

# Caring for a child with severe intellectual disability in China: The example of Rett syndrome

## Contributors

Faye Lim <sup>a</sup>

Jenny Downs <sup>ab</sup>

Jianghong Li <sup>ac</sup>

Xin-Hua Bao <sup>d</sup>

Helen Leonard <sup>a</sup>

## Affiliations

<sup>a</sup> Telethon Institute for Child Health Research, Center for Child Health Research, The University of Western Australia. PO Box 855 West Perth, Western Australia 6872, Australia

<sup>b</sup> School of Physiotherapy and Curtin Health Innovation Research Institute, Curtin University, GPO Box U1987, Perth, Western Australia 6845, Australia.

<sup>c</sup> Curtin Health Innovation Research Institute, Centre for Population Health Research, Curtin University. GPO Box U1987, Perth, Western Australia 6845, Australia.

<sup>d</sup> Department of Paediatrics, Peking University First Hospital, No. 8 Xishiku Street, Xicheng District, Beijing 100034, China

## Corresponding author

Helen Leonard

Telethon Institute for Child Health Research, Centre for Child Health Research,

The University of Western Australia,

PO Box 855 West Perth, Western Australia 6872, Australia.

Telephone: + 61 8 9489 7790

Fax: + 61 8 9489 7700

E-mail: [hleonard@ichr.uwa.edu.au](mailto:hleonard@ichr.uwa.edu.au)

### **Implications for rehabilitation**

- Little is known about the lives of girls with Rett syndrome in China, a genetic condition associated with severe disability.
- Most girls experienced home-based care with limited access to appropriate equipment, education and rehabilitation.
- Limited information and discrimination and stigma from some community members reduced families' abilities to meet their child's needs.
- Greater access to health, education and information services in China should be supported.

## **ABSTRACT**

*Purpose:* Rett syndrome is one of several genetic disorders known to cause severe intellectual and physical disability, mostly in girls. Girls affected by Rett syndrome appear to develop normally in the first six months of life, after which the usual clinical presentation comprises regression of communication and hand skills, the appearance of hand stereotypies and impaired gait. Intellectual disability affects more than 1.5% of the population of children in developing countries yet we know little about the daily lives and support services available for them and their caregivers. *Method:* This qualitative study explored the daily experiences of 14 mothers and one grandmother caring for a child with Rett syndrome in China via telephone interviews. *Results:* Participants reported a lack of education, rehabilitation and support services available to them. Limited access to information reduced families' capacity to adequately meet the needs of their child. These gaps were further exacerbated by discrimination and perceived stigma from some members of the community. *Conclusions:* Additional support services and educational programs at the governmental level can improve the quality of life of persons with an intellectual disability and their families and programs involving community participation in the care of people with disabilities may help to address discrimination.

## **Keywords**

Rett syndrome; severe disability, China, developing countries; support services

## INTRODUCTION

Approximately 1.5% of children in Western Australia have a severe intellectual disability [1] and this proportion may be higher in developing countries [2]. Differences in definitions, methodology and population surveyed make it difficult to provide estimates on the overall prevalence of intellectual disability in the developing world [3, 4]. Higher prevalence may be partly attributable to increased exposure to risk factors such as poverty, toxins and malnutrition [2]. Investigations into the experiences of families caring for a child with intellectual disability have been conducted mainly in developed countries [5, 6], with few studies in developing countries [7]. In each of these studies, support services were considered valuable and their availability was associated with advantages for the child with a disability and also for family functioning [8]. It is therefore important to understand how families in the developing world care for a child with intellectual disability and the support services available to them.

China has a population of approximately 1.3 billion people and is one of the fastest developing economies in the world [9]. Even so, nearly 39 million people there live below the poverty line [9] with an unequal distribution of income across provinces [10]. Traditional Chinese beliefs perceive disability as a punishment for wrongdoings from a previous life [11]. Therefore, persons with a disability and their families may be subject to stigma and discrimination [12]. It is believed that many children with an intellectual disability in China are not formally diagnosed, receive very few services and their families experience financial stress in caring for their child [13]. Information about the daily lives and support services available for those with an intellectual disability is limited.

Many of the services required by children with an intellectual disability are within healthcare and education systems. The current education system in China provides limited opportunities for children with severe intellectual disability [14], although these children often receive some healthcare services [15]. The Chinese healthcare system is unique in that it incorporates aspects of both Western medicine and traditional Chinese medicine (TCM) [16]. TCM is based on the concept of *qi* (energy flow) and commonly used treatments include herbal remedies, acupuncture, moxibustion and massage [16]. Clinicians in China often study both Western medicine and TCM, although the level of training in each area may vary depending on the clinician's specialization [17]. Rehabilitation services often incorporate TCM together with approaches commonly used in developed countries, such as physiotherapy [18]. TCM represents a unique facet to the experiences of families in caring for a child with severe intellectual disability in China.

Rett syndrome is one of several genetic disorders known to cause severe intellectual and physical disability. It is caused by mutations in the *MECP2* gene [19] and in a population-based study in Australia, an estimated 1 in 10,000 girls will be diagnosed by 12 years of age [20]. Girls affected by Rett syndrome appear to develop normally in the first six months of life, after which characteristic signs of the condition occur, including regression of communication and hand skills, the appearance of hand stereotypies and impaired gait. Eye gazing is used commonly as a means of communication [21] although some girls do retain the ability to use words following the regression period [22]. A video study using Australian population-based data (n=144) has found hand function skills ranging from no functional hand use in approximately one third to a similar proportion being able to pick up small objects [23]. Hand stereotypies such as wringing, mouthing and clapping develop in almost

all girls and women, with the number of different types of movements and their frequency decreasing as the girls grow older [24]. Population data from Australia has found that nearly half the girls and women had never learned to walk [25]. Rett syndrome is also associated with other co-morbidities, such as breathing disturbances, scoliosis, epilepsy and poor growth [26].

