This is a pre-copy edited, author-produced version of an article accepted for publication in Applied Ergonomics following peer review.

The version of record is available online at: 10.1016/j.apergo.2020.103095

Citation:

Hay, G. J., Klonek, F. E., & Parker, S. K. (2020). Diagnosing rare diseases: A sociotechnical approach to the design of complex work systems. *Applied Ergonomics, 86*. https://doi.org/10.1016/j.apergo.2020.103095

# Diagnosing rare diseases: A sociotechnical approach to the design of complex work systems

*Hay, G.J. and Klonek, F.E. and Parker, S.K.*

## 1. Introduction

Delivering effective treatment and patient management is a primary goal of health care organisations (Davis et al., 2014). As such, patients without a diagnosis challenge the health care system. Without a diagnosis, patients suffer from suboptimal and poorly co-ordinated treatment, and it is more challenging for them to receive financial and social support. Unfortunately, being undiagnosed is a common problem for patients suffering from rare, multi-system disorders. While a rare disease, by definition, is uncommon (e.g., some have a prevalence of 1 in one million; Wakap, 2019); collectively, there are 5,000 to 8,000 different rare diseases which affect almost one in ten people during their lifetime (Aymé & Rodwell, 2014). Consequently, building a better system for diagnosing and managing these patients represents a global challenge (cf. Davis et al., 2014).

The multi-system nature of rare diseases means that they typically effect multiple physiological systems (e.g. endocrine, cardiopulmonary, and urology). Consequently, the diagnosis and treatment of rare diseases requires expertise from multiple clinical specialties (e.g., endocrinologists, cardiologists, and urologists). However, the structure of clinical knowledge within the health system is notoriously siloed (Meneses & Caseiro, 2018). Consequently, patients often embark on what is known as a 'diagnostic odyssey', in which they feel like no single practitioner is looking at them 'as a whole' (Molster et al., 2016; p. 8). In Australia, research suggests that approximately 30% of patients with rare diseases wait more than five years to reach a diagnosis (and in 30% of cases, it takes 5 to upwards of 20 years), 30% of patients see more than six specialists, and 50% of patients receive at least one incorrect diagnosis (Molster et al., 2016). The resulting "chaos that coexists with being undiagnosed" (Spillmann et al., 2017; p. 1) is very frustrating for patients and their families. Furthermore, managing patients with rare diseases is also costly, with research estimating an annual cost of AUD395 million to the health system. In some states, this means that the 2% of the population with a rare disease account for about 10.5% of hospital inpatient expenditure (Walker et al., 2017). While it is difficult to estimate exactly, it is likely that the duplicative, resource-intensive nature of the diagnostic process also bears a significant cost to health systems.

The inefficiency of the current system of managing patients who must 'move' between multiple clinical specialists and various healthcare sub-systems (cf. Waterson et al., 2018) exemplifies the need for better patient *care transitions* (i.e., instances in which patients have to move from one healthcare sub-system to another; Coleman, 2003). Effective care transitions require seamless continuity of information and coordination amongst clinical specialists (Australian Medical Association, 2006; McDonald et al., 2007) – which is rarely achieved in practice (Department of Health, 2019). The inherent complexity of diagnosing rare diseases means that it is a particularly 'extreme case' (cf. Bell et al., 2018) of care transitions, from which much can be learnt.

In the current study, we examine an effort to re-design the work system for diagnosing rare disease patients in an Australian state. We seek to understand how such complex systems within healthcare can be redesigned to reduce, or more effectively manage, these care transitions. By 'work system', we mean a set of social components (e.g., people with different skills) and technical components (e.g., technologies, tools) that are highly interconnected, and also affected by the external environment (Davis, 2014). We conduct a sociotechnical systems (STS) analysis using two frameworks. First, we compare the work systems before and after the program initiation (cf. Holden et al., 2013). Second, we analyse the process stages of the system re-design (cf. Clegg, 1988), and identify insights regarding the effective design, implementation, and operation of such new socio-technical systems.

Next, we elaborate on our research aims and use of existing theory, followed by an introduction to the research context.

## 2. Research Aims and Theory

At its core, socio-technical systems (STS) theory advocates for the joint design and optimisation of social and technical system components, on the basis that "most contemporary problems in society are systemic in their origin" (Davis et al., 2014, p. 16). This systems-level perspective entails analysing the complex interconnections between the multiple social and technical components that exist in a given work system, and their effect on work-related outcomes (for a detailed overview, see Davis et al., 2014).

An STS perspective is particularly useful in the context of the complex, non-linear nature of the healthcare sector. For example, work within hospitals connects multiple complex sub-systems and different employee groups (e.g., doctors, nurses, administrative and policy staff) who operate within these sub-systems (Werner & Holden, 2015; Braithwaite, Clay-Williams, Nugus, & Plumb, 2013; Carayon & Friesdorf, 2006; Janowitz et al., 2006). Consequently, in the domain of healthcare, STS has already contributed to a better understanding of the system factors that affect care transitions (Waterson et al., 2018) – for example, by identifying coordination behaviours and information systems that are crucial for effective care transitions (Bisantz et al., 2012; McDonald et al., 2007). Here, we further extend this literature by applying it to the context of the work system for the diagnosis and management of rare, multi-system disorders – and the re-design of this work system – thus addressing two important research questions (see Figure 1).

