age.

All eight infant siblings failed the expressive component of the WILSTAAR screen. The sole expressive item on this screen relates to the use of variegated babbling. Although often thought to be developmentally more advanced, variegated babbling has been shown to co-occur with reduplicated babbling (Mitchell & Kent, 1990). Research has yet to establish the clinical significance of a lack of variegated babbling by 9 months of age, despite knowledge that the production of canonical babbling by 10 months is an important communication milestone (Oller et al., 1999). Nonetheless, it is interesting that each of the siblings (and none of the comparison infants) failed the WILSTAAR screen. The siblings as a group at 9 months also showed lower scores than the comparison group on the Speech composite of the CSBS infant-toddler checklist, suggesting restricted vocalisation development.

In addition to lagging in vocalisation development, the siblings demonstrated significantly poorer fine motor skills (on both the ASQ fine motor and CSBS Object Use subtests), even though at some timepoints no individual scored less than the recommended cut-off for concern. This pattern of overall depressed fine motor skills in the siblings (as a group) is consistent with the close relationship between fine motor and speech motor development proposed to exist in normal development (Locke & Pearson, 1990), as well as descriptions of motor coordination difficulties in children with speech sound disorders (Bradford & Dodd, 1996). It is possible that any broader phenotype of CAS may include relatively poorer fine motor development.

Across time, expressive language was poorer in the siblings group. When measured in the prelinguistic period (i.e., before the child is actually talking) measures of this skill typically encompass broad conceptualisations of 'language', including vocalisation, babbling and gesture use. As development progresses, expressive language is typically defined more by word use and the development of syntax. The generally weaker skills in these areas observed for the siblings culminated in significantly lower expressive vocabulary scores (at 18 and 24 months) and weaker sentence complexity at 24 months of age. It is interesting to note that even though two siblings showed clinically-important depressions in expressive vocabulary, another four siblings showed expressive vocabularies below the lowest reported for the comparison infants.

By the time the children were 24 months, communication skills were within normal limits for most of the infants studied. None of the comparison infants evidenced any speech and language difficulties at this age. Descriptively, the siblings as a group scored lower than the comparison infants in the areas of speech sound production, fine motor development, expressive vocabulary and sentence complexity. Two of the infant siblings showed clinically-important deficits in communication ability at 24 months of age.

The observation of generally lower scores on speech and language measures for the siblings group, and that two of the infant siblings presented at 2 years with significantly delayed communication skills but varied profiles, is consistent with a verbal trait deficit proposed by Lewis and colleagues (Lewis, Freebairn, Hansen,

Taylor et al., 2004). In their study of familial aggregation of speech and language disorders in the family members of children with CAS, siblings presented with a range of disorders including mild articulation problems, severe language and speech sound disorders, and CAS. The authors proposed that traits underlying CAS may be polygenic. This is consistent with findings reported on the FOXP2 gene mutation in that significant numbers of children with CAS features have not shown the specific mutation identified in the KE family and individual clinical cases of CAS (Alcock, Passingham, Watkins, & Vargha-Khadem, 2000a; MacDermot et al., 2005; Watkins et al., 2002).

That two out of the eight siblings studied in the present study presented with delayed or disordered communication development suggests a significant 'affection' rate, consistent with previous research. Higher affection rates were reported by Thoonen and colleagues (Thoonen et al., 1997), and Lewis and colleagues (Lewis, Freebairn, Hansen, Taylor et al., 2004), who reported a family history of speech and language disorders for 6 out of 11 (55%), and 19 out of 22 (86%), children with CAS studied, respectively. The rates are not directly comparable however, considering the differences in study purpose and design. The present study used family history to identify infant siblings for investigation, rather than gathering epidemiological data on how many children with a CAS diagnosis have a family history (including parental family history) of the disorder. In contrast, the Thoonen (Thoonen et al., 1997) and Lewis (Lewis, Freebairn, Hansen, Taylor et al., 2004) studies reported rates of family history of speech/language disorders in children with CAS. However, results such as these suggest that there may be a role for screening younger siblings in clinical populations. Tools such as the CSBS Infant-Toddler Checklist show promise for this purpose based on the results of the present study. The siblings showed significantly lower scores on the speech composite of the checklist, and in contrast to the WILSTAAR screen (which all of the siblings failed and thus appears to inflate the rate of 'false-positives'), the CSBS Infant-Toddler identified three siblings to be below expectations (including SIB2 , but not SIB1).

One infant sibling in this study (SIB2) showed a pattern of not only delayed communication development, but also features consistent with an early CAS-type profile (Davis & Velleman, 2000). This included a significantly restricted phonetic inventory, lack of consonant-vowel babble, a highly developed system of gestures/signs, and late motor milestones. At 2 years of age, it is inappropriate and impossible to confirm if CAS is the appropriate diagnosis for this child (Davis & Velleman, 2000); however, the pattern of performance over time and clinical presentation was highly suggestive. Investigation of this infant's (hereafter, SIB2) communication skills, measured on standardised tools from 9 months of age to 24 months of age, revealed a pattern consistent with theoretical predictions about the presentation of CAS, explored further below.

Cognitive and Conceptual Skills

As estimated by the Problem-Solving subscale of the ASQ, SIB2 showed normal cognitive ability, consistently achieving scores well within the normal range over time. General observation of abilities over time also corroborated this finding. Although CAS can occur in children with cognitive deficits (American Speech-Language-Hearing Association, 2007), the observation of normal cognitive skills removes the possibility of this confound for this case. Moreover, consistent with the notion of initial independence from the emerging speech motor control system (Levelt et al., 1999), conceptual development was strong. Measures of communicative intent (i.e., the Gesture and Expression-Eye Gaze clusters of the CSBS) were consistently well within the normal range for this infant.

Receptive and Expressive Language

Receptive language is often reported to be a relative strength for children with CAS, and a developmental perspective emphasises that this would be most evident early on, prior to the interactive processes involved in development (Karmiloff-Smith, Scerif, & Ansari, 2003). At each age sampled, receptive language skills were found to be age-appropriate for SIB2, again consistent with the notion of initially independent speech motor and conceptual development in infancy.

In contrast, difficulties with expressive language are often (almost universally) reported in children with CAS (Ekelman & Aram, 1983; Lewis, Freebairn, Hansen, Iyengar, & Taylor, 2004). SIB2 showed consistent delays in expressive language from 9 to 24 months of age, reflected in REEL-3 language ability scores. By 15 months, delayed expressive vocabulary development was evident, with scores on the CSBS Words cluster below age expectations. By 24 months, expressive vocabulary was showing further delays, and sentence complexity was restricted. Usually explained by the presence of concomitant language disorder, recent theoretical approaches to CAS account for such language deficits as emerging as a consequence of an original speech motor deficit in a developing system (e.g., Maassen, 2002). The restricted set of articulatory gestures in the protosyllabary implies that although receptive vocabulary can continue expanding, the toddler's expressive vocabulary is limited by an impaired ability in production.

Speech Sound and Syllable Development

As measured by the Sounds subtest of the CSBS Caregiver Questionnaire, SIB2 showed impaired development of syllables, even from 9 months of age. This deficit persisted at each age sampled, and manifested in a restricted phonetic inventory and range of syllables. Only one other sibling was below age-expectations in the sounds subtest at 9 months of age, and this did not persist across the sampling timepoints. Such a persistent deficit in SIB2 is consistent with a core deficit in speech motor control (Maassen, 2002), explored further below with reference to the vocalisation samples.

Rate of Vocalisation

Comparison of the vocalisation samples at 9 months of age suggested that the rate of vocalisation for SIB2 was not significantly less than that of the comparison infants. This finding is interesting and potentially in contrast to previous anecdotal suggestions about CAS. Anecdotally, CAS children have been described as being quiet as infants (Davis & Velleman, 2000), and the parent report results in Study 1 support this depiction. Objective quantification of vocalisation rates for the infant in question in this study does not appear to support this assertion. However, it is possible (and perhaps probable) that parental perception of how vocal an infant is may be related to the amount of *canonical* vocalisations, rather than total

vocalisations in general. That is, infants with CAS-type features may well vocalise, but not using readily-identified canonical syllables that are universally and intuitively noticed by parents (Oller, Eilers, & Basinger, 2001).

Vocalisation Type

Consistent with observations in large groups of typically developing infants, all comparison infants had entered the canonical stage by 9 months of age (Nathani et al., 2006; Oller, 2000; Oller et al., 1999). This was reflected both in the parent report of sounds used, as well as in the vocalisation samples. All comparison infants were reported to be producing canonical syllables, with all also reportedly using variegated forms. Analysis of the vocalisation samples corroborated the parent report, with all producing a range of canonical syllables (on average, 13% of vocalisations were canonical, with another 8% representing advanced forms). Oller and colleagues (Oller et al., 1999) have demonstrated the robustness of canonical babbling, with emergence occurring between 6 and 10 months in typical development.

For the two siblings showing delayed language development at 2 years of age, no canonical syllables were observed in their 9 month vocalisation samples. For SIB2, this was also evident in parental report – canonical syllables were not documented until 12 months of age. SIB1 was reported to be using some canonical syllables at 9 months, although this was not observed during the vocalisation sample. Analysis of their 12 month vocalisation samples revealed that SIB2 was still using significantly less canonical syllables (2.5%) compared to the typically developing 9 month olds (who averaged 17% with none producing less than 6%). SIB1 used approximately 15% by this age, perhaps suggesting that the initial lag represented a delay rather than impaired speech motor control.

The results reported here for SIB2 are consistent with the few descriptions of young children with CAS reported in the literature. The case studies of two preschool children with CAS described by Velleman (1994) included reported histories of delayed or decreased babbling and late emergence of first words. Tate (1991, as cited in Shriberg, Aram, & Kwiatkowski, 1997b) similarly reported a history of delayed babbling in a case study of a child with CAS. Reduced, absent or delayed canonical babbling, however, may not be specific to CAS (Oller, 2000). There is growing

evidence for the continuity of vocalisation and language development, with restricted vocabulary development evident in groups of children showing restricted vocalisation development in infancy (Oller et al., 1999). In the present study, SIB1, who at two years of age was showing delayed expressive language development but age-appropriate speech sound acquisition, also showed deficits in prelinguistic vocalisations. As was seen in Table 35, the profile over time for SIB1 included depressions in measures of conceptualiser development at 9 months (receptive language, gesture use, and expression and eye gaze), followed by subsequent deficits in expressive language, expressive vocabulary and syntactic development, persisting to 2 years of age. Prelinguistically, her overall rate of vocalisation, though not reaching statistical significance, was descriptively lower than the range observed in the comparison infants, perhaps suggesting a different source of delayed speech/language development in this infant. Parent report for SIB1, in contrast to that for SIB2, indicated the presence of canonical syllables at 9 months despite the lack of such syllables in the vocalisation sample.

A core tenet of the theory investigated in the present thesis predicts that the source of deficit in children with CAS affects speech motor control prelinguistically. Acoustic analyses were therefore used to explore the nature of prelinguistic vocalisations further.

Acoustic Measures

Canonical syllables were investigated acoustically for evidence of a core speech motor control deficit. In typical development, canonical syllables represent the first 'adult-like' syllables containing a closant and vocant. It was hypothesised that acoustic measures may reflect a core deficit in articulatory control, suggesting a qualitative difference over and above any quantitative difference that may reflect delayed development.

Duration. Syllable durations for the comparison infants were consistent with the range normally found in typically developing infants (i.e., 100 to 500ms, Rvachew et al., 2002). Total durations were not significantly different to the comparison infants for either SIB1 or SIB2. In the case of SIB2 who showed a profile consistent with early features of CAS, longer syllable durations were predicted based on notions of slower articulation rates or less coarticulation. Longer syllable

and word durations, for example, have been reported for children with CAS (Bahr, 2005; Nijland, Maassen, & van der Meulen, 2003). However, the observation of 'normal' syllable durations may instead support the absence of any dysarthric element in this infant. The duration of the vocalic portion of the syllables was longer for SIB1 than the comparison infants. This may be reflective of the predominance of nasal onsets for this infant.

Fundamental frequency (F_o). Average fundamental frequency of the canonical syllables produced by the comparison group infants was within the range reported for typically developing infants (Kent & Murray, 1982). Consistent with the older age at which canonical syllables were produced, and thus potential biological effects of vocal tract size and length of vocal cords (Kent, 1976; Voperian & Kent, 2007), SIB1's mean Fo was significantly lower than that of the comparison group. In contrast, fundamental frequency for the infant hypothesised to be showing early CAS-related features was significantly higher. Variation in fundamental frequency was also significantly greater. These results are particularly interesting as they go against what would be expected based on maturational differences alone. They suggest atypical speech motor control (Kent, 1976). Large variability in fundamental frequency may reflect poor laryngeal control and /or neurological immaturity of the speech motor control system (Lieberman, 1969; Bosma, Truby & Lind, 1965, as cited in Kent, 1976). No such significant variability was observed for SIB1.

F_1 and F_2. Measures of the first two formants in canonical syllables produced by the infants indicated no differences in F1 for either of the infant siblings compared to the comparison group infants. F2, however was significantly lower for both infant siblings. The most obvious explanation of this finding relates to the fact that F2 typically lowers with age as a consequence of biological changes in the vocal tract size (Kent, 1976), although it has been suggested to be relatively stable from 4 to 24 months of age (Voperian & Kent, 2007). However, as F2 is sensitive to tongue advancement, the results could also suggest a slightly more retracted tongue position or a lack of production of front vowels. Coefficient of variation calculated for both formants (in order to more suitably control for variation vocal tract size as well as the lack of control over phonetic context) suggested greater F1 variability for SIB2, but not for F2 for the same infant. Research has indicated that although variability of both formants tends to decrease with age, F1 may achieve stability earlier than F2, with the hypothesis that jaw stability is achieved earlier than motor control of other

articulators (Nittrouer, 1993). If this is so, the results of the present study may suggest particularly immature motor control (perhaps affecting jaw stability) for SIB2. This suggestion of greater variability in F1 requires replication given it received some, but not strong, statistical support. No differences in these measures compared to the comparison sample were observed for SIB1.

SIB2's F1 and F2 formant patterns were also atypical in terms of there being a particularly strong correlation between the two formants; a pattern not observed in any such strength in the comparison infants (and also not observed in SIB1). This may suggest tighter coupling of the articulators (with F1 reflecting tongue height, and F2 tongue advancement), consistent with theories of articulatory phonology (Browman & Goldstein, 1992) and patterns underlying babbling (MacNeilage & Davis, 1990). Articulatory phonology views gestures as the basic units underlying phonological contrasts. The Frame-Content theory of babbling also posits syllabic articulatory gestures to consist initially of gross movements of the jaw. It is only over time that the articulators begin to move independently of one another in speech. Thus, tighter coupling, like that observed for SIB2 between tongue height and advancement, may reflect immature movement patterns or impaired speech motor control.