The International Rett Syndrome Phenotype Database (InterRett) was established in 2002 to collect information from families and clinicians to facilitate collaboration on genotype-phenotype studies in Rett syndrome [27]. To date, InterRett has information from over 2000 families in 53 countries and this is because of strong collaborative ties with clinicians and family associations around the world, including China. All families from China participating in InterRett were first ascertained by clinicians at Peking University First Hospital [27]. This qualitative study describes the daily experiences of a sample of families living in China with a daughter diagnosed with Rett syndrome and who were participating in InterRett.

## **METHODS**

This research was approved by the Human Research Ethics Committee, The University of Western Australia and by the Department of Pediatrics, Peking University First Hospital. Participants were recruited from the InterRett database, the inclusion criteria including (1) a confirmed clinical or genetic diagnosis of Rett syndrome for the child [26] and; (2) that the caregiver had a good command of written and spoken Mandarin (Putonghua). All study materials including interview questions and schedules and consent forms were translated into Mandarin.

Sampling was designed to capture variability in life experiences [28]. Potential families were divided into sampling groups based on the child's age at the time of recruitment ( $\leq 5.5$  years,  $5.5 < 10.5$  years and  $>10.5$  years) and distance from Beijing ( $\leq 500\text{km}$ ,  $500 < 1000\text{km}$  and  $>1000\text{km}$ ). Three families from each category were sampled at random using sampling commands in Stata9 [29]. A second sampling for families with a child older than 5.5 years and who lived  $>500\text{km}$  and  $>1000\text{km}$  from Beijing was required as nine families in the first sample either declined ( $n=1$ ), were not interested in participating ( $n=3$ ) or were not contactable ( $n=5$ ). The second sampling yielded one additional family, resulting in a total of 14 families.

The Australian Rett Syndrome Study aims to develop and maintain an ongoing population-based data collection for research on the genetic and clinical characteristics of Rett syndrome as well as its impact on daily living [30]. The study has a Consumer Reference Group - a group of parents with a daughter with Rett syndrome from around Australia who meet by teleconference to provide a consumer perspective on research directions and the methods being used to undertake this research [31]. For this study, an interview schedule was designed in consultation with the Australian Consumer Reference Group and subsequently piloted with three Chinese families who met the inclusion criteria for this study. Both groups provided input on the appropriateness and comprehensiveness of the interview questions from a family perspective, with the latter group providing additional feedback on cultural sensitivity. The interview schedule included questions on the functional skills of the child, educational and social experiences, and concerns for the child's future. All suggestions received during the piloting processes were incorporated into the final interview schedule.

Following recruitment, mothers of the affected child were interviewed in Mandarin via telephone. Tape-recorded interviews were conducted by FJL, a Malaysian-born ethnic Chinese with background in Mandarin, and lasted between 30 minutes and 1 ½ hours. Interview questions were guided by the interview schedule although the sequence of questions changed according to the flow of conversation. Interviews were transcribed in Mandarin, including breaks in the conversation and hesitations, and were then sent to parents for checking (member checking). The final transcript was then translated into English and translations were reviewed by one of three native Mandarin speakers to ensure that cultural nuances were maintained. Over the course of the interviews, the interviewer kept notes on the questions and topics discussed to improve technique and thus the quality of the data.

### ***Analysis***

Content analysis was conducted using data from translated interview transcripts and the interviewer's notes. This process began during data collection and continued after data collection was complete. Data were grouped by the content area of the question by FJL and then read and re-read to gain familiarity and form initial ideas of the data set. The researcher then coded recurring words, phrases or concepts within these areas, and integrated similar codes to define the key themes. The themes were marked within the data to allow further reflection on each thematic decision and either confirm, refute or modify the original interpretations.



## RESULTS AND COMMENTARY

Of the 23 families contacted, mothers from 14 families (61%) consented and participated in the interviews. Six families lived in Beijing or in Shandong, Shanxi or Hebei Provinces, all within 500km of Beijing. Eight families lived more than 500km away from Beijing and were from Henan or Jiangsu Province (500-1000km away); or Guangdong and Guangxi Province (>1000km away). Seven families had two children and in all but two families, the child with Rett syndrome was the eldest sibling. One family had dizygotic twins, the daughter was affected by Rett syndrome and the son was unaffected. The other family had an unaffected older son and the younger daughter was affected by Rett syndrome. Ten families lived in a city, three in towns and one family in a village. Age of the mothers ranged from 28 to 55 years and 12/14 held post-secondary qualifications. The girls with Rett syndrome were aged between 3 and 18 years at the time of the study. Individual family data are shown in Table 1. A summary of the identified thematic codes is presented in Figure 1.

### ***“My greatest wish is for her to be able to take care of herself”***

A large part of parents' recounts about daily living related to their daughter's functional skills, in particular relating to walking, feeding and communication. Of these skills, parents placed most emphasis on her ability to walk. All girls except one were able to walk independently or with some assistance (Table 1). The mother of the girl who was non-ambulatory described the difficulties in moving her daughter around the home. For the girls who could walk, families tried to maintain their daughter's skills by taking her for walks to the park or around their residential compound at least once a day. One parent said that she had previously used a wheelchair for transporting her child but that it was now too small for

her daughter and she has been unable to buy a larger one. Other families said that it was inconvenient to take the child out as she was getting bigger and was too heavy for her grandparents or parents to carry over distances. The majority of the girls were said to particularly enjoy going out to places that were bustling or “*renao*”.