Functionally, this infant produced a limited range of syllabic articulatory gestures, with a productive consonant inventory at 12 months of only 3 consonants (i.e., [b], [d], and [m]), and correspondingly limited vowels. The acoustic findings reported above are consistent with the limited phonetic picture observed for the child. However, it should be noted that the acoustic findings require replication, especially given the low number of canonical syllables that were available from this infant for analysis.

Dissociation Between Conceptualiser and Speech Motor Systems

As hypothesised, a significant classical dissociation was observed between conceptualiser and speech motor control abilities in infancy for SIB2. This infant showed significantly poorer speech motor development than the comparison sample, in the context of intact conceptual development, and a significantly larger discrepancy in scores. This finding has important implications for modelling of speech and language development. Levelt's (1999) adapted model of early speech production

posits two initially independent systems in infancy – a conceptual system and an articulatory motor system. The results of the present study support this proposition. Even though such a dissociation was found for only one infant, such a classical dissociation in abilities supports initially independent systems in early development. The affected infant demonstrated a significantly restricted repertoire of syllabic articulatory gestures, despite intact conceptual/conceptualiser skills.

Limitations

A number of methodological issues need to be considered when interpreting the results of Study 3. Although longitudinal studies may provide the best way to investigate the natural progression of CAS and identify the core deficit, the timeframe of the present study did not allow confirmation of a CAS diagnosis. It is not yet possible or appropriate to diagnose CAS in infants or toddlers (Davis & Velleman, 2000), so although features consistent with such a diagnosis were identified in one infant sibling, further investigation over a longer time frame is necessary to draw firm conclusions for this individual.

Moreover, there are obvious restrictions in generalising such results from one child. The single case methodology utilised in the present study, although avoiding many of the difficulties associated with group studies (Bishop, 1997; Caramazza, 1986; Crawford & Howell, 1998), restricts the degree to which conclusions can be made regarding the CAS population as a whole. However, the case demonstrates that it is possible for an infant to show the type of dissociation predicted from a prelinguistic speech motor deficit account of CAS. Larger group longitudinal studies are required to see whether this type of origin typifies children who later meet clinical diagnosis for CAS, or whether CAS can result from alternative developmental pathways.

A limitation of the acoustic data relates to differences in the age of production of pre-linguistic canonical vocalisations. Although vocalisation samples were obtained at 9, 12 and 18 months of age, acoustic analyses focussed only on prelinguistic canonical vocalisations. This was to address speech motor control prior to the coupling of the conceptual and articulatory-motor systems (Levelt et al., 1999). For the typically developing comparison infants, these were produced at the 9 month data collection session. The two siblings showing communication deficits at 24 months of age did

not produce any canonical vocalisations in their samples until 12 months of age, however. Differences in acoustic measures could therefore be attributed to biological differences relating to size of the vocal tract, as described above (Kent, 1976). However, the finding of a significantly higher mean and variability in the fundamental frequency, and the significantly stronger correlation in F1 and F2 values, with associated restricted vowel space for SIB2, can not be explained solely by biological factors.

A secondary limitation arising from the acoustic analysis of infant canonical syllables is that, unlike analyses of older children's speech, it is not possible to control the syllable type and number produced by each infant. Thus infants differed in terms of which syllables they spontaneously produced, and the amount of these. Although this presented limitations in the nature and interpretation of acoustic analyses, this information in itself is rare and informative with respect to description of the vocalisations of infants who may show increased risk of CAS.

Conclusions

Despite these limitations, the present study makes important contributions to the study of CAS and to theoretical accounts of both normal and disordered communication development. There are presently no published longitudinal investigations of CAS from pre-speech to speech (Zeigler & Maassen, 2004), despite there being an established need for such studies (American Speech-Language-Hearing Association, 2007). The present study documents the developmental trajectory of speech and language development in infants with a family history of CAS. Moreover, description of two infants who at 2 years of age show delayed and/or disordered development allowed direct investigation of a core deficit in speech motor control hypothesis of CAS.

Group comparisons, showing generally poorer speech and language skills in the siblings, as well as the observation of delayed and/or disordered communication ability in two of the siblings, provided support for the verbal trait deficit hypothesis proposed by Lewis et al. (Lewis, Freebairn, Hansen, Taylor et al., 2004). Moreover, the results of the present study confirm that it is possible for a child with heightened risk of CAS to show, pre-linguistically, a dissociation between modalities consistent

with a core deficit in the emerging speech motor control system. Such a motoric deficit has been previously proposed by a number of researchers, but rarely interpreted in the context of the developing infant system (Maassen, 2002). Importantly, the present study also highlighted the utility of longitudinal paradigms in the study of CAS, using knowledge of familial aggregation to identify infants for investigation. Theoretical, research and clinical implications from this and Studies 1 and 2 will be considered further in the following chapter.

CHAPTER 5

GENERAL DISCUSSION

"Speech production is a highly precise and practiced motor skill"
(Hodge, 1994, p.92)

Overview

The present research aimed to examine a theoretical account of CAS which encompasses a developmental model of speech production. In this account, a core deficit in speech motor control, affecting perceptuo-motor learning, is hypothesised to be responsible for the array of characteristics observed in children with the disorder (Maassen, 2002). The notion predicts that there will be evidence of such a deficit prelinguistically, and thus the present research focussed on this developmental period. Results from the three studies were broadly consistent with this notion, notwithstanding ongoing debate concerning the differentially diagnostic phenotype of CAS and exceptions to the general pattern of observations.

Generated from the core speech motor control deficit account, two broad research questions were explored:

1. Do children with CAS show deficits in early vocalisation development consistent with a speech motor control account of the disorder?
2. Do infants at risk of CAS show a profile consistent with evidence of a dissociation between conceptual and speech motor control abilities in early development?

A combination of methodologies was employed to investigate these questions: retrospective parent report, analysis of retrospective infant data, and prospective longitudinal investigation of infants considered at risk of CAS. Similar

methodologies have been used in the study of other developmental disorders, most notably autism spectrum disorders (e.g., Bryson et al., 2007; Coonrod & Stone, 2004; Iverson & Wozniak, 2007). Despite acknowledgement of the urgent need for such investigations in the area of Childhood Apraxia of Speech (American Speech-Language-Hearing Association, 2007), the present research appears to be the first to apply such combined methodologies to the disorder.

Study 1 quantified parental report of early vocalisation behaviours in children with sCAS. In comparison to children with SLI and typically developing speech and language development, the sCAS group were reported to have specific differences in early development: namely, being less likely to have babbled, being later in the emergence of two word combinations and showing commonly constrained language and motor development. These results are consistent with expectations based on theoretical models of language development (Bailly, 1997; Maassen, 2002) and previous anecdotal suggestions (Hall, 2003a). However, a lack of comprehensive clinical data on the sCAS children, and the reliance on the assumed reliability of parental report, indicated the need for further research to corroborate and extend the findings.

In Study 2, investigation of retrospective infant data for a clinical sample of children allowed further investigation of the speech motor control deficit hypothesis. Results from the first phase identified and documented CAS features in the children, with specific criteria employed to quantify the presence of commonly reported characteristics. Infant data available for the same children, when compared to infant data for a large sample of children without identified persisting communication deficits, allowed more direct investigation of hypothesised early CAS features. The results supported the notion of impaired speech motor control as being a potential core deficit in CAS. In particular, the child showing the greatest number and highest severity of CAS features showed the predicted pattern of limited syllabic articulatory gestures but intact conceptual development in infancy. However, a range of profiles were reported, both in the infant data and in the 3-4 year old data.
A prospective longitudinal study of infant siblings of children with CAS (Study 3) allowed detailed investigation of vocalisations and developmental trajectories for infants from 9 months of age to 24 months of age. Whilst the data collection

timeframe did not allow CAS to be diagnosed in any of the toddlers, the results highlighted one case in particular with a profile suggestive of the disorder, potentially representing the first longitudinal investigation of CAS from infancy. The results supported the notion of initially independent conceptual and speech motor development, the possibility of dissociated development at this early stage (Levelt, Roelofs, & Meyer, 1999), and the presence of a core deficit in speech motor control (Maassen, 2002).

Participant Similarities Across Studies

Similarities in the characteristics of a number of participants across the three studies are worth comment. In Study 1, the participants with the highest number of CAS features were those where parent report for vocalisation behaviours was mostly negative – that is, they were reported to be relatively quiet as infants, not to have babbled, were later in the emergence of first words and two word combinations, and were also later in most motor milestones. Similarly, Study 2 demonstrated that the participant with the most number and greatest severity of CAS features showed a particular pattern of a lack of consonant-vowel babble in infancy and dissociated impairment in expressive but not receptive and conceptual abilities. Consistent with the first two studies, Study 3 demonstrated the presence of atypical vocalisation development in the context of intact conceptual skills in the one infant showing a pattern most suggestive of CAS at 2 years of age.

These converging results support theoretical and clinical hypotheses and highlight the need for further longitudinal research. A number of researchers have suggested that the clinical features of CAS may be evident from very early on in development. Maassen (2002), for example, suggested "among the first signs of a dyspraxic development, often assessed in retrospect, is reduced babbling in combination with a delayed or deviant oral motor development" (p. 260). Anecdotal reports similarly suggest such early deficits in vocalisations (Hall, 2003a). Moreover, application of a developmental model of early communication proposed the possibility of an isolated core deficit, with negative effects on subsequent linguistic development (Levelt et al., 1999).

CAS – An Impairment With a Core Deficit in Speech Motor Control?

As outlined in the introductory chapter, historically, CAS was initially conceptualised as a deficit of speech motor abilities (Morley, 1965). The disorder was hypothesised to reflect impaired motor planning and/or programming (Stackhouse, 1992), located 'downstream' from linguistic processes but further 'upstream' than actual execution of movements. Observation of language deficits in most children with CAS, however, led researchers to question the adequacy of such a 'motoric' theory in accounting for the seemingly divergent characteristics. Alternative explanations emerged, positing a linguistic deficit as underlying the disorder. Core impairments in timing (Peter & Stoel-Gammon, 2008), phonological representations (Marion, Sussman, & Marquardt, 1993), and/or the assignment of lexical stress (Shriberg et al., 2003) are examples of linguistic accounts of CAS that have been proposed over the years.

The two alternative explanations of CAS (i.e., one proposing speech motor control as the locus of core deficit; the other suggesting it to be linguistically-based) have traditionally been framed as being mutually exclusive theories. More recently, researchers have reframed the debate as more appropriately being conceptualised as a motoric-only deficit versus a linguistic *and* motoric impairment (American Speech-Language-Hearing Association, 2007), implying co-morbidity in the latter explanation. The present thesis, however, proposed that typical accounts of CAS are limited by their lack of interpretation within a developmental framework. A core deficit in speech motor control, when interpreted in the context of a developmental model, is able to account for the presence of motoric *and* linguistic impairments in CAS, evident after a period of development. The hypothesis predicts, importantly, that the core impairment would be evident in infancy, in the context of intact conceptualiser development. It is the developmental process itself that results in the varied presentation and degree of impairment in individual children (Karmiloff-Smith, 1998; Karmiloff-Smith, Scerif, & Ansari, 2003). The results of the present research were consistent with this hypothesis, and demonstrated the viability of impaired speech motor control as being implicated as an original source of deficit with ongoing negative consequences for the emerging speech and language system.

Further discussion of these results in relation to the proposed developmental trajectory of CAS is presented below.

Setting up the Protosyllabary

Maassen (2002) proposed that a core deficit in articulatory motor (speech motor) control, affecting perceptuo-motor learning, may underlie CAS. Previously, numerous researchers proposed a core deficit in speech motor control, but few interpreted this deficit within a developmental framework. According to Levelt and colleagues (Levelt et al., 1999), the articulo-motor system enables the production of various speech gestures – a set of babbles that begin to form the 'protosyllabary' in the infant system. In the case of an infant with impaired speech motor control, the protosyllabary would be restricted. Few studies have directly investigated this account, however. This notion was certainly supported in the present research. Reduced or delayed babbling identified via parental recall (Study 1) and retrospective infant data (Study 2) provided support for the idea of a restricted set of gestures in the protosyllabary for children with CAS. Study 3 demonstrated objectively that an infant showing a profile suggestive of CAS had not entered the canonical babbling stage by the age expected in normal development, and thus also demonstrated a restricted protosyllabary, prelinguistically.

Many current theoretical accounts of prelinguistic vocal development highlight the importance of babbling for later speech production ability. Davis and MacNeilage's Frame-Content theory, for example, emphasises the motoric basis of canonical syllables (Davis & MacNeilage, 1995; MacNeilage, 1998). Babbling occurs when the infant combines vocalisation with rhythmical oscillations of the jaw (MacNeilage, Davis, Kinney, & Matyear, 2000; MacNeilage & Davis, 2000). The resulting syllables are thought to represent the emergence of speech motor control (Moore & Ruark, 1996). If a core deficit in this system is present, the infant's protosyllabary would be necessarily restricted. Moreover, cross-discipline research into vocal motor learning suggests a mechanism whereby impaired speech motor control affects not only the establishment of a set of syllables for later use, but also the *process* of vocal learning (Haesler et al., 2007; Pytte & Suthers, 2000).

As discussed throughout this thesis, limited or restricted babbling is not necessarily specific to CAS. Hearing impairment (David et al., 2002), structural defects affecting the vocal apparatus (Chapman, Hardin-Jones, Schulte, & Halter, 2001; Locke & Pearson, 1990, 1992), and congenital cognitive impairments (Stoel-Gammon, 1997), for example, have been shown to negatively impact the emergence of babbling. Of more direct relevance to the present study, however, is research demonstrating the continuity of prelinguistic vocal development in normal and disordered communication development alike, in the absence of known disorder of the systems listed above (Eilers, Neal, & Oller, 1996; MacNeilage, Davis, & Matyear, 1997; Oller, Eilers, Neal, & Schwartz, 1999; Stoel-Gammon, 1989; Whitehurst, Smith, Fischel, Arnold, & Lonigan, 1991). In Study 1, the children with SLI, although all having been reported as having babbled in infancy, were late to do so. Oller and colleagues documented persistently restricted expressive vocabularies in toddlers who, as infants, had not commenced babbling by 10 months of age (Oller, Eilers, Neal, & Schwartz, 1999). Whether any of these children had features consistent with CAS is unknown; however, this and other research on late talkers suggests a greater role for prelinguistic vocal development than was once acknowledged.

While prelinguistic vocal development may also be restricted in children who are not suspected to have CAS, the source of impairment is presumed to differ to the core speech motor control deficit hypothesised for CAS. It may be that auditory perceptual skills are immature or underdeveloped (Tallal & Stark, 1981). There may be an overall delay in the communication system as a whole, secondary to some neurological immaturity (Beitchman, Hood, Rochon, & Peterson, 1989), affecting both articulatory motor development and development of the conceptualiser and subsequent linguistic processes. In contrast, a more specific deficit in articulatory-motor, or speech motor, control is hypothesised for CAS. Acoustic measures utilised in Study 3, when combined with information from the standardised assessments, supported the viability of this explanation. Mean fundamental frequency, as well as variation in this measure, were unusually high in the canonical syllables of SIB2. Formants 1 and 2 were also highly correlated, suggesting coupling of the articulators and a lack of maturity of the articulatory system. While these results are preliminary and require replication, they are consistent with the notion of impaired underlying speech motor control, and present an important avenue for future research.