“She can’t walk, and she can’t sit anymore. Usually, she just lies on the bed. It’s only during mealtimes when we bring her down and sit her on the chair... even on the wheelchair she is sometimes not really stable, she keeps slipping left and right, that kind of thing... she can only sit for a few seconds before she falls... basically she just lies in bed.” – Mrs. L

“...someone suggested that we bring her out for walks everyday. That is to say, it is also training (exercise). We try to, we cannot *not* care about her walking. If she can’t walk, it will be more troublesome, right?” – Mrs. D

Hand function, particularly in relation to the ability to self-feed was also of concern to parents. Only one parent in this group reported that her daughter had some functional hand use. This girl was able to take bread and milk from the fridge although could not extract it from the packaging. Nevertheless, this child needed assistance to feed. Parents in this study expressed the view that hand stereotypies prevented their daughter from learning to use her hands purposefully. In particular, hand stereotypies that were self-injurious in nature, such as hitting themselves on the head, were particularly concerning. In some settings, hand or elbow splints can be used to reduce the frequency of stereotypies and self-injury, [32] but this occurs on a case-by-case basis and only if better function is demonstrated. The

parents in this study said that they did not have access to such equipment although one parent had tried to make her own form of splint.

“She hits her head, claps her hands and also puts her hand in her mouth... I use a paper box and make a tube-like thing from it, make it heavy by hanging something on the paper tube, put her arm in it, and fix it at the elbow area to prevent her arm from bending, so she can’t hit (herself)...we can only limit her, try to allow her to use her hands...to stop her from harming her hands. We want her to stop hitting (herself), it’s very hard for parents (to watch).” – Mrs. D

***“If only she could speak and express what she feels in her heart”***

Generally, parents felt that their daughter understood more than she could say as has been observed in Rett syndrome [33]. Some girls and women retain the ability to say words [34] and this was described by a few parents in this group.

“Now she can still say “Mama”, but she can’t say “Dada” anymore. In this aspect, she has regressed a bit. What she could say previously, she can’t now. Sometimes when she’s scared, that’s when she calls out “Mama”...That is, if she sees anything startling, or if she fell down, or something, that’s when she will call “Mama, mama”.

Nowadays she usually doesn’t call her dad anymore. – Mrs. M

None of the parents in this group reported using augmented communication systems or technologies to assist their daughter’s communication and their daughter’s inability to

communicate effectively, particularly when she appeared to be in pain, was a source of anxiety for parents.

“When she cries, sometimes it’s because she’s sick, but we don’t know (if) it’s a stomach-ache or a headache. Because we don’t understand. Sometimes we just rub some (ointment) on her head and on her stomach (to ease the pain). We just feel that, sometimes when we rub the ointment (medicine) on her, we feel that if only she could talk. For instance, even if her intelligence isn’t that high, or whatever, these aren’t very important. If only she could speak and express what she feels in her heart.” - Mrs. B

***“Since my daughter was young until now, essentially she hasn’t stopped taking medicine”***

Parents often expressed a sense of helplessness when dealing with medical issues and said that “there is nothing I can do”. Previously collected data through the InterRett family questionnaire indicated that five of the fourteen girls had been diagnosed with scoliosis. However, only two of the five discussed scoliosis briefly during the interview. A further two families noticed that their daughter’s spine was curved but did not mention the term ‘scoliosis’.

Most girls in this group had epilepsy and were taking prescribed medications. The most common medication used was sodium valproate, also commonly prescribed in Rett syndrome in Australia [35]. One parent also reported using *tuotai*, the Chinese equivalent of topiramate, another anti-epileptic medication. Most parents whose children were taking

epileptic medications felt that their effectiveness was limited and were greatly concerned about the side effects that medications might have over the long term. However, they were unsure if they could or should reduce the dosage and were fearful that their child's condition would deteriorate if they did this. Even so, one parent had tried to reduce the number of medicines her daughter was taking but was unsure if the child's grandmother, who was caring for the child at the time, heeded her advice.

“... she's [her daughter] been taking too many medicines, it's not good for her body. So I tell her grandmother, to please reduce the medicines that she takes. Because it's useless, and the medicine she eats is bitter... Maybe now her grandmother still gives her some (liquid) supplements to help her (brain growth).” – Mrs. I

Nutrition was less of an issue for concern for parents as their daughters often ate the same foods as they did. One parent even said that she thought her daughter ate better than the rest of the family as her parents tried to buy foods that she liked and did not give her the same dishes for two or three consecutive meals. Poor growth is a common issue in Rett syndrome [36] and the Chinese parents who expressed concern in this area felt that their daughter was too skinny and “lacked muscle”. These parents felt that there was not much they could do to rectify the situation apart from increasing her dietary intake and a few children were given supplements to promote brain development and growth.

***“I would like something like that. That is to say, some place that can help her”***

Compared to their Western counterparts, a major difference in day-to-day management was the absence of structured education or rehabilitation programs. Although the evidence base is sparse, rehabilitation programs may improve the functional skills of girls and women with Rett syndrome [5] and address parents’ concern in this area. Although some special education schools and rehabilitation centres exist in China [37], efforts to enroll the child at these institutions had been unsuccessful and all girls were home-bound. Schools or rehabilitation centres that parents had contacted had sometimes refused to admit the child because of incontinence.

“We have asked around to a lot of schools, including those private ones. They all said that since she can’t manage herself (incontinent), they can’t accept her.” – Mrs. J

Other institutions were located far away from where the family lived. Furthermore, some institutions required an adult to accompany the child at all times, although some parents indicated that they would have been happy to accommodate this condition if only the school or centre was willing to accept their child.