Initially Independent Speech Motor and Conceptual Systems

A key tenet of Levelt and colleagues' developmental model (highlighted by Maassen, 2002) is that initially, in infancy, the developing speech motor and conceptual systems are independent of each other (Levelt et al., 1999). This implies that it is possible to have an isolated impairment, at these early stages, in one system, as is proposed for CAS. The results of the present research support this notion. In Study 1, although children with CAS were reported to be less likely to babble, they were not later in the emergence of smiling, which could be reflective of intact pre-conceptual development given its sensitivity to disorders affecting communicative intent (Sabbagh, 1999; Wetherby et al., 2004; Wong, Huia, Lee, & Leung, 2004). Moreover, Study 3, in particular, demonstrated that it is possible for an individual to show dissociated development in these areas, with selectively impaired speech motor control but age-appropriate conceptual development. Caramazza and Coltheart (2006) highlight the importance of individual cases in evaluating theoretical explanations of normal and impaired systems. That one sibling showed this very clear pattern of dissociation in two areas – with selectively impaired speech motor control yet intact conceptual development – presents a strong argument for the plausibility of both the underlying developmental model of speech production and the speech motor control deficit account of CAS. Whether such a deficit applies to each and every clinical case of CAS requires further investigation.

Coupling of the Speech Motor and Conceptual Systems

Levelt and colleagues (Levelt et al., 1999) proposed that first words are produced when the typically developing infant couples previously babbled gestural scores from the protosyllabary with lexical concepts stored in the conceptual system. In the case of a core impairment in speech motor control, the conceptual system is hypothesised to be intact. However, the protosyllabary is restricted, meaning there is a lack of speech motor patterns available for meaningful word production. Thus the emergence of first words would be expected to be delayed in CAS, and the rate of expressive vocabulary expansion would be reduced. Parents of children with sCAS (Study 1) reported first words to emerge significantly later than children with typical speech and language ability. Study 2 also showed delayed word emergence in children with a CAS profile. The infant in Study 3 with early CAS features also

produced first words at a later age than expected in normal development, consistent with anecdotal reports and theoretical hypotheses.

Subsequent 'Linguistic Development'

As discussed throughout the present thesis, speech motor deficit theories of CAS have traditionally been viewed as being inadequate in terms of their ability to account for the varied linguistic impairments typically seen in children with the disorder. Language and literacy difficulties, for example, have previously been seen as incongruent with a core deficit in speech motor control. Such impairments have thus been seen as commonly co-morbid (Lewis, Freebairn, Hansen, Iyengar, & Taylor, 2004; Stackhouse, 1992), rather than the secondary sequelae of a motoric core impairment.

However, the interpretation of a core speech motor control deficit within a developmental framework not only accounts for language and literacy difficulties, but predicts such impairments, *especially* after a period of development. In typically developing infants, Levelt and colleagues (Levelt et al., 1999) suggest that the morphological and phonological encoding systems develop as a consequence of the pressure of a growing vocabulary. The protosyllabary essentially becomes overtaxed, necessitating the dismantling of whole-word gestures into smaller units. Such a pivotal role for lexical growth in subsequent syntactic development (Bates & Goodman, 1997) is supported by observations that children rarely begin to combine words until their expressive vocabulary has exceeded 50 single words (Rescorla, 1989; Rescorla & Alley, 2001).

In contrast, such developments would predictably be delayed in children with CAS, if a core deficit in speech motor control exists. In the present research, children with CAS demonstrated these linguistic deficits. In Study 1, for example, the sCAS group were reported as producing two word combinations significantly later than both children with typical development, *and* those with SLI. Expressive vocabulary at 24 months of age for the infant with a CAS-type profile in Study 3 was 32 single words, significantly below age expectations and presumably too restricted to overtax the protosyllabary and support the establishment of the phonological and morphological

encoding systems. These studies highlight the importance of intact phonetic skill for subsequent linguistic development.

Children with CAS often also present with phonological awareness and literacy difficulties (Hall, 2003c; Lewis et al., 2004; Marion et al., 1993). It is well documented that impoverished phonological representations may underlie such difficulties (American Speech-Language-Hearing Association, 2007). In the developmental context, the establishment of well-specified phonological representations may be reliant on phonetic development (as well as input processes) (Maassen, 2002; Storkel & Morrisette, 2002). Thus, in a child with an initial core deficit in speech motor control, establishment of well-specified phonological representations would be impaired. The pivotal role of speech motor control and early perceptuo-motor development for subsequent phonological development is well supported (McCune & Vihman, 2001; Storkel & Morrisette, 2002; Velleman, 1994). The close interaction between speech motor, phonological and lexical development has been acknowledged (Mitchell, 1995).

Associated Areas of Impairment

As detailed in earlier chapters, whilst the exact phenotype for CAS is still being debated (American Speech-Language-Hearing Association, 2007), a number of commonly-reported features have been identified. These include inconsistency in production, vowel errors, speech sequencing difficulties and prosodic anomalies. While speech motor control explanations of CAS have often been viewed as being unable to account for such divergent characteristics, when viewed developmentally, the features can be accommodated.

The inconsistency observed in children with CAS, whereby multiple productions of the same word are produced differently, may be reflective of an impoverished syllabary and poor phonologic encoding. The protosyllabary described by Levelt and colleagues (Levelt et al., 1999), if restricted in the case of CAS, would subsequently result in a restricted syllabary. The syllabary is said to contain gestural scores for frequently used syllables (Levelt et al., 1999); thus in the individual with CAS, this repository of gestures would be restricted. Moreover, such "highly overlearned gestural patterns…. need not be recomputed time and again" in speakers with intact

abilities (Levelt et al., 1999, p. 5). This would not be the case for speakers with impoverished syllabaries, leading to inconsistency in production. The lexical advantage observed for words over nonwords for typically developing children, but not seen in children with CAS (Thoonen, Maassen, Gabreëls, Schreuder, & de Swart, 1997) may also be an artefact of an impoverished syllabary. Real words may not have had the benefit of repeated accurate production in children with CAS, therefore functioning similarly to phonetically legal nonsense words in repetition tasks.

The vowel errors observed in children with CAS (Davis, Jacks, & Marquardt, 2005) are consistent with a deficit in speech motor control. In the movements underlying babbling, oscillation of the jaw, with initial passive movement of the tongue, is thought to result in the patterns of syllables produced by typically developing children (Davis & MacNeilage, 1995). Back vowels, for example, have been shown to predominantly co-occur with velar consonants, and front vowels with alveolar consonants (MacNeilage, 1998). Restricted vowel development, therefore, may be a consequence of a limited range of articulatory gestures or speech movement patterns. Normal development typically involves a gradual 'uncoupling' of individual articulators (Browman & Goldstein, 1992); in the present study a tighter coupling was suggested for the infant whose profile was most suggestive of CAS.

A similar pattern of gradual uncoupling in movement patterns of the limbs has been observed in normal motor development (Piek, 2002). For example, objective instrumentation demonstrates initial tight coupling of joints in the first stages of learning a skill (Piek & Gasson, 1999). Such tight joint coupling (e.g., in leg joints) is hypothesised to reduce movement complexity, effectively minimising extraneous movements that may inhibit learning of the movement (Piek, 2002). Extended periods of tight coupling between relevant limb joints has been documented for infants at risk of motor impairments (Vaal, van Soest, Hopkins, Sie, & van der Knaap, 2000). If the same principle applies to the movement patterns underlying speech production, tight coupling of the articulators may be inferred in the case of strongly correlated F1 and F2 values as was observed in SIB2 in Study 3.

As described in Chapter 1, the prosodic anomalies observed in CAS have been interpreted in a number of ways. Shriberg and colleagues (Shriberg, Aram, & Kwiatkowski, 1997c), for example, initially interpreted prosodic difficulties in children with CAS as being reflective of a core deficit with linguistic representations. However, such deficits have also been viewed as being more reflective of a deficit with speech motor control (Shriberg et al., 2003). A core deficit in speech motor control, impacting on the ability to develop a protosyllabary and subsequent morpho-phonological encoding, may parsimoniously account for prosodic anomalies. Velleman and Shriberg (1999) demonstrated via metrical analyses that the stress errors observed in children with CAS were similar to those seen in younger typically developing children. Specifically, the presence of a high degree of weak syllable deletion was noted, in both children with CAS and younger typically developing children. Thus children with CAS may not develop the stores and processes required for production of appropriate lexical stress. Moreover, the dissociation in speech motor and conceptual skills may make any weak syllable deletion and stress anomalies more apparent: perhaps the greater length of sentences attempted (but not successfully articulated) in an effort to convey more ideas results in an atypically high degree of syllable deletion. Further research is needed to investigate the nature of prosodic anomalies that are often perceptually apparent (Odell & Shriberg, 2001; Skinder, Strand, & Mignerey, 1999; Velleman & Shriberg, 1999), but not always acoustically evident (Munson, Bjorum, & Windsor, 2003; Skinder et al., 1999), in children with CAS.

Another area of deficit often reported in children with CAS is impaired motor development, particularly fine motor skills (Hall, 2003c). In the present research, motor milestones were significantly correlated with language milestones for the children with sCAS in Study 1. In Study 2, data from only one time-point were available, which did not indicate significant motor delays for children with CAS. However, in Study 3, the infant siblings as a group showed significantly lower fine motor scores even when no individual scored below the normal range.

Although the speech motor control deficit hypothesised as being the core impairment in CAS does not directly predict fine motor difficulties, research demonstrating a link between these two areas of development may account for these findings. Rhythmical hand banging, for example, has shown to co-occur with the emergence of canonical babbling (Ejiri & Masataka, 2001). Anatomically, the neurological substrates underlying both skills are proposed to be similarly located (Locke, 2004; Locke & Pearson, 1990). Whether the effect is neurologically or behaviourally mediated, constraints in fine motor and speech motor development appeared to co-occur in the present research, suggesting the need for additional research to understand this trend.

Strengths and Limitations

The present research appears to be amongst the first to investigate the prelinguistic period in CAS, using a combination of methodologies not yet applied to the disorder. Study 1, in quantifying parental report of the vocalisation behaviours of children with a clinical diagnosis of CAS, gave an important insight into the prelinguistic phase of development in these children, and identified a number of areas for further investigation (i.e., babbling, motor development). The relatively large number of clinical participants, recruited over an extended timeframe, was a particular strength of the preliminary study.

Study 2 was unique in its use of retrospective data available for clinically-ascertained children, allowing the prelinguistic period for children with CAS features at 3 to 4 years to be investigated. Retrospective data designs have been used previously for other developmental disorders such as autism and dyslexia, but have not yet been applied in any published studies on CAS. A secondary strength of this study was the large number of infants whose data were available for comparison. In addition, the study operationally defined the CAS-related characteristics reported in the clinical sample participants. In the absence of a set of validated diagnostic features for the disorder, detailed participant description has been acknowledged as vital (American Speech-Language-Hearing Association, 2007), yet is not often reported for the children studied.

A significant strength of Study 3 was its longitudinal design, using family history as an initial method of recruiting infants potentially 'at-risk' of CAS. To the best of our knowledge, this methodology has not previously been applied to the study of CAS. Investigation of group and individual profiles provides an important first step to objectively document the natural history of the disorder and its potential broader phenotype. Examination of vocalisation data, including the use of acoustic analyses, was another unique contribution to the study of the way in which CAS may manifest in its earliest form. There appear to be no published studies investigating vocalisation and acoustic data of children considered at risk of CAS, and thus Study 3 makes an important contribution in that regard.

Despite the strengths identified above, a number of methodological limitations need to be considered when interpreting the findings of the present research. These relate mostly to each specific study, and while they are highlighted in the relevant chapters, they are also summarised here from an integrated standpoint. As with all research into CAS, the results of the present research are limited by the current lack of a validated set of diagnostic criteria for CAS. The children in Study 1, for example, were identified via clinical means and were not assessed by the researcher. Study 2 applied more detailed techniques to provide more comprehensive participant description than is typically observed in CAS research, but still suffers from the lack of validated criteria that is typically available for other disorders. The results of Study 3, whilst providing important information relevant to the developmental trajectory of CAS, suffer from the limited time-frame available for longitudinal follow-up of the infant siblings, meaning that a specific diagnosis could not be confirmed.

The nature of data utilised in each study also requires consideration. Whilst Study 1 represented an important insight into the prelinguistic vocalisations of children with CAS and SLI, it was based on how parents recalled this information, and thus was dependent on the reliability of the parents' recall. Study 2 had the benefit of objective infant data, collected prospectively, but as these data were not originally designed for the present research, they contained limitations that restricted the scope of the hypotheses. The third study contained important longitudinal data collected solely for the present research. However, the comparison of similarly prelinguistic canonical

syllables, produced at different ages by the infants, coupled with the inability to confirm a CAS diagnosis in the toddler age, is an important limitation for the present research. Future follow up of the infants involved will allow confirmation of the infants' developmental status. Investigation of patterns of connected speech in comparison to the early prelinguistic samples may provide an important insight into potential similarities in acoustic patterns.

Reflecting the preliminary nature of investigations of the prelinguistic period in this population, the large number of statistical analyses performed within the data sets brings with it a threat of an inflated rate of Type 1 errors in interpreting the data (Tabachnick & Fidell, 2001). However, as hypotheses were theory-driven, and to protect against the risk of making a Type 2 error, interpretation of analyses proceeded at the standard per-test alpha level of .05, except where post hoc comparisons were made. For the single case comparison, conclusions were based on patterns of extremeness unlikely to be Type 1 errors (Crawford & Garthwaite, 2002). Despite these factors, further research is required to confirm patterns observed in the present data.

Moreover, although the present research appropriately utilised single case methodology where participant numbers were small and individuals' profiles were of importance, such designs suffer from limitations in generalising the results (Caramazza, 1986). The results observed in the present study therefore require replication in order to establish their application in explaining the broader CAS population. However, the observed patterns support the viability of the hypothesised speech motor control deficit as a possible explanation for CAS. Whether such a deficit accounts for each and every clinical case of CAS is not clear from the present research, and larger longitudinal group studies are required to investigate this issue. Replication of the dissociation observed in the infant in Study 3 showing a CAS-type profile will be important in evaluating the generalisability of the results.

Theoretical Implications

The results of the present research support the notion of a core deficit in speech motor control underlying CAS. They also provide support for a model of early communication development which proposes the initial independence of conceptual and speech motor systems in infancy (Levelt et al., 1999). In this account, such independence in systems is short-lived, with first words emerging from the coupling of the two systems. Thus, a clear cascading effect of a deficit at one level is predicted, affecting subsequent development of linguistic systems.