“I’ve looked for it, a rehabilitation centre. At the rehabilitation centre, I went looking for a specialist. I wanted to know if they could help me train her, if they can, I’ll stop working and just accompany her there everyday...after looking at my child, they said that, they can’t treat [this kind of child] at that rehabilitation centre.” – Mrs. A

This parent went on to express her desire for a place that would accept her child, a sentiment that was echoed by other parents. The only family successful in enrolling their child into a rehabilitation centre subsequently became greatly dissatisfied with the quality of staff and the program and removed their daughter after a year.

“...The teachers there ... they don’t have any enthusiasm, or honour or compassion to speak of. I feel that when they help treat these kids, they just blunder through it...They usually [just] sit there and chat...” – Mrs. N

In lieu of conventional rehabilitation or education programs, parents turned to traditional Chinese medicine (TCM) and music to manage symptoms. Many families in this group had consulted with TCM clinicians prior to and after the child was diagnosed with Rett syndrome and had tried using massage or acupuncture. However, parents felt that these treatments were ineffective and discontinued them. Girls and women with Rett syndrome are generally very responsive to music and this has been used to develop therapies to encourage functional skills in some settings [38, 39]. Similarly, Chinese parents also reported that their child particularly enjoyed and responded to certain types of music, however, none of the parents indicated awareness of its use in therapies.

“One more thing is music. My child loves music, it doesn’t matter if it is day or night...She particularly likes the live shows...Because music can activate brain cell activity so I still recommend that kids listen to music more...I don’t recommend that kids take any medicine. It’s better if they just listen to music more.” – Mrs. J

***“I wanted to know what I could do to provide a good standard of living for my child.”***

Information about Rett syndrome, its cause, what to expect over time and how it can be managed was of paramount importance to the parents. For Chinese families, the main sources of information on management strategies were clinicians at Peking University First Hospital, the Internet and the online QQ family support group. The QQ group is an online instant messaging group, similar to MSN Messenger or Yahoo Chat, which was set up in 2007 and currently comprises close to 100 families from all over China. The QQ group enables families to exchange ideas and information about Rett syndrome and is the Chinese equivalent of the US-based Rettnet - an email listserv for families, moderated by the International Rett Syndrome Foundation [40]. Although information on Rett syndrome from other sources is available on the Internet, limited knowledge of the English language was a common stumbling block for parents.

“...I went online and looked up some information. But there is very little information in Mandarin on the Net...Anything about caring for her, or how to do some therapy... all of these were on English websites...I couldn't read it, because my English isn't that good (laughs).” – Mrs. K

Therefore, parents often relied on the QQ group for information. Online support groups like Rettnet and QQ can provide information and emotional support to families, particularly those whose child has been newly diagnosed with the disorder [41]. In addition to exchanging information on managing the symptoms of Rett syndrome, parents sometimes used it to express their frustrations, as one parent described.



“The parents will talk but we don’t exchange news that often. Everyone goes online to talk about their misfortune, “Sigh, my daughter is kicking a fuss again” or “The old folks at home are angry with me again, and my husband isn’t happy with me” or “My child is sick, what kind of medicine should I use” that kind of thing. To be honest, no one can help anyone.” – Mrs. N

***“They don’t care about your feelings, they stare at you with that odd look in their eyes”***

Discrimination and stigma from people around them influenced every aspect of how parents cared for their child, including the carers they employed and the daily activities they undertook with their child. Under most circumstances, having grandparents care for young children is common in China [42] and grandparents were the main carers for the child in more than half the families in this sample (Table 1). Parents relied on extended family members to provide childcare because there were few alternatives available to them. However, when the child has a disability, not all grandparents were willing to care for the child, as one parent (who had twins, one of whom was a girl with Rett syndrome) revealed.

“Her grandmother was initially very concerned for our family... after we gave birth to the kids, she helped us to care for them. Later she said that she couldn’t do it anymore... She said that as soon as she saw this daughter, this granddaughter, she will get upset, very upset... That is she couldn’t tolerate it, she couldn’t tolerate to look (at her). That’s why she, when the kids turned 3 years old, she stopped helping us care for them.” – Mrs. N

This family subsequently hired a live-in nanny to care for their children although this option was also problematic. Hiring a nanny imposed additional financial burdens on families and it was also difficult to find a suitable nanny as nannies caring for a child with a disability were at risk of being ostracized by their peers.

“Because nannies are under pressure too. If she was caring for a normal child, she will have a lot of freedom. When she goes out to chat with other nannies, she is their equal. Maybe if you bring this kind of child (child with a disability) out with you, other nannies will look down on you a bit. They will also discriminate against her...The other nannies, when they see her they too, shy away. That’s why our nanny, she will tell me, “When I go out, I’m discriminated against, so I will be in a bad mood. When I come home, I don’t even feel like eating.”” – Mrs. N

Another alternative was for the mother to stay at home and care for her child but this was not an option for many families because they relied on income from both parents for living expenses. Thus, working mothers often felt guilty because they could not care for their child as much as they would have liked. On the other hand, stay-at-home mothers felt guilty because they were not contributing to the family income and might blame the child for this situation.

“If you have this kind of child, the limitations on the parents are...large. If parents want to do something, they can’t do it anymore, she stopped it all. We have no choice. My work, also my future, it’s all ruined... I’m currently on leave; I’ve been on leave for 4 years. [My father] is particularly good; he takes quite good care of me too,

really. But I thought it over, I need to go to work, or it will be too much to care for this child (we will not make enough money to care for this child)... If we look for a carer, it's hard to find one right now and you can't find very good ones. I've been looking, I looked at carers from several hospitals, and I still have not found one. No one is willing to look after this kind of child." – Mrs. D

While stigma from within the family influenced who cared for the child, stigma from outside the family influenced family activities. Fear of being stared at and inconveniencing others influenced the timing and frequency of activities. One parent described how she would only bring her daughter out for her daily walk in the evening while other families took their daughter out less frequently because they felt uncomfortable when others stared and feared that she might cry or yell out unexpectedly.

"Usually when we bring her shopping, we seldom go to places where there are too many people. Because other people may not understand this illness, right? If other people see, they may wonder what's wrong with her, that's why we don't like to go to places where there are a lot of people; we just go for walks in the park." – Mrs. I

"If you lead her by the hand to walk, anyone can see that she isn't a normal child. Also a lot of people stare at you very rudely, right? They don't care about your feelings (aren't sensitive to your feelings), they stare at you with that odd look in their eye." – Mrs. N

Over time, parents appeared to have learned to cope with these stares and “odd looks” and said that it didn’t bother them anymore.

On the other hand, some parents also described positive encounters with more compassionate members of society. One parent described how her neighbour had offered to care for her daughter if her family was busy. Another parent said that neighbours and passer-bys were sympathetic and eager to be of help. This helped some parents to take a more positive approach to their daughter’s care and one parent even submitted an article to a relatively well-known newspaper to educate her community about Rett syndrome.

## **DISCUSSION**

For the Chinese girls with Rett syndrome in this study, a typical day involved home-based care, mostly by grandparents, with limited educational and rehabilitation options. The girls showed some variability in their functional skills, likely to have been related in part to the specific genotype [43]. Many frustrations had been experienced by these families whilst searching for a diagnosis for their daughter [44] and these frustrations continued in their current lives in terms of limited access to appropriate equipment, education and rehabilitation opportunities and poor access to information about Rett syndrome.

Those with Rett syndrome need support to optimise functional performance in daily life but there was little support for the developmental issues faced by these girls and their families. Whilst most girls and women with Rett syndrome in Australia use mobility aids and other equipment regularly [45], families in China reported little, if any, equipment use stating that it was difficult to even purchase a size-appropriate wheelchair. Alternative communication

methods include eye-gaze training, flash cards or computer-based communication boards are used in Rett syndrome [46] but were not reported in this study. This contrast was more pronounced in families' access to education and rehabilitation services. Even though China's policy stipulates that all children have a right to education, research suggests that there is a shortfall in overall availability of services with variation by region, and in practice, services are less likely to be extended to children with an intellectual disability [14]. Parents reported that access to rehabilitation services was unavailable for children who were incontinent, and this is usually the case in Rett syndrome. Factors that may hinder the availability and access of these services include limited resources, the lack of experience and training for healthcare and education professionals as well as prevailing stigma towards people with disabilities [11, 12]. Although these issues were identified specifically in a Chinese context, they are likely to apply to other developing nations as well [47].

Common co-morbidities in Rett syndrome include scoliosis, poor growth and gastrointestinal issues, and epilepsy [26]. Over two thirds of girls and women with Rett syndrome in Australia have scoliosis [48], however, families in this group only discussed the issue briefly. This contrast can be partly explained by an age bias towards younger girls in this study since girls with Rett syndrome are more likely to develop scoliosis as they grow older [48]. Two of the five families whose daughter had a diagnosis of scoliosis prior to joining InterRett discussed it during the interview. The other three families reported that their daughter with scoliosis could still walk independently. The ability to walk has been shown to be protective against the development of scoliosis in previous population-based research [48] and perhaps the scoliosis in these girls was relatively mild. A further two families had noticed scoliosis in their daughter but had not previously reported a diagnosis

of scoliosis when completing the InterRett questionnaire. This discrepancy can be explained by the time lag between previous completion of the questionnaire and the interview.

Although poor growth and nutrition is often an issue for girls and women with Rett syndrome [36], all families in this sample reported that their daughter ate well. Nearly 20% of girls and women with Rett syndrome in Australia have a gastrostomy tube [36], an intervention that was not mentioned by any family in this study and may not be routinely available in China. Epilepsy also represents a significant co-morbidity in Rett syndrome with over 80% reported to have epilepsy in a population study of girls and women with Rett syndrome [49]. The anti-epileptic medications, sodium valproate and topiramate, used by families in China, are also used in other countries [35]. Parents expressed concerns about the potential side-effects of long term use of anti-epileptic medications and a sense of helplessness saying “there is nothing I can do”. It is possible that the paucity of information on managing the symptoms of Rett syndrome that was available to the families could have contributed to this sense of helplessness.

Greater availability and access to information on potential therapies was highlighted as an area of need. Research suggests that this need is not limited to families with a child with Rett syndrome but rather is one that is felt by many families caring for a child with a disability [50, 51]. These needs may be addressed in part with specialised training for healthcare professionals and also support to existing information networks for parents. In China, these needs are beginning to be addressed. For example, dedicated funding is now provided to hospitals for the clinical care of children with disabilities, and there are specific schemes to fund knowledge dissemination in the form of workshops and seminars with

invited Chinese and foreign experts to educate clinicians about the current knowledge of causes of child intellectual disabilities. In the Kunming area, a non-government-funded association holds workshops for families with disabled children in rural and remote (Personal communication, Dr Lui Yun, Kunming).

In addition, Internet-based support groups have been shown to be a valuable source of information for parents about the management of a disorder as well as providing emotional support [41]. An Internet-based support group also has the added advantage of facilitating parents to connect with one another across wider geographical boundaries, a significant advantage for families in a country as large as China. Research shows that mothers caring for a child with Rett syndrome have poorer physical and mental health compared to the general population, possibly due to the high level of care required by their daughter [52]. Although informal support networks such as the QQ group can help to support the emotional and mental well-being of these mothers, it may be insufficient in filling the gaps which are due to the absence of other structured support networks. To further develop support for families with a daughter with Rett syndrome, one of the participants in the current study has recently spearheaded the formation of the Rett Family Care Centre. Its goals are to increase awareness and respect for those with Rett syndrome, encourage the development of treatments for the disorder, and provide support for families [53]. Advocacy efforts include active participation in media reports: there is much to be achieved.