This notion is consistent with dynamic systems theory, whereby developmental domains are interactive (Mitchell, 1995), but it adds an important qualifier relating to the timing of such interactivity. Applied to movement patterns, dynamic systems theory presumes interaction between the organism and the environment, as well as within the individual (Thelen, 1981). The present research is consistent with the interactive nature of development, especially over time. However, it also suggests that, as with Levelt's model, a form of 'modularity' exists within the speech/language domain, in the form of initially dissociated conceptual and speech motor systems that have the potential to be selectively impaired.

Although the results were broadly consistent with the notion of impaired speech motor control being a viable explanation of the core deficit in CAS, they do not isolate the biological explanation for such a deficit. The children involved in the present research did not present with frank disorders that would give rise to dysarthria, nor did they present with oral musculature features consistent with an idiopathic form of the disorder. It has been noted that CAS and dysarthria often co-occur (American Speech-Language-Hearing Association, 2007), however. Future research is needed to explore the overlap in disorders of speech motor control and to further delineate the processes and systems involved. Moreover, a core deficit in speech motor control may arise (or be present) for a number of reasons. One commonly held view is that children with CAS have subtle, but as yet undetectable, abnormalities in aspects of the brain (American Speech-Language-Hearing Association, 2007; Strand, 1992). It remains for future research to examine putative biological factors underlying the disorder.

Furthermore, a relatively broad conceptualisation of deficient speech motor ability was considered in the present study. As introduced in the opening chapter, the term speech motor control refers to the processes and systems involved in transforming a phonologic representation of language into an acoustic signal (Kent, 2000). From a developmental perspective, the core deficit in CAS is hypothesised as a deficit in articulo-motor, or speech motor control, affecting perceptuo-motor learning. Current research using computational neural modelling techniques is attempting to isolate the nature of such a deficit (Maassen & Terband, 2008). Investigations with the DIVA model, for example, have shown that poor feedforward control (consisting of unstable commands for producing speech sounds), possibly arising from either degraded oral sensitivity and/or altered levels of neural noise (Maassen & Terband, 2008), simulates some of the key characteristics of CAS. Thus, although the present research demonstrates the viability of a core speech motor control deficit (as opposed to a deficit originating purely in the linguistic system), it does not allow investigation of potential pathways to this deficit.

The present research supports the utility of developmental perspectives for studying developmental disorders such as CAS (Hulme & Snowling, 2009). As highlighted by Bishop (1997) and Karmiloff-Smith and colleagues (Karmiloff-Smith, 1998; Karmiloff-Smith, 1999; Karmiloff-Smith et al., 2003; Karmiloff-Smith, Scerif, & Thomas, 2002), much of the heterogeneity observed in children with various developmental disorders may be the result of the unfolding and interactive nature of development. Children with the same underlying core deficit may present with varying features over time, making it a challenging task to identify diagnostic criteria that are inclusive enough to account for individual difference yet specific enough to clearly identify instances of the disorder. Models that include mechanisms for change over time are clearly vital in understanding the dynamic nature of speech and language development.

Clinical Implications

The results from the present studies suggest a number of clinical implications relating to the diagnosis, early identification and treatment of CAS. Although the thesis focussed specifically on CAS, broader implications for developmental speech and language impairments in general are also apparent.

Diagnosis and definition of CAS. Although the present research did not aim to identify a set of differentially diagnostic features for CAS, it does contribute to discussion about such features. Studies 1 and 2 identified a number of clinical characteristics in children with sCAS. Not every child considered to have CAS displayed every CAS-related feature, however. This is consistent with previous research and the current lack of validated differentially diagnostic criteria (American Speech-Language-Hearing Association, 2007; Peter & Stoel-Gammon, 2008). In phase one of Study 2, CAS-related characteristics were operationally defined in order to determine presence or absence of each feature. As outlined in Chapter 3, such detail is often lacking from many studies of CAS. In the absence of validated differentially diagnostic features, future research should similarly provide detailed description of how such features are measured and identified, to allow greater consistency across researchers.

A core deficit originating in lower-level speech motor control, affecting the ability to develop a protosyllabary and restricting subsequent language acquisition, has implications for the definition and description of CAS as a diagnostic category. It supports the definition of CAS proposed by ASHA, particularly the focus on speech *movements* and *movement sequences*. Although there are many associated features, the nature of the core deficit may initially be isolated to lower-level speech motor control ability.

The present research, consistent with Maassen's (2002) proposal for the need to interpret CAS in a developmental framework, goes some way to providing an explanation for the inconsistent findings relating to CAS when it is researched in children. Specifically, the interactive nature of development and cascading effects of a deficit at one level of the system on subsequent phonetic, phonological and

linguistic development accommodates the varied findings reported in the literature. As discussed previously, researchers have reported children with CAS to show deficits in aspects of speech motor control, language skills, speech sequencing, phonological awareness and literacy, and even perception. Models of early language development and perspectives on the interactive nature of development *predict* varied additional deficits *especially* after a period of development. This suggests that the best time to identify a core deficit in speech motor control is much earlier than our knowledge and tools currently allow.

Early identification. Mirroring research in other developmental disorders (Landa & Garrett-Mayer, 2006; Wetherby et al., 2004), there is great interest in the early identification of speech and language disorders, including CAS (Reilly et al., 2007). Factors contributing to this interest include legislative issues, with a focus on early identification, increased consumer awareness, as well as issues relating to the high heritability of speech and language issues in general. The present research contributes to discussion in the early identification area.

Finding anomalies in pre-linguistic vocalisations (by parent report – Study 1, and by inspection of retrospective infant data- Study 2) for some children who have clinical diagnoses of sCAS suggests the possibility of identifying infants who may be at increased 'risk' of CAS. Furthermore, a particular pattern of impairment, with intact conceptual development and age-appropriate receptive language skills in the context of atypical vocalisation development, may be more suggestive of a motor-planning type of speech deficit. It remains not yet possible to diagnose CAS in infants or toddlers (Davis & Velleman, 2000). The absence of canonical babbling by 10 months of age, however, should indicate the need for careful observation (Oller et al., 1999). Presuming audiological problems are ruled out, infants who are not producing canonical syllables by this age may be at increased risk of speech and language delays. Further research is needed to establish the role of early vocalisation and language measures for predicting specific patterns of later impairment.

A number of tools suitable for screening were used in the present investigations. The infant in Study 3 who showed a profile consistent with early CAS features failed the WILSTAAR screen, the communication subtest of the ASQ, and the Speech composite of the CSBS Infant Toddler checklist at 9 months of age. However, all siblings in Study 3 failed the WILSTAAR screen, as did all infants in Study 2, suggesting a lack of specificity of this tool. In contrast, only one sibling failed the ASQ communication subtest – the one who later showed CAS characteristics. This same infant also failed the speech composite of the CSBS infant toddler checklist, supporting the sensitivity of the tool (although he was not the only child to fail on the checklist – two other infants who evidenced age-appropriate speech and language skills at 2 years of age also failed on the checklist). These results suggest that tools such as the ASQ and CSBS infant toddler checklist, readily available and time-efficient, may be useful in the monitoring of large samples of infants, or more specific monitoring of those genetically at increased risk. Screening siblings of children with CAS, or speech and language disorders in general, for example, may be indicated.

The notion of early identification often implies the possibility of early intervention. However, even once early identification is possible, further research would be needed to establish the effectiveness or otherwise of specific intervention approaches. Early screening also brings with it ethical issues regarding increasing parental concern. These issues are beyond the scope of the present thesis, but remain important areas for consideration for future research. Implications for treatment approaches, however, are indicated, based on the proposed core deficit originating in speech motor control.

Treatment approaches. Despite acknowledgement of the negative consequences of CAS for the child, family and community (American Speech-Language-Hearing Association, 2007), relatively few research studies have specifically investigated the effectiveness of treatment approaches for the disorder. Children with CAS have often been described as being 'resistant' or slower to respond to therapy. Of the small number of treatment studies that have been reported to date, research has evaluated the effectiveness of specific techniques to improve speech production (e.g., integral stimulation, Strand & Debertine, 2000),

augmentative and alternative communication systems (e.g., Cumley & Swanson, 1999), and phonological awareness training (Moriarty & Gillon, 2006). Additional treatment approaches have been described but not yet evaluated (Crary & Towne, 1984; Velleman, 2002).

The present research provides support for techniques that focus on remediation of the core deficit in speech production, as well as for limiting the negative effects on vocabulary acquisition and expansion, development of syntax, and subsequent phonological awareness. Given the dissociation observed, with intact conceptual development, the results also highlight the importance of recognising potential strengths in areas such as communicative intent and gesture use, and the potential difficulties (such as frustration) that may arise with such mis-matched skills.

Hypothetically, if it was possible to identify CAS in infancy, treatment targeting a core deficit in speech motor control may focus on enhancing the opportunities for production and expansion of canonical syllables and vocalisations. Given the theoretical and anatomical suggestion of the co-occurrence of hand-banging and other rhythmic movements with canonical babbling, encouraging such movements (e.g., providing ample opportunities for shaking rattles) may theoretically help to entrain vocal production. Such rhythmical movements are hypothesised to bridge the gap between uncoordinated and coordinated movement (Mitchell, 1995; Thelen, 1981). A team approach, including input from Speech Pathologists, Occupational Therapists and/or Physiotherapists may also facilitate optimal progress (Hall, 2003d; Hodge, 2003).

Given the relative strength in conceptual skills, strategies to utilise these skills and limit the negative effects of the speech motor control deficit may be indicated. Extra focus on imitating vocalisations may be indicated, in the context of enjoyable play with a familiar care-giver, in light of research supporting a role for mirror neurons (Corballis, 2004; Ito, 2004; Westermann & Miranda, 2004). This may include the encouragement of symbolic noise (e.g., animal and transport noise), to attempt to increase the child's phonemic inventory. Encouraging word production within the child's current phonemic repertoire, however restricted this may be, is a technique suitable for the early linguistic period. A child with only /b/ in their inventory, for

example, may be encouraged to use words such as 'ball', 'byebye', 'boo', and 'baby', or approximations of them. Moreover, encouraging the use of gestural or other augmentative communication devices may be appropriate, especially to reduce frustration and encourage the continual expansion of (non-verbal) language skills. Although clinicians may use approaches such as that described above for children with CAS features, research is needed to objectively evaluate the benefit and efficacy of each component, and/or combinational therapies.

Future Directions

Despite much interest in the disorder, there are many aspects of CAS that remain poorly understood (American Speech-Language-Hearing Association, 2007). A number of areas for future research are suggested from the results of the present investigations. Future research should continue to explore the phenotype and diagnostic criteria for CAS, for research and clinical applications. The present research (Study 2) operationally defined features commonly reported in CAS, for the purpose of increased detail in participant description. However, alternative methods for measuring and defining the characteristics may be indicated, and those features showing diagnostic promise require validation.

The differences in early development reported retrospectively by parents of children with sCAS may be more meaningful if they could be compared to that of children with non-apraxic phonological disorder. As pointed out in Chapter 2, it was not clear whether the way in which children with sCAS were reported (i.e., as being less likely to babble, later in the emergence of two word combinations, for example) was influenced by their current (persistent) speech production deficits. Future research should therefore compare the early development of children with both CAS and phonological disorder, including children whose earlier speech output deficits have essentially resolved.

Observation of general trends for weaker communication and fine motor ability in siblings of children with CAS suggests the need for further investigation of siblings of children with CAS. Although only 2 of the 8 infant siblings showed atypical

speech and language development by 2 years of age, and only one of these showed CAS-related features, the results support the need for further research into the familial aggregation of CAS.

Moreover, longitudinal research investigating CAS should continue, focussing on 'at-risk' samples such as the one described herein. The relatively late age that CAS can be diagnosed in children (coupled with its relative infrequency) is likely to have contributed the lack of such studies from an early age (American Speech-Language-Hearing Association, 2007). However, applying paradigms utilised in the dyslexia and autism literature is one way to address the lack of studies into the CAS. Within the present data set, continuing longitudinal investigation of the infants is planned. It will be informative to evaluate the children's profiles over time. In addition, further analysis of the vocalisation samples may provide more information regarding the nature of the proposed speech motor control deficit. Exploring vocalisations at later timepoints, for example, may highlight whether the acoustic anomalies observed for SIB2 are a persistent feature of this child's speech.

Replication of the methodology applied in Study 3, with larger numbers and over a longer period of development would be an important avenue for future research into both CAS and speech and language disorders in general. The utilisation of kinematic measurement of articulator movement may provide further information to complement the use of acoustic analysis in investigating the nature of prelinguistic vocalisations. This type of measurement has been applied to children from 12 months of age (Green, Moore, Higashakawa, & Steeve, 2000), but appears not to have been applied to the study of putatively 'at risk' siblings of children with CAS.

Conclusion

The present research employed a combination of methodologies to investigate the core deficit in CAS. Results were consistent with a core deficit in speech motor control, affecting perceptuo-motor learning and having negative effects for subsequent linguistic development. As highlighted in the opening quote to this chapter, speech production in typically developing children becomes a highly practiced motor skill. For children with CAS, this high degree of practice and

resulting precision does not appear to feature in their early prelinguistic development. In a climate where speech and language skill has been recognised as a clear determinant contributing to the future health and wellbeing of individuals and communities (Anderson et al., 2003), identifying the core deficit in CAS as early as possible in development is of vital importance.

The results of the present investigation, whilst preliminary given their basis on single cases, supported the notion of a core deficit in speech motor control, evident in prelinguistic vocalisations and in the context of intact conceptual development. Such a core deficit, when interpreted developmentally, predicts the range of impairments that are often observed in children with CAS. Such an account should provide a foundation for research into CAS and other speech disorders alike.

REFERENCES

Alcock, K. J., Passingham, R. E., Watkins, K., & Vargha-Khadem, F. (2000a). Oral dyspraxia in inherited speech and language impairment and acquired dysphasia. [Science Direct]. *Brain and Language, 75*, 17-33.

Alcock, K. J., Passingham, R. E., Watkins, K., & Vargha-Khadem, F. (2000b). Pitch and Timing Abilities in Inherited Speech and Language Impairment. *Brain and Language, 75*(1), 34-46. Retrieved: August 6, 2004, from Elsiever database.

American Speech-Language Hearing Association (2006). *Childhood Apraxia of Speech (Draft Technical Report).*

American Speech-Language-Hearing Association. (2007). *Childhood Apraxia of Speech (Technical Report)*. Available from www.asha.org/policy

Anderson, L. M., Shinn, C., Fullilove, M. T., Scrimshaw, S. C., Fielding, J. E., Normand, J., et al. (2003). The effectiveness of early childhood development programs: A systematic review. *American Journal of Preventative Medicine, 24*(3), 32-46.

Archibald, L. M. D., & Alloway, T. P. (2008). Comparing language profiles: children with specific language impairment and developmental coordination disorder. [Informa Healthcare]. *International Journal of Language & Communication Disorders, 43*(2), 165-180.

Bahr, R. H. (2005). Differential diagnosis of severe speech disorders using speech gestures. *Topics in Language Disorders, 25*(3), 254-265.