Another overarching theme related to encounters with stigma. Cultural beliefs and limited opportunities to interact with people with disabilities are likely to play a role in prevailing stigma, not just in China but potentially other developing nations as well such as India [54].

Stigma towards people with disabilities has the potential to hinder the use of existing services and restrict future plans for additional support services. However, there were reports of positive experiences with “more compassionate” members of society highlighting the importance of public awareness relating to disability [55]. Increased awareness of disabilities goes hand-in-hand with the provision of support services that families need. Government-funded programs and community-based rehabilitation programs have the potential to change negative attitudes and encourage greater integration of Rett syndrome children into mainstream society whilst also meeting the needs of people with a disability [18, 51]. Although programs with funding for clinical centres and families are beginning to be adopted in China (personal communication with Chinese clinicians and observations by JL in China), there remains much to do before this ideal is achieved [18].

Participants in this study were limited to mothers of a child with Rett syndrome with one exception. One parent asked that the child’s grandmother also be interviewed as the grandmother was the one who cared for the child on a daily basis. The information provided by this grandparent was similar to that provided by the mothers in other families. It is common for grandparents in China to care for young children while their parents were at work, however, the role of grandparents in childcare is a neglected area of research that deserves further examination [42]. Further, most of the mothers had post-secondary school qualifications. It is likely that this was a more resourced group of families and we could be underestimating the difficulties encountered in daily life for girls with Rett syndrome in China. As this was an exploratory study, some aspects of daily living were not as thoroughly examined, for instance, sibling relationships and coping with stigma. Additional in-depth discussion relating to the management of co-morbidities could also be useful. Moreover,



this sample does not capture the full range of variability in Rett syndrome, for instance, in relation to functional skills, as to do so would require a much larger number of participants. Further information on these areas would add depth to our understanding of families' experiences.

Despite these limitations, this study has started to address some of the priority research questions identified by the international community [56] and contributed new information to our understanding of parents' experiences in caring for a child with severe intellectual and physical disability in China. Our methodology included careful development of the interview schedule, translation of the data and member checking to ensure cultural sensitivity. We have included population data on Rett syndrome to support the story provided by our Chinese families. We found that families perceived a lack of support services and limited access to information to adequately meet the needs of their child, further exacerbated by discrimination and stigma from some members of the community. Better access to education, rehabilitation and appropriate equipment could not only reduce the burden of care on families [57] and future healthcare care costs [58] but also contribute to a better quality of life for the child with Rett syndrome and her family. The development of the Rett Family Care Centre will contribute to achieving these goals.

## **ACKNOWLEDGEMENTS**

The authors would like to acknowledge all the families who have participated in this study. We would also like to acknowledge the contributions of Jenny Song and Lisa Yang with translation of interview transcripts. Special thanks to members of the Australian Rett Syndrome Community Reference Group who generously donated their time to giving feedback to interview questions.

**DECLARATION OF INTERESTS:** We would also like to acknowledge the International Rett Syndrome Foundation who provides funding for the International Rett Syndrome Phenotypic Database. Helen Leonard's current funding is from an NHMRC Senior Research Fellowship #572568. There are no known conflicts of interest for each of the authors of this paper.

## **REFERENCES**

- [1] Leonard H, Petterson B, Bower C, Sanders R. Prevalence of intellectual disability in Western Australia. *Paediatric and Perinatal Epidemiology* 2003;17:58-67.
- [2] Durkin M. The epidemiology of developmental disabilities in low-income countries. *Mental Retardation and Developmental Disabilities Research Reviews* 2002;8(3):206-11.
- [3] Leonard H, Wen X. The epidemiology of mental retardation: Challenges and opportunities in the new millenium. *Mental Retardation and Developmental Disabilities Research Reviews* 2002;8:117-34.

[4] Fujiura GT, Park HJ, Rutkowski-Kmitta V. Disability statistics in the developing world: A reflection on the meanings in our numbers. *Journal of Applied Research in Intellectual Disabilities* 2005;18(4):295-304.

[5] Larsson G, Engerström IW. Gross motor ability in Rett syndrome – the power of expectation, motivation and planning. *Brain and Development* 2001;23:S77-81.

[6] McConkey R, Truesdale-Kennedy M, Chang M-Y, Jarrah S, Shukri R. The impact on mothers of bringing up a child with intellectual disabilities: A cross-cultural study. *International Journal of Nursing Studies* 2008;45(1):65-74.

[7] Vaidya S. A sociological study of families with autism in Delhi: Issues and Challenges. In. *A sociological study of families with autism in Delhi: Issues and Challenges*. Asian Pasific Autism Conference; 21 August 2009; Sydney, Australia; 2009.

[8] Pearson V, Chan TWL. The relationship between parenting stress and social support in mothers of children with learning disabilities: A Chinese experience. *Social Science and Medicine* 1993;37(2):267-74.

[9] CIA - The World Factbook -- China [Internet]. Washington, DC: Central Intelligence Agency; 5 March 2010 - [cited 25 March 2010]; Available from: <https://www.cia.gov/library/publications/the-world-factbook/geos/ch.html>.

- [10] Ravallion M, Chen S. China's (uneven) progress against poverty. *Journal of Development Economics* 2007;82(1):1-42.
- [11] Su H, Dyke DCV. Breaking the silence and overcoming the invisibility: Down Syndrome in China Part I. *International Pediatrics* 2005;20(1):25-33.
- [12] Shang X, Fisher KR, Xie J. Discrimination against children with disability in China. *International Journal of Social Welfare* 2011;20(3):298-308.
- [13] Xiong N, Yang L, Yu Y, Hou J, Li J, Li Y, Liu H, Zhang Y, Jiao Z. Investigation of raising burden of children with autism, physical disability and mental disability in China. *Research in Developmental Disabilities* 2011;32(1):306-11.
- [14] McLoughlin CS, Zhou Z, Clark E. Reflections on the development and status of contemporary special education services in China. *Psychology in the Schools* 2005;42(3):273-83.
- [15] Wong VC, Hui SL. Epidemiological study of Autism Spectrum Disorder in China. *Journal of Child Neurology* 2008;23(1):67-72.
- [16] Chen K, Xu H. The integration of traditional Chinese medicine and Western medicine. *European Review* 2003;11(02):225-35.

- [17] Xu D, Sun B, Wan X, Ke Y. Reformation of medical education in China. *Lancet* 2010;375:1502-04.
- [18] Chung EY-H, Packer T, Yau M. When East meets West: community-based rehabilitation in Chinese communities. *Disability and Rehabilitation* 2011;33(8):697-705.
- [19] Amir RE, Van den Veyver IB, Wan M, Tran CQ, Francke U, Zoghbi HY. Rett syndrome is caused by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2. *Nature Genetics* 1999;23(2):185.
- [20] Fehr S, Bebbington A, Nassar N, Downs J, Ronen G, de Klerk N, Leonard H. Trends in the diagnosis of Rett syndrome in Australia. *Pediatric Research* 2011;70:313-19.
- [21] Didden R, Korzilius H, Smeets E, Green V, Lang R, Lancioni G, Curfs L. Communication in Individuals with Rett Syndrome: an Assessment of Forms and Functions. *Journal of Developmental and Physical Disabilities* 2010;22(2):105-18.
- [22] Renieri A, Mari F, Mencarelli MA, Scala E, Ariani F, Longo I, Meloni I, Cevenini G, Pini G, Hayek G, Zappella M. Diagnostic criteria for the Zappella variant of Rett syndrome (the preserved speech variant). *Brain and Development* 2009;31(3):208-16.
- [23] Downs J, Bebbington A, Jacoby P, Williams A-M, Ghosh S, Kaufmann WE, Leonard H. The level of purposeful hand function is a marker for clinical severity in Rett syndrome *Developmental Medicine and Child Neurology* 2010:16.

[24] Carter P, Downs J, Bebbington A, Williams S, Jacoby P, Kaufmann WE, Leonard H. Stereotypical hand movements in 144 subjects with Rett syndrome from the population-based Australian database. *Movement Disorders* 2010;25(3):282-88.

[25] Fehr S, Bebbington A, Ellaway C, Rowe P, Leonard H, Downs J. Altered attainment of developmental milestones influences the age of diagnosis of Rett syndrome. *Journal of Child Neurology* 2011;26:980-87.

[26] Neul JL, Kaufmann WE, Glaze DG, Christodoulou J, Clarke AJ, Bahi-Buisson N, Leonard H, Bailey MES, Schanen NC, Zappella M, Renieri A, Huppke P, Percy AK. Rett syndrome: Revised diagnostic criteria and nomenclature. *Annals of Neurology* 2010;68(6):944-50.

[27] Fyfe S, Cream A, de Klerk N, Christodoulou J, Leonard H. InterRett and RettBASE: International Rett Syndrome Association databases for Rett syndrome. *Journal of Child Neurology* 2003;18(10):709-13.

[28] Patton MQ. *Qualitative research and evaluation methods*. 3rd ed. Thousand Oaks, California: Sage Publications.

[29] StataCorp. *Stata Statistical Software*. In. *Stata Statistical Software*; Collage Station, TX; 2007.

[30] Laurvick CL, de Klerk N, Bower C, Christodoulou J, Ravine D, Ellaway C, Williamson S, Leonard H. Rett syndrome in Australia: A review of the epidemiology. *Journal of Pediatrics* 2006;148(3):347-52.

[31] McKenzie A, Hanley B. Consumer and community participation in health and medical research: A practical guide for health and medical research organisations. Perth: The University of Western Australia & Telethon Institute for Child Health Research.

[32] Naganuma GM, Billingsley FF. Effect of hand splints on stereotypic hand behavior of three girls with Rett syndrome. *Physical Therapy* 1988;68(5):664-71.

[33] Sandberg AD, Ehlers S, Hagberg B, Gillberg C. The Rett syndrome complex. *Autism* 2000;4(3):249-67.

[34] Zappella M, Gillberg C, Ehlers S. The Preserved Speech Variant: A subgroup of the Rett complex: A clinical report of 30 cases. *Journal of Autism and Developmental Disorders* 1998;28(6):519-26.

[35] Jian L, Nagarajan L, Klerk Nd, Ravine D, Christodoulou J, Leonard H. Seizures in Rett syndrome: An overview from a one-year calendar study. *European Journal of Paediatric Neurology* 2007;11:310-17.

[36] Oddy WH, Webb KG, Baikie G, Thompson SM, Reilly S, Fyfe SD, Young D, Anderson AM, Leonard H. Feeding experiences and growth status in a Rett syndrome population. *Journal of Pediatric Gastroenterology and Nutrition* 2007;45(5):582-90.

[37] Ellsworth NJ, Zhang C. Progress and challenges in China's special education development. *Remedial and Special Education* 2007;28(1):58-64.

[38] Wigram T, Lawrence M. Music therapy as a tool for assessing hand use and communicativeness in children with Rett Syndrome. *Brain and Development* 2005;27(Supplement 1).