Bahr, R. H., Velleman, S. L., & Ziegler, M. A. (1999). Meeting the challenge of suspected developmental apraxia of speech through inclusion. [ProQuest 5000]. *Topics in Language Disorders, 19*(3), 19-35.

Ball, M., & Gibbon, F. (2002). *Vowel Disorders*. Boston: Butterworth-Heinemann.

Betz, S., & Stoel-Gammon, C. (2005). Measuring articulatory error consistency in children with developmental apraxia of speech. *Clinical Linguistics & Phonetics, 19*(1), 53-66.

Bishop, D. V. M. (1997). Cognitive neuropsychology and developmental disorders: Uncomfortable bedfellows. *The Quarterly Journal of Experimental Psychology, 50*(4), 899-923.

Bishop, D. V. M. (2002). Motor immaturity and specific speech and language impairment: Evidence for a common genetic basis. *American Journal of Medical Genetics, 114*, 56-63.

Boersma, P., & Weeink, D. (2002). PRAAT: doing phonetics by computer (Version 4.0.28). Amsterdam: Institute of Phonetic Sciences, University of Amsterdam.

Boyce, B. A., & Poteat, G. M. (2005). Review - Ages and stages questionnaires: a parent-completed child-monitoring system. *Mental Measurements Yearbook, 16*.

Bradford, A., & Dodd, B. (1996). Do all speech-disordered children have motor deficits? *Clinical Linguistics & Phonetics, 10*(2), 77-101.

Bricker, D., Squires, J., Mounts, L., Potter, L., Nickel, R., Twombly, E., et al. (1999). *Ages and stages questionnaire: User's guide* (2nd ed.). Baltimore: Paul H Brooks Publishing Company.

Bridgeman, E., & Snowling, M. (1988). The perception of phoneme sequence: A comparison of dyspraxic and normal children. *International Journal of Language & Communication Disorders, 23*(3), 245 - 252.

Broomfield, J., & Dodd, B. (2004). The nature of referred subtypes of primary speech disability. [ProQuest 5000]. *Child Language Teaching and Therapy, 20*(2), 135-151.

Browman, C. P., & Goldstein, L. (1992). Articulatory phonology: an overview. *Phonetica, 49*(3-4), 155-180.

Bryson, S., Zwaigenbaum, L., Brian, J., Roberts, W., Szatmari, P., Rombough, V., et al. (2007). A Prospective Case Series of High-risk Infants who Developed Autism. *Journal of Autism and Developmental Disorders, 37*(1), 12-24.

Bzoch, K. R., & League, R. (1991). *Receptive-expressive emergent language scale* (2nd ed.). Texas: Pro-Ed.

Bzoch, K. R., League, R., & Brown, V. (2003). Receptive-Expressive Emergent Language Test (3rd ed.). Austin, Texas: PRO-ED.

Caramazza, A. (1986). On drawing inferences about the structure of normal cognitive systems from the analysis of patterns of impaired performance: The case for single-patient studies. *Brain and Cognition, 5*, 41-66.

Chiat, S., & Roy, P. (2007). The preschool repetition test: an evaluation of performance in typically developing and clinically referred children. [ProQuest 5000]. *Journal of Speech, Language and Hearing Research 50*(2), 429-443.

Coonrod, E. E., & Stone, W. L. (2004). Early Concerns of Parents of Children With Autistic and Nonautistic Disorders. [ProQuest 5000]. *Infants and Young Children, 17*(3), 258-268.

Corballis, M. C. (2004). FOXP2 and the mirror system. [Science Direct]. *Trends in Cognitive Sciences, 8*(3), 95-96.

Crary, M. A. (1993). *Developmental Motor Speech Disorders*. San Diego: Singular Publishing Group.

Crary, M. A., & Towne, R. L. (1984). The asynergistic nature of developmental verbal dyspraxia. *Australian Journal of Human Communication Disorders, 12*(2), 27-28.

Crawford, J. R., & Garthwaite, P. H. (2002). Investigation of the single case in neuropsychology: confidence limits on the abnormality of test scores and test score differences. *Neuropsychologia, 40*, 1196-1208.

Crawford, J. R., & Garthwaite, P. H. (2005). Testing for suspected impairments and dissociations in neuropsychology: Evaluation of alternatives using Monte Carlo simulations and revised test for dissociations. *Neuropsychology, 19*(3), 318-331.

Crawford, J. R., Garthwaite, P. H., & Gray, C. D. (2003). Wanted: Fully operational definitions of dissociations in single-case studies. *Cortex, 39*, 357-370.

Crawford, J. R., Garthwaite, P. H., Howell, D. C., & Gray, C. D. (2004). Inferential methods for comparing a single case with a control sample: Modified t-tests versus mycroft et al.'s (2002) modified ANOVA. *Cognitive Neuropsychology, 21*(7), 750-755.

Crawford, J. R., & Howell, D. C. (1998). Comparing an individual's test score against norms derived from small samples. *The Clinical Neuropsychologist, 12*(4), 482-486.

Crawford, J. R., Howell, D. C., & Garthwaite, P. H. (1998). Payne and Jones revisited: Estimating the abnormality of test score differences using a modified paired samples t test. *Journal of Clinical and Experimental Neuropsychology, 20*(6), 898-905.

Darrah, J., Hodge, M., Magill-Evans, J., & Kembhavi, G. (2003). Stability of serial assessments of motor and communication abilities in typically developing infants--implications for screening. [Science Direct]. *Early Human Development, 72*(2), 97-110.

Davis, B., Jakielski, K. J., & Marquardt, T. P. (1998). Developmental apraxia of speech: determiners of differential diagnosis. *Clinical Linguistics & Phonetics, 12*(1), 25-45.

Davis, B. L., Jacks, A., & Marquardt, T. P. (2005). Vowel patterns in developmental apraxia of speech: three longitudinal case studies. [Stretswise]. *Clinical Linguistics & Phonetics, 19*(4), 249-274.

Davis, B. L., & MacNeilage, P. F. (1995). The articulatory basis of babbling. *Journal of Speech & Hearing Research., 38*, 1199-1211.

Davis, B. L., & Velleman, S. L. (2000). Differential diagnosis and treatment of developmental apraxia of speech in infants and toddlers. *Infant-Toddler Intervention, 10*(3), 177-192.

Dawson, G., Osterling, J., Meltzoff, A. N., & Kuhl, P. (2000). Case Study of the Development of an Infant with Autism from Birth to Two Years of Age. [Science Direct]. *Journal of Applied Developmental Psychology, 21*(3), 299-313.

Dell, G., Schwartz, M. F., Martin, N., Saffran, E. M., & Gagnon, D. A. (1997). Lexical access in aphasic and nonaphasic speakers. *Psychological Review, 104*(4), 801-838.

Dewey, D. (1995). What Is Developmental Dyspraxia. [Science Direct]. *Brain and Cognition, 29*(3), 254-274.

Dodd, B. (1995). *Differential diagnosis and treatment of children with speech disorder*. London: Whurr Publishers.

Dodd, B. (2007). Evidence-Based Practice and Speech-Language Pathology: Strengths, weaknesses, ppportunities and threats. *Folia Phoniatrica et Logopaedica, 59*(3), 118-129.

Dodd, B., Hua, Z., Crosbie, S., Holm, A., & Ozanne, A. (2002). *Diagnostic Evaluation of Articulation and Phonology*. London: The Psychological Corporation.

Dodd, B., & McIntosh, B. (2008). The input processing, cognitive linguistic and oro-motor skills of children with speech difficulty. *International Journal of Speech-Language Pathology, 10*, 169-178.

Duffy, J. R. (2002). Apraxia of speech: Historical overview and clinical manifestations of the acquired and developmental forms. In L. Shriberg & T. Campbell (Eds.), *Proceedings of the 2002 Childhood Apraxia of Speech Research Symposium* (pp. 3-12). Arizona: The Hendrix Foundation.

Ejiri, K., & Masataka, N. (2001). Co-occurence of preverbal vocal behavior and motor action in early infancy. *Developmental Science, 4*(1), 40-48.

Ekelman, B. L., & Aram, D. (1983). Syntactic findings in developmental verbal apraxia. *Journal of Communication Disorders, 16*, 237-250.

Ertmer, D. J., & Mellon, J. A. (2001). Beginning to talk at 20 months: early vocal development in a young cochlear implant recipient. [ProQuest 5000]. *Journal of Speech, Language and Hearing Research, 44*(1), 192-206.

Ertmer, D. J., Young, N. M., Grohne, K., Mellon, J. A., Johnson, C., Corbett, K., et al. (2002). Vocal development in young children with cochlear implants: Profiles and implications for intervention. *Language, Speech & Hearing Services in Schools, 33*, 184-195.

Evans, J. L. (2001). An emergent account of language impairments in children with SLI: implications for assessment and intervention. [Science Direct]. *Journal of Communication Disorders, 34*(1-2), 39-54.

Fenson, L., Dale, P. S., Reznick, S., Thal, D., Bates, E., Hartung, J. P., et al. (1993). MacArthur Communicative Development Inventories. San Diego, CA: Singular Publishing Group.

Forrest, K. (2003). Diagnostic criteria of developmental apraxia of speech used by clinical speech-language pathologists. [ProQuest 5000]. *American Journal of Speech - Language Pathology, 12*(3), 376-380.

Forrest, K., & Morrisette, M. L. (1999). Feature analysis of segmental errors in children with phonological disorders. [Proquest 5000]. *Journal of Speech, Language, and Hearing Research, 42*(1), 187-194.

Gathercole, S., & Baddeley, A. (1990). Phonological memory deficits in language disordered children: Is there a causal connection? *Journal of Memory and Language, 29*(3), 336-360.

Green, J. R., Moore, C. A., Higashakawa, M., & Steeve, R. W. (2000). The physiological development of speech motor control: lip and jaw coordination. *Journal of Speech, Language and Hearing Research, 43*(1), 239-255.

Goodgold-Edwards, S., & Cermak, S. (1990). Integrating motor control and motor learning concepts with neuropsychological perspectives on apraxia and developmental dyspraxia. *American Journal of Occupational Therapy, 44*(5), 431-439.

Groenen, P., Maassen, B., Crul, T., & Thoonen, G. (1996). The specific relation between perception and production errors for place of articulation in developmental apraxia of speech. *Journal of Speech and Hearing Research, 39*, 468-482.

Guenther, F., H. (1995). Speech sound acquisition, coarticulation, and rate effects in a neural network model of speech production. *Psychological Review, 102*(3), 594-621.

Guenther, F. H. (2006). Cortical interactions underlying the production of speech sounds. [Science Direct]. *Journal of Communication Disorders, 39*(5), 350-365.

Guenther, F. H., Ghosh, S. S., & Tourville, J. A. (2006). Neural modeling and imaging of the cortical interactions underlying syllable production. [Science Direct]. *Brain and Language, 96*(3), 280-301.

Guenther, F., H, Hampson, M., & Johnson, D. (1998). A theoretical investigation of reference frames for the planning of speech movements. *Psychological Review, 105*(4), 611-633.

Guyette, T., & Diedrich, W. M. (1981). A critical review of developmental apraxia of speech. In N. J. Lass (Ed.), *Speech and Language: Advances in Basic Research and Practice* (Vol. 5, pp. 1-49). New York: Academic Press.

Haesler, S., Rochefort, C., Georgi, B., Licznerski, P., Osten, P., & Scharff, C. (2007). Incomplete and inaccurate vocal imitation after knockdown of FoxP2 in songbird basal ganglia nucleus area X. *PLoS Biology, 5*(12), 1-13.

Hall, P. K. (2003a). A letter to the parent(s) of a child with developmental apraxia of speech: Part I: Speech characteristics of the disorder. [ProQuest 5000]. *Language, Speech & Hearing Services in Schools, 31*(2), 169-172.

Hall, P. K. (2003c). A letter to the parent(s) of a child with developmental apraxia of speech: Part III: Other problems often associated with the disorder. [ProQuest 5000]. *Language, Speech & Hearing Services in Schools, 31*(2), 176-179.

Hall, P. K. (2003d). A letter to the parent(s) of a child with developmental apraxia of speech: Part IV: Treatment of DAS. [ProQuest 5000]. *Language, Speech & Hearing Services in Schools, 31*(2), 179-182.

Hay, L. (1984). Discontinuity in the development of motor control in children. In W. Prinz & A. F. Sanders (Eds.), *Cognition and motor processes* (pp. 351-360). Berlin: Springer-Verlag.

Highman, C., Hennessey, N. W., Sherwood, M., & Leitão, S. (2008). Retrospective parent report of early vocal behaviours in children with suspected Childhood Apraxia of Speech (sCAS). *Child Language Teaching and Therapy, 24(3),* 285-306.

Hill, E. L. (2001). Non-specific nature of specific language impairment: a review of the literature with regard to concomittant motor impairments. [StretsWise]. *International Journal of Language & Communication Disorders, 36*(2), 149-171.

Hodge, M. (1994). Assessment of children with developmental apraxia of speech: a rationale. *Clinics in Communication Disorders, 4*(2), 91-101.

Hodge, M. (2003). *Clinical classification of children with developmental speech disorders: A transdisciplinary perspective.* Paper presented at the 2002 Childhood Apraxia of Speech Research Symposium, Arizona.

Hodge, M., & Hancock, H. R. (1994). Assessment of children with developmental apraxia of speech: a procedure. *Clinics in Communication Disorders, 4*(2), 102-118.

Horner, T. (1988). Single versus repeated assessments of infant abilities using the Baylee Scales of Infant Development. *Infant Mental Health Journal, 9*(3), 209-217.

Hulme, C., & Snowling, M. J. (2009). Understanding developmental cognitive disorders. In *Developmental Disorders of Language Learning and Cognition* (pp. 1-36). West Sussex: Wiley-Blackwell.

Hurford, D. P., & Stutman, G. (2004). *The 16th Mental Measurements Yearbook* (Vol. 16). Lincoln: The Buros Institute of Mental Measurements.

Impara, J. C., & Plake, B. S. (1998). *The thirteenth mental measurements yearbook*. Lincoln: Buros Institute of Mental Measurements.

Ito, M. (2004). 'Nurturing the brain' as an emerging research field involving child neurology. [Science Direct]. *Brain and Development, 26*(7), 429-433.

Iverson, J. M., & Wozniak, R. H. (2007). Variation in vocal-motor development in infant siblings of children with autism. [ProQuest 5000]. *Journal of Autism and Developmental Disorders 37*(1), 158 - 170.

Jacks, A., Marquardt, T. P., & Davis, B. L. (2006). Consonant and syllable structure patterns in childhood apraxia of speech: Developmental change in three children. [Science Direct]. *Journal of Communication Disorders, 39*(6), 424-441.

Karmiloff-Smith, A. (1998). Development itself is the key to understanding developmental disorders. [Science Direct]. *Trends in Cognitive Sciences, 2*(10), 389-398.

Karmiloff-Smith, A. (1999). Taking development seriously. *Human Development, 42*(6), 325-327.

Karmiloff-Smith, A., Scerif, G., & Ansari, D. (2003). Double dissociations in developmental disorders? Theoretically misconceived, empirically dubious. *Cortex, 39*(1), 161-163.