[39] Yasuhara A, Sugiyama Y. Music therapy for children with Rett syndrome. *Brain and Development* 2001;23(Supplement 1):S82-S84.

[40] RettNet - rettsyndrome.org [Internet]. International Rett Syndrome Foundation; 2008 - [cited 16 November 2010]; Available from: <http://www.rettsyndrome.org/online-communities/online-communities/rettnet.html>.

[41] Leonard H, Slack-Smith L, Phillips T, Richardson S, D'Orsogna L, Mulroy S. How can the Internet help parents of children with a rare neurological disorder? *Journal of Child Neurology* 2004;19(11):902-07.



[42] Goh ECL. Raising the precious single child in urban China-An intergenerational joint mission between parents and grandparents. *Journal of Intergenerational Relationships* 2006;4(3):6-28.

[43] Bebbington A, Anderson A, Ravine D, Fyfe S, Pineda M, de Klerk N, Ben-Zeev B, Yatawara N, Percy A, Kaufmann WE, Leonard H. Investigating genotype-phenotype relationships in Rett syndrome using an international data set. *Neurology* 2008;70(11):868-75.

[44] Lim F, Downs J, Li J, Bao X-H, Leonard H. Barriers to diagnosis of a rare neurological disorder in China—Lived experiences of Rett syndrome families. *American Journal of Medical Genetics Part A* 2012;158A(1):1-9.

[45] Urbanowicz A, Downs J, Bebbington A, Jacoby P, Girdler S, Leonard H. Use of equipment and respite services and caregiver health among Australian families living with Rett syndrome. *Research in Autism Spectrum Disorders* 2011;5:722–32.

[46] Sigafoos J, Green VA, Schlosser R, O'Eilly MF, Lancioni GE, Rispoli M, Lang R. Communication intervention in Rett syndrome: A systematic review. *Research in Autism Spectrum Disorders* 2009;3(2):304-18.

[47] Maulik PK, Darmstadt GL. Childhood disability in low- and middle-income countries: Overview of screening, prevention, services, legislation, and epidemiology. *Pediatrics* 2007;120(Supplement 1):S1-55.

[48] Ager S, Fyfe S, Christodoulou J, Jacoby P, Schmitt L, Leonard H. Predictors of scoliosis in Rett syndrome. *Journal of Child Neurology* 2006;21(9):809-13.

[49] Jian L, Nagarajan L, Klerk Nd, Ravine D, Bower C, Anderson A, Williamson S, Christodoulou J, Leonard H. Predictors of seizure onset in Rett syndrome. *Journal of Paediatrics* 2006;149:542-7.

[50] Wong S, Wong TK, Martinson I, Lai AC, Chen W, He Y. Needs of Chinese parents of children with developmental disability. *Journal of Learning Disabilities* 2004;8(2):141-58.

[51] Wang P, Michaels CA. Chinese Families of Children with Severe Disabilities: Family Needs and Available Support. *Research & Practice for Persons with Severe Disabilities* 2009;34(5):21-32.

[52] Laurvick CL, Msall ME, Silburn S, Bower C, de Klerk N, Leonard H. Physical and mental health of mothers caring for a child with Rett syndrome. *Pediatrics* 2006;118(4):e1152-64.

[53] [Internet]. - [cited Available from: <http://blog.sina.com.cn/u/1936766310>].

[54] Edwardraj S, Mumtaj K, Prasad JH, Kuruvilla A, Jacob KS. Perceptions about intellectual disability: a qualitative study from Vellore, South India. *Journal of Intellectual Disability Research* 2010;54(8):736-48.

[55] Wang J-B, Guo JJ, Yang L, Zhang Y-D, Sun Z-Q, Zhang Y-J. Rare diseases and legislation in China. *The Lancet* 2010;375(9716):708-09.

[56] Tomlinson M. Research priorities for health of people with disabilities: an expert opinion exercise. *Lancet* 2009;374(9704):1857.

[57] Ostensjo S, Carlberg EB, Vollestad NK. The use and impact of assistive devices and other environmental modifications on everyday activities and care in young children with cerebral palsy. *Disability and Rehabilitation* 2005;27(14):849-61.

[58] Hendrie D, Bebbington A, Bower C, Leonard H. Measuring use and cost of health sector and related care in a population of girls and young women with Rett syndrome. *Research in Autism Spectrum Disorders* 2011;5:901-09.

## Caring for a child with severe intellectual disability in China: An example of Rett syndrome

**Table 1: Demographic information for participating families**

Family ID	Age of mother (y)	Highest education level of mother	Municipality	Child age at interview (y)	Level of mobility	Main carer (daytime)
A	55	Trade/technical certificate	City	18	Walk with assistance	Grandparent; aunt
B	37	High school	Town	15	Walk with assistance	Grandparents; aunt
C	28	Tertiary qualification	City	5	Walk with assistance	Grandparents
D	31	Tertiary qualification	Village	6	Walk with assistance	Grandparents
E	33	Tertiary qualification	Town	4	Walk independently	Grandparents
F	32	Trade/technical certificate	City	5	Walk with assistance	Grandparents
G	36	Tertiary qualification	City	7	Walk with assistance	Nanny <sup>a</sup>
H	34	Tertiary qualification	Town	9	Walk independently	Grandparents
I	31	High school	City	8	Walk independently	Grandparents
J	34	Tertiary qualification	City	8	Walk independently	Mother
K	36	Tertiary qualification	City	7	Non-ambulant	Grandparents
L	38	Trade/technical certificate	City	11	Walk independently	Nanny
M	Unknown	Tertiary qualification	City	15	Walk independently	Mother
N	42	Tertiary qualification	City	6	Walk independently	Nanny

<sup>a</sup> Grandparents also provided care on a regular basis

**Figure 1: Thematic codes used in this study**