Karmiloff-Smith, A., Scerif, G., & Thomas, M. (2002). Different approaches to relating genotype to phenotype in developmental disorders. [Web of Science]. *Developmental Psychobiology, 40*(3), 311-322.

Kent, R. D. (1976). Anatomical and neuromuscular maturation of the speech mechanism: Evidence from acoustic studies. *Journal of Speech and Hearing Research, 19*, 421-447.

Kent, R. D. (2000). Research on speech motor control and its disorders: A review and prospective. [Science Direct]. *Journal of Communication Disorders, 33*(5), 391-428.

Kent, R. D., & Murray, A. D. (1982). Acoustic features of infant vocalic utterances at 3, 6, and 9 months. *Journal of the Acoustical Society of America, 72*(2), 353-365.

Kent, R. D., Mitchell, P. R., & Sancier, M. (1991). Evidence and role of rhythmic organization in early vocal development in human infants. In J. Fagard & P. H. Wolff (Eds.), *The Development of Timing Control and Temporal Organization in Coordinated Action* (pp. 135-149). Amsterdam: North-Holland.

Koster, C., Been, P. H., Krikhaar, E. M., Zwarts, F., Diepstra, H., & Leeuwen, T. V. (2005). Differences at 17 months: productive language patterns in infants at familial risk for dyslexia and typically developing infants. [ProQuest]. *Journal of Speech, Language, and Hearing Research, 48*(2), 426- 438.

Landa, R., & Garrett-Mayer, E. (2006). Development in infants with autism spectrum disorders: a prospective study. *Journal of Child Psychology and Psychiatry, 47*(6), 629-638.

Leitão, S., & Fletcher, J. (2004). Literacy outcomes for students with speech impairment: long-term follow-up. [Swetswise]. *International Journal of Language & Communication Disorders, 39*(2), 245-256.

Leonard, L., McGregor, K., & Allen, G. (1992). Grammatical morphology and speech perception in children with specific language impairment. *Journal of Speech, Language and Hearing Research, 35*, 1076-1085.

Levelt, W. J. M. (1989). *Speaking: From intention to articulation*. Cambridge: MIT Press.

Levelt, W. J. M., Roelofs, A., & Meyer, A. S. (1999). A theory of lexical access in speech production. [StretsWise]. *Behavioral and Brain Sciences, 22*, 1-75.

Lewis, B. A., Freebairn, L. A., Hansen, A., Taylor, G. H., Iyengar, S., & Shriberg, L. D. (2004). Family pedigrees of children with suspected childhood apraxia of speech. [Science Direct]. *Journal of Communication Disorders, 37*(2), 157-175.

Lewis, B. A., Freebairn, L. A., Hansen, A. J., Iyengar, S. K., & Taylor, H. G. (2004). School-Age Follow-Up of Children With Childhood Apraxia of Speech. [ProQuest 5000]. *Language, Speech & Hearing Services in Schools, 35*(2), 122-140.

Locke, J. L. (2002). Vocal development in the human infant: Functions and phonetics. In F. Windsor, M. L. Kelly & N. Hewlett (Eds.), *Investigations in Clinical Phonetics and Linguistics* (pp. 243-256). New Jersey: Lawrence Erlbaum Associates.

Locke, J. L. (2004). How do infants come to control the organs of speech? In B. Maassen, R. D. Kent, H. Peters, P. van Lieshout & W. Hulstijn (Eds.), *Speech Motor Control in Normal and Disordered Speech* (pp. 175-190). Oxford: Oxford University Press.

Locke, J. L., & Pearson, D. M. (1990). Linguistic significance of babbling: evidence from a tracheostomized infant. *Journal of Child Language, 1990*, 1-16.

Locke, J. L., & Pearson, D. M. (1992). Vocal learning and the emergence of phonological capacity. In C. A. Ferguson, L. Menn & C. Stoel-Gammon (Eds.), *Phonological Development: Models, Research, Implications* (pp. 92-129). Maryland: York Press.

Lyytinen, H., Ahonen, T., Eklund, K., Guttorm, T. K., Laakso, M. L., Leinonen, S., et al. (2001). Developmental pathways of children with and without familial risk for dyslexia during the first years of life. *Developmental Neuropsychology, 20*(2), 535-254.

Maassen, B. (2002). Issues contrasting adult acquired versus developmental apraxia of speech. *Seminars in Speech and Language, 23*(4), 257-266.

Maassen, B. (2003). *Cognitive neuropsychological approaches in speech motor disorders in children.* Paper presented at the 2002 Childhood Apraxia of Speech Research Symposium, Arizona.

Maassen, B., Groenen, P., & Crul, T. (2003). Auditory and phonetic perception of vowels in children with apraxic speech disorders. *Clinical Linguistics & Phonetics, 17*(6), 447-467.

Maassen, B., & Terband, H. (2008). *Computational neural modeling of feedback control in Childhood Apraxia of Speech (CAS)*. Paper presented at the 12th Meeting of the International Clinical Linguistics and Phonetics Association.

MacDermot, K. D., Bonora, E., Sykes, N., Coupe, A. M., Lai, C. S. L., Vernes, S. C., et al. (2005). Identification of FOXP2 truncation as a novel cause of developmental speech and language deficits. *American Journal of Human Genetics, 76*, 1074-1080.

MacNeilage, P. F. (1998). The frame/content theory of evolution of speech production. [PubMed]. *Behavioral and Brain Sciences, 21*, 499-546.

MacNeilage, P. F., & Davis, B. (1990). Acquisition of speech production: Frames, then content. In M. Jeannerod (Ed.), *Attention and Performance XIII: Motor Representation and Control* (pp. 453-176). London: Lawrence Erlbaum Associates.

MacNeilage, P. F., Davis, B., Kinney, A., & Matyear, C. L. (2000). The motor core of speech: a comparison of serial organization patterns in infants and languages. [Web of Science]. *Child Development, 71*(1), 153-163.

MacNeilage, P. F., & Davis, B. L. (2000). On the Origin of Internal Structure of Word Forms. [Science]. *Science, 288*(5465), 527-531.

MacNeilage, P. F., & Davis, B. L. (2001). Motor mechanisms in speech ontogeny: phylogenetic, neurobiological and linguistic implications. [Science Direct]. *Current Opinion in Neurobiology, 11*(6), 696-700.

Marion, M. J., Sussman, H. M., & Marquardt, T. P. (1993). The perception and production of rhyme in normal and developmentally apraxic children. [Science Direct]. *Journal of Communication Disorders, 26*(3), 129-160.

Marquardt, T. P., Jacks, A., & Davis, B. (2004). Token-to-token variability in developmental apraxia of speech: three longitudinal case studies. *Clinical Linguistics & Phonetics, 18*(2), 127-144.

Marquardt, T. P., Sussman, H. M., Snow, T., & Jacks, A. (2002). The integrity of the syllable in developmental apraxia of speech. [Science Direct]. *Journal of Communication Disorders, 35*(1), 31-49.

Matson, J. L., Wilkins, J., & Gonzalez, M. (2007). Early identification and diagnosis in autism spectrum disorders in young children and infants: How early is too early? [Science Direct]. *Research in Autism Spectrum Disorders, In Press, Corrected Proof*.

McCabe, P., Rosenthal, J. B., & McLeod, S. (1998). Features of developmental dyspraxia in the general speech-impaired population? *Clinical Linguistics & Phonetics, 12*(2), 105-126.

McCauley, R. J., & Strand, E. A. (2008). A review of standardized tests of nonverbal oral and speech motor performance in children. *American Journal of Speech Language Patholology, 17*(1), 81-91.

McCune, L. (1992). First words: a dynamic systems view. In C. A. Ferguson, L. Menn & C. Stoel-Gammon (Eds.), *Phonological Development: Models, Research, Implications* (pp. 313-336). Timonium: York Press.

McCune, L., & Vihman, M. M. (2001). Early phonetic and lexical development: A productivity approach. [ProQuest 5000]. *Journal of Speech, Language, and Hearing Research, 44*(3), 670-684.

Meng, X.-L., Rosenthal, R., & Rubin, D. B. (1992). Comparing correlated correlation coefficients. *Psychological Bulletin, 111*, 172-175.

Millar, C., Kail, R., & Leonard, L. (2001). Speed of processing in children with specific language impairment. *Journal of Speech, Language and Hearing Research, 44*, 416-433.

Mitchell, J. V. (1985). *The ninth mental measurements yearbook*. Lincoln: The Buros Institute of Mental Measurements.

Mitchell, P. R. (1995). A dynamic interactive developmental view of early speech and language production: application to clinical practice in motor speech disorders. *Seminars in Speech and Language, 16*(2), 100-109.

Mitchell, P. R., & Kent, R. D. (1990). Phonetic variation in multisyllable babbling. [PubMed]. *Journal of Child Language, 17*(2), 247-265.

Moore, C. A., & Ruark, J. L. (1996). Does speech emerge from earlier appearing oral motor behaviors? *Journal of Speech & Hearing Research, 39*, 1034-1047.

Moriarty, B. C., & Gillon, G. T. (2006). Phonological awareness intervention for children with childhood apraxia of speech. [Taylor & Francis]. *International Journal of Language & Communication Disorders, 41*(6), 713 - 734.

Morley, M. (1965). *The development and disorders of speech in childhood*. London: Churchill Livingstone.

Moyle, M. J., Ellis Weismer, S., Evans, J. L., & Lindstrom, M. J. (2007). Longitudinal relationships between lexical and grammatical development in typical and late-talking children. [ProQuest 5000]. *Journal of Speech, Language and Hearing Research 50*(2), 508-528.

Munson, B., Bjorum, E. M., & Windsor, J. (2003). Acoustic and perceptual correlates of stress in nonwords produced by children with suspected developmental apraxia of speech and children with phonological disorder. [CINAHL]. *Journal of Speech Language & Hearing Research, 46*(1), 189-202.

Nathani, S., Ertmer, D. J., & Stark, R. E. (2006). Assessing vocal development in infants and toddlers. *Clinical Linguistics and Phonetics, 20*(5), 351-369.

Nijland, L., Maassen, B., & van der Meulen, S. (2002). Speech motor subprocesses in DAS studied with a bite-block. In F. Windsor, M. L. Kelly & N. Hewlett (Eds.), *Invesitgations in Clinical Phonetics and Linguistics* (pp. 257-266). New Jersey: Lawrence Erlbaum Associates.

Nijland, L., Maassen, B., & van der Meulen, S. (2003). Evidence of motor programming deficits in children diagnosed with DAS. [ProQuest 5000]. *Journal of Speech, Language, and Hearing Research, 46*(2), 437-450.

Nijland, L., Maassen, B., van der Meulen, S., Gabreels, F., Kraaimaat, F. W., & Schreuder, R. (2002). Coarticulation patterns in children with developmental apraxia of speech. *Clinical Linguistics & Phonetics, 16*(6), 461-483.

Nijland, L., Maassen, B., van der Meulen, S., Gabreels, F., Kraaimaat, F. W., & Schreuder, R. (2003). Planning of syllables in children with developmental apraxia of speech. *Clinical Linguistics & Phonetics, 17*(1), 1-24.

Nittrouer, S. (1993). The emergence of mature gestural patterns is not uniform: Evidence from an acoustic study. *Journal of Speech and Hearing Research, 36*(5), 959-972.

Odell, K. M., & Shriberg, L. D. (2001). Prosody-voice characteristics of children and adults with apraxia of speech. *Clinical Linguistics & Phonetics, 15*(4), 275-307.

Oller, D. K. (2000). *The emergence of the speech capacity*. Mahwah: Lawrence Erlbaum Associates.

Oller, D. K., Eilers, R. E., & Basinger, D. (2001). Intuitive identification of infant vocal sounds by parents. *Developmental Science, 4*(1), 49-60.

Oller, D. K., Eilers, R. E., Neal, A. R., & Schwartz, H. K. (1999). Precursors to speech in infancy: The prediction of speech and language disorders. [Science Direct]. *Journal of Communication Disorders, 32*(4), 223-245.

Paterson, S. J., Brown, J. H., Gsodl, M. K., Johnson, M. H., & Karmiloff-Smith, A. (1999). Cognitive Modularity and Genetic Disorders. [Science Online]. *Science, 286*(5448), 2355-2358.

Peter, B., & Stoel-Gammon, C. (2008). Central timing deficits in subtypes of primary speech disorders. [Informa Healthcare]. *Clinical Linguistics & Phonetics, 22*(3), 171 - 198.

Peters, H. F. M., Hulstijn, W., & Lieshout, P. H. H. M. V. (2000). Recent developments in speech motor research into stuttering. [ProQuest 5000]. *Folia Phoniatrica et Logopaedica, 52*(1-3), 103.

Piek, J. P. (2002). The role of variability in early motor development. [Science Direct]. *Infant Behavior and Development, 25*(4), 452-465.

Piek, J. P. (2006). Theoretical approaches to motor development. In *Infant motor development* (pp. 31-63). Champaigne: Human Kinetics.

Piek, J. P., & Gasson, N. (1999). Spontaneous kicking in fullterm and preterm infants: Are there leg asymmetries? *Human Movement Science, 18*(2-3), 377-395.

Pytte, C. L., & Suthers, R. A. (2000). Sensitive period for sensorimotor integration during vocal motor learning. *Journal of Neurobiology, 42*(2), 172-189.

Reilly, S., Wake, M., Bavin, E., L., Prior, M., Williams, J., Bretherton, L., et al. (2007). Predicting Language at 2 Years of Age: A Prospective Community Study. [Pediatrics]. *Pediatrics, 120*(6), 1441-1449.

Rescorla, L. (1989). The Language Development Survey: a screening tool for delayed language in toddlers. *Journal of Speech and Hearing Disorders, 54*, 587-599.

Rescorla, L. (2002). Language and reading outcomes to age 9 in late-talking toddlers. *Journal of Speech, Language and Hearing Research, 45*(2), 360-371.

Rescorla, L., & Alley, A. (2001). Validation of the language development survey (LDS): a parent report tool for identifying language delay in toddlers. *Journal of Speech, Language and Hearing Research, 44*(2), 434-445.

Rescorla, L., Ratner, N., Jusczyk, P., & Jusczyk, A. (2005). Concurrent Validity of the Language Development Survey: Associations With the MacArthur-Bates Communicative Development Inventories: Words and Sentences. [Proquest 5000]. *American Journal of Speech - Language Pathology, 14*(2), 156-163.

Rescorla, L., Ross, G. S., & McClure, S. (2007). Language delay and behavioural/emotional problems in toddlers: Findings from two developmental clinics. *Journal of Speech, Language and Hearing Research, 50*(4), 1063-1078.

Rice, M. L., & Wexler, K. (1996). Toward tense as a clinical marker of specific language impairment in English-speaking children. *Journal of Speech, Language and Hearing Research, 39*, 1239-1257.

Roelofs, A. (1997). The WEAVER model of word-form encoding in speech production. [Science Direct]. *Cognition, 64*(3), 249-284.

Rothganger, H. (2003) Analysis of the sounds of the child in the first year of age and a comparison to the language. *Early Human Development, 75,* 55-69.

Roy, P., & Chiat, S. (2004). A Prosodically Controlled Word and Nonword Repetition Task for 2- to 4-Year-Olds: Evidence From Typically Developing Children. [ProQuest 5000]. *Journal of Speech, Language, and Hearing Research, 47*(1), 223-234.

Rvachew, S., Creighton, D., Feldman, N., & Sauve, R. (2002). Acoustic-phonetic description of infant speech samples: coding reliability and related methodological issues. *Acoustic Research Letters Online, 3*(1), 24-28.

Sabbagh, M. A. (1999). Communicative intentions and language: evidence from right-hemisphere damage and autism. *Brain and Language, 70*, 29-69.

Selby, J., Robb, M., & Gilbert, H. (2000). Normal vowel articulations between 15 and 36 months of age *Clinical Linguistics and Phonetics, 14*(4), 255-265.

Shriberg, L. D. (2003). Diagnostic markers for child speech-sound disorders: introductory comments. *Clinical Linguistics & Phonetics, 17*(7), 501-505.

Shriberg, L. D., Aram, D., & Kwiatkowski, J. (1997a). Developmental apraxia of speech: I. Descriptive and theoretical perspectives. [ProQuest 5000]. *Journal of Speech, Language, and Hearing Research, 40*(2), 273.

Shriberg, L. D., Aram, D., & Kwiatkowski, J. (1997b). Developmental apraxia of speech: II. Toward a diagnostic marker. [ProQuest 5000]. *Journal of Speech, Language, and Hearing Research, 40*(2), 286-312.

Shriberg, L. D., Aram, D., & Kwiatkowski, J. (1997c). Developmental apraxia of speech: III. A subtype marked by inappropriate stress. [ProQuest 5000]. *Journal of Speech, Language, and Hearing Research, 40*(2), 313.

Shriberg, L. D., Campbell, T. F., Karlsson, H. B., Brown, R. L., McSweeny, J. L., & Nadler, C. J. (2003). A diagnostic marker for childhood apraxia of speech: the lexical stress ratio. [CINAHL]. *Clinical Linguistics & Phonetics., 17*(7), 549-574.

Shriberg, L. D., Green, C. L., Campbell, T. F., McSweeny, J. L., & Scheer, A. R. (2003). A diagnostic marker for childhood apraxia of speech: the coefficient of variation ratio. [CINAHL]. *Clinical Linguistics & Phonetics, 17*(7), 575-595.

Shriberg, L. D., & Tomblin, J. B. (1999). Prevalence of speech delay in 6-year-old children and comorbidity with langauge impairment. *Journal of Speech, Language and Hearing Research, 42*(1461-1481).

Sivberg, B. (2003). Parents' detection of early signs in their children having an autistic spectrum disorder. *Journal of Pediatric Nursing, 18*(6), 433-439.

Skinder, A., Connaghan, K. P., Strand, E. A., & Betz, S. (2000). Acoustic correlates of perceived lexical stress errors in children with developmental apraxia of speech. *Journal of Medical Speech-Language Pathology., 8*(4), 279-284.

Skinder, A., Strand, E. A., & Mignerey, M. (1999). Perceptual and acoustic analysis of lexical and sentential stress in children with developmental apraxia of speech. *Journal of Medical Speech-Language Pathology., 7*(2), 133-144.

St James-Roberts, I. (2004). Evaluation of the WILSTAAR scheme. *Literacy Today, 40*.

St James-Roberts, I., & Alston, E. (2006). Attention development in 10-month-old infants selected by the WILSTAAR screen for pre-language difficulties. *Journal of Child Psychology and Psychiatry, 47*(1), 63-68.

Stackhouse, J. (1992). Developmental verbal dyspraxia I: A review and critique. *European Journal of Disorders of Communication, 27*, 19-34.

Stackhouse, J., & Snowling, M. (1992). Developmental verbal dyspraxia II: A developmental perspective on 2 case studies. *European Journal of Disorders of Communication, 27*(1), 35-51.

Stackhouse, J., & Wells, B. (1997). *Children's speech and literacy difficulties I: A psycholinguistic framework*. London: Whurr.

Stark, R. E., Bernstein, L. E., & Demorest, M. E. (1993). Vocal communication in the first 18 months of life. *Journal of Speech and Hearing Research, 36*, 548-558.

Stoel-Gammon, C. (1989). Prespeech and early speech development of two late talkers. *First Language, 9*, 207-224.

Stoel-Gammon, C. (1992). Prelinguistic vocal development: measurements and predictions. In C. A. Ferguson, L. Menn & C. Stoel-Gammon (Eds.), *Phonological Development: Models, Research, Implications* (pp. 439-456). Maryland: York Press.

Strand, E. A. (1992). The integration of speech motor control and language formulation in process models of acquisition. In R. S. Chapman (Ed.), *Processes in Language Acquisition and Disorders* (pp. 86-107). Saint Louis: Mosby Yearbook.

Strand, E. (2002). An examination of childhood apraxia of speech, *Basic Science in Speech and Language*. Atlanta, US.

Strand, E. (2003). Childhood apraxia of speech: Suggested diagnostic markers for the younger child. In L. Shriberg & T. Campbell (Eds.), *Proceedings of the 2002 Childhood Apraxia of Speech Research Symposium* (pp. 75-80). Arizona: The Hendrix Foundation.

Strand, E. A., & Debertine, P. (2000). The efficacy of integral stimulation with developmental apraxia of speech. *Journal of Medical Speech-Language Pathology., 8*(4), 295-300.

Sutherland, D., & Gillon, G. T. (2007). Development of phonological representations and phonological awareness in children with speech impairment. *International Journal of Language & Communication Disorders, 42*(2), 229-250.

Tabachnick, B., & Fidell, L. (2001). *Using multivariate statistics*. Boston: Allyn and Bacon.

Tallal, P., & Stark, R. E. (1981). Speech acoustic-cue discrimination abilities of normally developing and language-impaired children. *The Journal of the Acoustical Society of America, 69*(2), 568-574.

Thelen, E. (1981). Rhythmical behavior in infancy: an ethological perspective. *Developmental Psychology, 17*(3), 237-257.

Thoonen, G., Maassen, B., Gabreels, F., & Schreuder, R. (1999). Validity of maximum performance tasks to diagnose motor speech disorders in children. [ProQuest 5000]. *Clinical Linguistics & Phonetics, 13*(1), 1-13.

Thoonen, G., Maassen, B., Gabreels, F., Schreuder, R., & de Swart, B. (1997). Towards a standardised assessment procedure for developmental apraxia of speech. *International Journal of Language & Communication Disorders, 32*(1), 37-60.

Thoonen, G., Maassen, B., Wit, J., Gabreels, F., & Schreuder, R. (1996). The integrated use of maximum performance tasks in differential diagnostic evaluations among children with motor speech disorders. *Clinical Linguistics and Phonetics, 10*(4), 311-336.

Tomblin, J. B., Records, N. L., Buckwalter, P., Zhang, X., & Smith, E. (1997). Prevalence of specific langauge impairment in kindergarten children. *Journal of Speech, Language and Hearing Research, 40*, 1245-1260.

Vaal, J., van Soest, A. J., Hopkins, B., Sie, L. T. L., & van der Knaap, M. S. (2000). Development of spontaneous leg movements in infants with and without periventricular leukomalacia. *Experimental Brain Research, 135*(1), 94-105.

Vargha-Khadem, F., Gadian, D. G., Copp, A., & Mishkin, M. (2005). FOXP2 and the neuroanatomy of speech and language. *Neuroscience, 6*, 131-138.

Varley, R., & Whiteside, S. P. (2001). What is the underlying impairment in acquired apraxia of speech? *Aphasiology, 15*(1), 39-84.

Velleman, S., & Strand, K. (1994). Developmental verbal dyspraxia. In J. E. Bernthall & N. W. Bankson (Eds.), *Child Phonology: Characteristics, assessment and intervention with special populations* (pp. 110-139). New York: Thieme Medical.

Velleman, S. L. (1994). The interaction of phonetics and phonology in developmental verbal dyspraxia: two case studies. *Clinics in Communication Disorders, 4*(1), 66-77.

Velleman, S. L. (2002). Phonotactic therapy. *Seminars in Speech and Language, 23*(1), 43-55.

Velleman, S. L., & Shriberg, L. D. (1999). Metrical analysis of the speech of children with suspected developmental apraxia of speech. [CINAHL]. *Journal of Speech Language & Hearing Research., 42*(6), 1444-1460.

Vernes, S. C., Nicod, J., Elahi, F. M., Coventry, J. A., Kenny, N., Coupe, A. M., et al. (2006). Functional genetic analysis of mutations implicated in a human speech and language disorder. [ProQuest]. *Human Molecular Genetics [NLM - MEDLINE], 15*(21), 3154-3167.

Viholainen, H., Ahonen, T., Lyytinen, P., Cantell, M., Tolvanen, A., & Lyytinen, H. (2006). Early motor development and later language and reading skills in children at risk of familial dyslexia. [Cambridge Press]. *Developmental Medicine and Child Neurology, 48*(5), 367-373.

Voperian, H., & Kent, R. (2007). Vowel acoustic space development in children: A synthesis of acoustic and anatomic data. *Journal of Speech, Language and Hearing Research, 50*, 1510-1545.

Ward, S. (1992). The predictive validity and accuracy of a screening test for language delay and auditory perceptual disorder. *European Journal of Disorders of Communication, 27*, 55-72.

Watkins, K. E., Vargha-Khadem, F., Ashburner, J., Passingham, R. E., Connelly, A., Friston, K. J., et al. (2002). MRI analysis of an inherited speech and language disorder: structural brain abnormalities. [PubMed]. *Brain, 125*(3), 465-478.

Watson, L. R., Baranek, G. T., & DiLavore, P. C. (2003). Toddlers with Autism: Developmental perspectives. *Infants and Young Children, 16*(3), 201-214.

Watt, N., Wetherby, A., & Shumway, S. (2006). Prelinguistic predictors of language outcome at 3 years of age. *Journal of Speech, Language and Hearing Research, 49*(6), 1224-1237.

Wechsler, D. (2002). *Wechsler Preschool and Primary Scale of Intelligence - 3rd Edition*. San Antonio: PsychCorp.

Weismer, S. E., Evans, J., & Hesketh, L. J. (1999). An examination of verbal working memory capacity in children with specific language impairment. *Journal of Speech, Language and Hearing Research, 42*(5), 1249-1260.

Westermann, G., & Miranda, E. R. (2004). A new model of sensorimotor coupling in the development of speech. [Science Direct]. *Brain and Language, 89*(2), 393-400.

Wetherby, A. M., Allen, L., Cleary, J., Kublin, K., & Golstein, H. (2002). Validity and reliability of the communication and symbolic behavior scales developmental profile with very young children. *Journal of Speech, Language and Hearing Research, 45*(6), 1202-1218.

Wetherby, A. M., & Prizant, B. M. (2002). *Communication and Symbolic Behavior Scales - Developmental Profile*. Baltimore: Paul H Brookes Publishing Company.

Wetherby, A. M., Woods, J., Allen, L., Cleary, J., Dickinson, H., & Lord, C. (2004). Early indicators of autism spectrum disorders in the second year of life. *Journal of Autism and Developmental Disorders, 34*(5), 473-493.

Whitehurst, G. J., Smith, M., Fischel, J. E., Arnold, D. S., & Lonigan, C. J. (1991). The continuity of babble and speech in children with specific expressive language delay. *Journal of Speech & Hearing Research, 34*, 1121-1129.

Wiig, E. H., Secord, W., & Semel, E. (1992). *Clinical Evaluation of Language Fundamentals - Preschool*. Texas: The Psychological Corporation.

Williams, P., & Stackhouse, J. (2000). Rate, accuracy and consistency: diadochokinetic performance of young, normally developing children. *Clinical Linguistics & Phonetics, 14*(4), 267-293.

Williams, R., & Inghman, R. J. (1981). A further analysis for developmental apraxia of speech in children with defective articulation. *Journal of Speech and Hearing Research, 24*, 496 - 505.

Wong, V., Huia, L. S., Lee, W., & Leung, L. (2004). A modified screening tool for autism. *Pediatrics, 114*(2), 479 - 480.

Zeigler, W., & Maassen, B. (2004). The role of the syllable in disorders of spoken language production. In B. Maassen, R. D. Kent, H. Peters, P. van Lieshout & W. Hulstijn (Eds.), *Speech Motor Control in Normal and Disordered Speech* (pp. 415-447). Oxford: Oxford University Press.

Zubrick, S. R., Taylor, C. L., Rice, M. L., & Slegers, D. W. (2007). Late language emergence at 24 months: An epidemiological study of prevalance, predictors, and covariates. *Journal of Speech, Language and Hearing Research, 50*, 1562-1592.

APPENDICES

APPENDIX A

Before First Words Questionnaire

Pre-speech questionnaire

The questions that follow will require you to think back to when your child was a baby. It can be very difficult to remember what children did back then, so you might want to ask other relatives (e.g. Grandparents, Aunts and Uncles), or look at your child's yellow personal health record book, or even look at early videos if you have them.

EARLY SOUNDS

1. Did your child make many sounds as a baby (particularly between the ages of 6 and 12 months)?

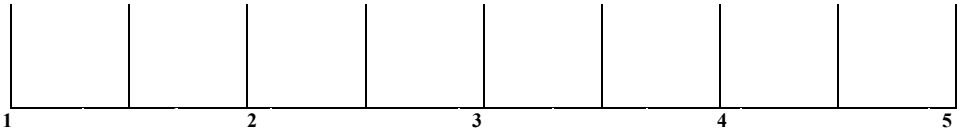
☐ yes ☐ no

2. Please describe the kinds of sounds your child made as a baby. *(You can answer generally, or put down different ages if you wish)*

_____
_____
_____
_____
_____
_____

The next question is asking you to think about the times when your child made sounds as a baby. We are talking about sounds that the baby made with his/her mouth that <u>WEREN'T</u> crying or burping noises.

3. How much did your child vocalise as a baby (particularly between the ages of 6 and 12 months)? Please rate on a scale of 1 to 5, with 1 indicating that your child seemed to vocalise rarely (or was a 'quiet' baby), and 5 indicating that your baby seemed to vocalise often (or was a 'vocal' baby).



Rarely vocalised except for crying & burps etc

Frequently vocalised

4. Did your child make 'cooing' and 'gooing' noises, for example making vowel type noises like 'ah', 'ee'?

☐ **yes** ☐ **no** ☐ **unsure**

If yes, at what age? (you can circle one particular month, or a range of months)

3 mnths	4 mnths	5 mnths	6 mnths	7 mnths	8 mnths	9 mnths	10 mnths	11 mnths	12 mnths	13 mnths	14 mnths	15 mnths

Other:_____ ☐ can't remember

5. **Did your child 'babble' as a baby? When I say 'babble', I mean did he/she say sounds like "ba-ba", "ma-ma", "da-da-da" where the sound is repeated a few times?**

☐ **yes** ☐ **no** ☐ **unsure**

If yes, at what age? (you can circle one particular month, or a range of months)

6 mnths	7 mnths	8 mnths	9 mnths	10 mnths	11 mnths	12 mnths	13 mnths	14 mnths	15 mnths	16 mnths	17 mnths	18 mnths

Other: _____ ☐ can't remember

6. Did your child ever produce babble where the consonant sound changed, for example, "ba-da", "gollygolly", "teda", "be-de-ga"?

☐ **yes** ☐ **no** ☐ **unsure**

If yes, at what age? (you can circle one particular month, or a range of months)

6 mnths	7 mnths	8 mnths	9 mnths	10 mnths	11 mnths	12 mnths	13 mnths	14 mnths	15 mnths	16 mnths	17 mnths	18 mnths

Other:_____ ☐ can't remember

7. Did he/she babble as much as other children? (please state who your are comparing to…. E.g. 'not as much as friend's children', 'more than his/her brothers/sisters')

☐ **babbled less** ☐ **babbled more** ☐ **babbled about the same**
compared to _____

☐ **unsure**

8. Did your child sound different to other children?

- ❑ Yes (please describe):
  _____
  _____
- ❑ no

9. Did your child make other noises as a baby (eg. Raspberries, squeals)? Please describe

☐ **yes** ☐ **no** ☐ **unsure**
_____
_____
_____

FEEDING

1. Did your child have any feeding difficulties as a baby? (eg. Poor suck, difficulty moving to lumpy foods, avoiding certain food textures)

- ❑ Yes (please describe):

  _____
  _____
  _____

- ❑ No

2. Did he/she have any issues with dribbling? If yes, please describe

- ❑ Yes (please describe):

  _____
  _____
  _____

- ❑ No

OTHER

1. Did anything about your child ever concern you as a baby?

- ❑ Yes (please describe):

  _____
  _____

- ❑ no

2. Please indicate the approximate ages that your baby:

Sat upright _____ Smiled _____
Began crawling _____ Took first steps _____
Said first word _____
Joined two words together (eg. 'dad gone', 'more juice')
_____

3. Is there anything else you would like to tell us about your child?

_____
_____
_____

Thankyou for taking the time to complete this questionnaire. I realise it is not an easy task to remember back to when your child was a baby, so I appreciate the effort involved.

APPENDIX B

Statement of Copyright and Authorship

Copyright

Chapter 2 of the present thesis appears as a peer-reviewed article in Child Language Teaching and Therapy, a journal published by SAGE (USA, UK, Singapore, India). Copyright permission information from the publisher states that authors may re-use their content in any print-form work written or edited by the Author, without seeking permission from SAGE. The final, definitive version of this paper has been published in Child Language Teaching and Therapy, 2008, Vol 24, by SAGE Publications Ltd, All rights reserved and © and appears at http://sagepub.com/cgi/content/abstract/24/3/285

Authorship

I, Chantelle Denise Highman, contributed the major conception, design, data collection, analysis, and interpretation aspects to the contents of the paper entitled:

> Highman, C., Hennessey, N., Sherwood, M., & Leitao, S. (2008). Retrospective parent report of early vocal behaviours in children with suspected Childhood Apraxia of Speech (sCAS). *Child Language Teaching and Therapy, 24*(3), 285-306.

The co-authors Hennessy and Leitão, as my PhD supervisors, contributed input appropriate to their supervisory role (being supervision of the project, design and analysis guidance, and assistance with editing of the paper where required). The co-author Sherwood, as a clinical specialist speech pathologist, contributed input regarding the clinical cases and practical support with recruitment.

January 2010

APPENDIX C

Description of the WILSTAAR program

In 2001, the Community Health Branch of the Health Department of Western Australia commenced the implementation of the WILSTAAR/Baby Talk program in selected areas. The program was based on the WILSTAAR program described by Ward and colleagues (Ward, 1992, 1999), and was aimed at providing an early identification and health promotion program to appropriate families. A summary of the program, which was based heavily on assumptions of environmental factors contributing to language delay and/or impairment, is provided below.

1. WILSTAAR screen (Ward, 1992) implemented to all infants attending their routine 8-9 month child health check (see Appendix D)
2. Child health nurse then sent completed forms to local speech pathologists
3. Speech pathologists scored WILSTAAR screen:
    a. If the infant passed, no further action was provided
    b. If the infant failed the screen, parents were sent a letter with an appointment for a home visit session to take part in a speech and language promotion program
4. Speech pathologists completed home visit assessment, consisting of the REEL-2 and WILSTAAR record forms:
    a. If the infant passed the REEL-2, they were considered a false positive and no further action was required
    b. If the infant failed the REEL-2, they were considered a true positive and offered the intervention program
5. The intervention program consisted of monthly home visits, whereby speech pathologists would provide standard information to the parent/s regarding activities to stimulate listening and language skills in their infant (e.g., encouraging the parents to make symbolic noise, talk about what they are doing with their infant, and use simple language). Infants were categorised

into one of 3 groups, depending on their profile, and were given programs according to this:

a. Group 1. Infants in this group failed the receptive component of the screen, and then failed either or both components of the REEL-2. The program focussed on strategies to develop the infant's selective attention skills (auditory perceptual), for example – showing the infant the source of environmental noises, and encouraging the parent to notice what the infant is looking at and using simple language
b. Group 2. Infants in this group failed the expressive component of the screen, and then failed the receptive (and expressive component) of the REEL-2. The program focussed on strategies to increase the quantity and quality of the parents' input, for example – saying rhymes and playsounds with the infant.
c. Group 3. Infants in this group failed the expressive component of the screen, and then (only) failed the expressive component of the REEL-2. The program focussed on strategies to encourage enjoyment in sound making and talking, for example – encouraging the parent to repeat words often and copy back the sounds the infant makes.

6. The REEL-2 was readministered at the completion of the program (length depended on how quickly parents/infants had moved through the techniques)

Despite the original positive reports of WILSTAAR's effectiveness (Ward, 1999), concerns regarding the validity of the screen (St James-Roberts & Alston, 2006) and whether the program (as opposed to factors within the infants or simply spontaneous improvement) was responsible for the positive results, led to much debate about the claims of the program (St James-Roberts, 2004). In Western Australia, the program was never fully 'rolled out' to every health service area, and eventually was scaled down and replaced by less targeted programs.

APPENDIX D

WILSTAAR screen questions

Receptive Section

Q1. Does s/he always notice sounds like people coming into the room or food preparation sounds?

Q2. Does s/he always notice when you call his/hername when s/he's not really concentrating on play?

Q3. Does s/he notice cars passing, dogs barking, the hoover, as much as ever?

Q4. a) Does s/he ever ignore interesting or unusual sounds?

If yes, b) Would s/he if not concentrating on something else?

Q5. Would s/he always turn a second time to an interesting sound like the rattle of a biscuit tin, if it came again soon after the first time?

Q6. Have you ever, at any time, thought s/he might have a hearing loss?

Expressive Section

Q7. Does s/he string different sounds together now e.g., ba dee goo dee bow?

APPENDIX E

Single case comparison results – Study 2B

Table E1.
T values (with p values in parentheses) for the case comparisons (Crawford & Garthwaite, 2002) of the clinical sample participants compared to the false positive group on the Ages and Stages Questionnaire (ASQ, Bricker et al., 1999)

	Subtest *t* values				
Participant	Comm	GM	FM	Prob	Pers-Soc
1	-	-	-	-	-
2	1.08 (.14)	0.88 (.19)	0.56 (.29)	0.80 (.22)	0.59 (.28)
3	0.50 (.33)	0.88 (.19)	1.31 (.10)	0.58 (.28)	0.66 (.26)
4	-	-	-	-	-
5	-	-	-	-	-
6	2.10 (.02)*	0.08 (.47)	1.31 (.10)	0.80 (.22)	0.04 (.49)
7	1.08 (.14)	1.52 (.07)	0.37 (.36)	0.11 (.46)	0.04 (.49)
8	0.96 (.17)	0.88 (.19)	0.56 (.29)	0.58 (.28)	0.66 (.26)
9	0.96 (.17)	0.88 (.19)	0.56 (.29)	0.58 (.28)	0.66 (.26)

Note. - = missing data
* statistically significant at $p < .05$

Table E2.
T values (with p values in parentheses) for the case comparisons (Crawford & Garthwaite, 2002) of the clinical sample participants compared to the true positive group on the Ages and Stages Questionnaire (ASQ, Bricker et al., 1999)

	Subtest *t* values				
Participant	Comm	GM	FM	Prob	Pers-Soc
1	-	-	-	-	-
2	0.96 (.17)	0.91 (.18)	0.54 (.30)	0.40 (.35)	0.47 (.32)
3	0.60 (.27)	0.91 (.18)	1.62 (.053)	0.89 (.19)	0.86 (.20)
4	-	-	-	-	-
5	-	-	-	-	-
6	1.99 (.02)*	0.11 (.46)	1.62 (.053)	0.40 (.35)	0.20 (.42)
7	0.96 (.17)	1.64 (.052)	0.54 (.30)	0.25 (.40)	0.20 (.42)
8	1.12 (.13)	0.91 (.18)	0.54 (.30)	0.89 (.19)	0.86 (.20)
9	1.12 (.13)	0.91 (.18)	0.54 (.30)	0.89 (.19)	0.86 (.20)

Note. - = missing data
* statistically significant at $p < .05$

Table E3.
T values (with p values in parentheses) for the case comparisons (Crawford & Garthwaite, 2002) of the clinical sample participants compared to the false positive group on the REEL-2 (Bzoch & League, 1991)

	Subtest			
Participant	Receptive		Expressive	
1	1.48	(.07)	1.32	(.10)
2	0.96	(.17)	2.60	(.01)*
3	1.99	(.03)*	0.68	(.25)
4	2.32	(.01)*	4.68	(<.001)**
5	2.32	(.01)*	2.95	(.002)**
6	0.96	(.17)	5.21	(<.001)**
7	1.99	(.03)*	1.96	(.03)*
8	0.96	(.17)	1.96	(.03)*
9	1.29	(.10)	1.09	(.14)

* statistically significant at $p < .05$ ** statistically significant at $p < .01$

Table E4.
T values (with p values in parentheses) for the case comparisons (Crawford & Garthwaite, 2002) of the clinical sample participants compared to the true positive group on the REEL-2 (Bzoch & League, 1991)

	Subtest			
Participant	Receptive		Expressive	
1	0.16	(.44)	0.82	(.21)
2	0.48	(.32)	1.19	(.12)
3	0.80	(.21)	1.83	(.04)*
4	1.21	(.11)	4.48	(<.001)**
5	1.21	(.11)	1.74	(.04)*
6	0.48	(.32)	5.30	(<.001)**
7	0.80	(.21)	0.18	(.43)
8	0.48	(.32)	0.18	(.43)
9	0.07	(.47)	1.19	(.12)

* statistically significant at $p < .05$ ** statistically significant at $p < .01$

APPENDIX F

Comparisons of number of sounds – Study 2B

Table F1.
T and p values for the case comparisons (Crawford & Garthwaite, 2002) of the clinical sample participants compared to both the false positive and true positive groups on the number of reported consonant sounds

		Comparison Group			
		False positives		True Positives	
Participant	# consonants	T value	*p* value	T value	*p* value
1	2	0.73	.23	0.34	.38
2	2	0.73	.23	0.34	.38
3	3	0.13	.50	0.50	.31
4	2	0.73	.23	0.34	.38
5	4	0.99	.16	1.34	.09
6	0	2.46	.01*	2.02	.02*
7	4	0.99	.16	1.34	.09
8	1	1.60	.06	1.18	.12
9	3	0.13	.50	0.50	.31

* statistically significant at $p < .05$

APPENDIX G

Results of two way mixed ANOVAs – Study 3

Table G1.
Analysis of variance results for the group by timepoint interaction effects on the Ages and Stages Questionnaire (ASQ, Bricker et al., 1999)

Subtest	*df*	*F*	*p*	$\eta^2_{partial}$
Communication	2, 14	0.85	.44	.06
Gross Motor	2, 14	2.86	.07	.17
Fine Motor	2, 14	1.37	.27	.09
Problem Solving	2, 14	0.06	.94	.01
Personal-Social	2, 14	0.09	.92	.01

Table G2.
Analysis of variance results for the group by timepoint interaction effects on Communication and Symbolic Behaviour Scales (CSBS) Caregiver Questionnaire (Wetherby & Prizant, 2002)

Subtest	*df*	*F*	*p*	$\eta^2_{partial}$
Expression and eye gaze	4, 14	0.53	.72	.04
Communication	4, 14	1.02	.40	.07
Gesture	4, 14	1.14	.35	.08
Sounds	4, 14	0.82	.52	.06
Words	4, 14	1.02	.41	.07
Understanding	4, 14	0.98	.43	.07
Object use	4, 14	0.66	.62	.05

APPENDIX H

Post hoc results for CSBS Caregiver Questionnaire main effect of timepoint – Study 3

Table H1.
Analysis of variance post hoc comparisons for the main effect of timepoint on the Communication and Symbolic Behaviour Scales (CSBS) Caregiver Questionnaire (Wetherby & Prizant, 2002)

Subtest	Timepoint Comparisons (months)		Mean Difference	*p*
Expression and eye gaze	9	12	-2.92	.07
		15	-2.95	.08
		18	-2.54	.29
		24	0.95	.03*
	12	15	-0.03	1.0
		18	0.38	1.0
		24	-0.47	1.0
	15	18	0.41	1.0
		24	0.44	1.0
	18	24	-0.85	1.0
Gesture	9	12	0.56	1.0
		15	-2.31	.59
		18	-4.19	.01*
		24	-5.17	.001**

	12	15	-2.88	.12
		18	-4.75	.001**
		24	-5.73	<.001**
	15	18	-1.88	.84
		24	-2.86	.16
	18	24	-0.98	1.0
Sounds	9	12	-0.31	1.0
		15	-0.47	1.0
		18	-1.38	.81
		24	-4.23	.01*
	12	15	-0.16	1.0
		18	-1.06	1.0
		24	-3.92	.001**
	15	18	-0.91	1.0
		24	-3.77	.048*
	18	24	-2.86	.07
Words	9	12	0.04	1.0
		15	1.07	1.0
		18	-0.58	1.0
		24	-3.12	.08
	12	15	1.03	.88
		18	-0.63	1.0
		24	-3.15	.10
	15	18	-1.66	.28
		24	-4.18	.01*
	18	24	-2.53	.24

Understanding	9	12	-1.08	.44
		15	-0.05	1.0
		18	-1.77	.54
		24	-4.30	.01*
	12	15	1.03	.22
		18	-0.69	1.0
		24	-3.21	.03*
	15	18	-1.72	.09
		24	-4.25	.01*
	18	24	-2.53	.12

* significant at $p < .05$ ** significant at $p < .01$