***Insert Figure 1 about here***

First, we examine **how re-designing diagnostic work affects the larger work system.** To do this, we compare the components and configuration of the work system before and after its re-design, and the outcomes of each configuration. To model these configurations, we used the approach proposed in the Systems Engineering Initiative for Patient Safety (SEIPS) 2.0 model (Holden et al., 2013). The SEIPS model is a specific STS framework originating in the human factors and ergonomics literature (Carayon et al. 2006, 2013) that has been tailored to, and well-used in, studies of healthcare (see Holden et al., 2013, for an overview). Another advantage of using the SEIPS model as the framework for this study is that, in comparison to other frameworks (e.g. Davis et al., 2014), the SEIPS includes the element of multi-organisation (or cross-cutting) systems – a characteristic feature in many domains of healthcare work, including in the work of rare disease diagnosis.

According to this model, work systems can be described by six interrelated components: persons, tasks, tools/technologies, organisation, internal environment and external environment. Applied to our research context, the component *persons* reflects the knowledge, skills, and abilities of the clinicians involved in diagnosing and managing patients with rare diseases. The component *tasks* reflects the work design (i.e., "the content and organization of work tasks, activities, relationships, and responsibilities", Parker, 2014, p. 662) of the clinicians. The component *organisation* reflects "the structures external to a person (but often put in place by people) which organise time, space, resources and activity" (Holden et al., 2013, p. 1672), and in this case, refer to hospital work structures. The component *tools and technology* reflects the "usability, accessibility, familiarity, level of automation, portability and functionality" of tools and technology in the system (Holden et al., 2013, p. 1672). The *internal environment* reflects the physical working conditions (e.g., light, noise, or available working

space), while *external environment* reflects "macro-level societal, economic, ecological and policy factors outside an organisation" (Holden et al., 2013, p. 1673). These six components of the work systems are interrelated, and changes in one component can affect any other (or multiple) component(s) at any given time. In the current study, our aim was to understand how different configurations of these components (before versus after the re-design) affected the processes and outcomes of diagnostic work.

Second, we examine the **multi-level factors influencing the re-design process,** distinguishing between the three stages proposed by Clegg (1988): (1) the Design stage, in which central actors (i.e., the *designers*) make key decisions about the novel work system[1]; (2) the Implementation stage, in which the new system is put into practice; and (3) the Operation stage, which focuses on the ongoing operation of the re-designed work system, including its sustainability. We use this model to identify the multi-level factors (i.e., barriers and enablers) that operate at each stage of design to affect its success.

Altogether, our research offers important contributions research and practise. Through exploring the redesign of care transitions within the novel context of the rare disease diagnosis work system, we unearth critical insights into the factors – both structural (Holden et al., 2013) and processual (cf. Clegg, 2000) – that influence the effective design of trans-organisational work systems in healthcare, drawing on insights from the organisational psychology and work design literature (cf. the call made by Clegg et al., 2017).

## 3. Methods

### *3.1 Research Context*

The re-design of the rare diseases work system was initiated by a team of geneticists and policymakers working in an Australia state, who were dissatisfied with the inefficiencies in the existing work system. Since 80% of rare diseases are associated with genetic factors (Department of Health, 2015), clinical genetic expertise is often heavily required in the management of rare disease patients – hence the central involvement of genetics specialists in this context.

All of the key stakeholders involved in the re-design worked across various parts of the healthcare system – in policy, as well as private and public hospitals/institutions. The Program Director and the program's genetic counsellor worked in the clinical genetics department of a public hospital, which provides state-wide genetic services. The policymakers worked in a unit of the state's Health Department that had a strategic focus on genetic disorders. Other key program stakeholders (e.g., hospital managers, executives, and clinicians) worked in: public metropolitan hospitals that hosted various clinical specialists; a state-wide pathology laboratory that specialised in genetic testing; and a private research facility that offered additional funding and infrastructure for the program.

[1] Compared to prototypical STS studies –in which criteria for the design are explicitly identified, and design options are judged against this criteria, all with the use of various STS tools and principles (see Baxter & Sommerville, 2011, for a brief review) – the STS design process that we have described is somewhat different. In this case, STS analysis was not conducted by the program directors; nor was there any use of STS tools. Instead, in this study, our retrospective analysis of the design and implementation process identified that many STS principles were adhered to intuitively (and unintentionally) – which we were able to uncover, post hoc, through the lens of STS theory. Thus, we, the authors, were passive observers, and not involved in influencing the approach to change. Further, due to a version of the program already operating in the United States, there was not a need for the program to be designed, per se, 'from scratch'. Instead, the program was adapted to suit the logistical nuances of the Australian context and the resources available to implement the program locally.

The re-design itself constituted the introduction of a state-wide program with the objective to facilitate better cross-specialty collaboration, thus improving the diagnosis and management of rare disease patients. This re-design changed both the social and technical structures of the existing work system – which we describe in more detail in the Findings sections (4.1 and 4.2).

***Data collection and analysis***

Using a two-year longitudinal design, we analysed the re-design of this work system using multi-source, qualitative data (i.e., archival documents, interviews, and on-site observations).

To familiarise ourselves with the existing work system, and its historical origin and context, we collected and analysed archival documents (e.g., clinical protocols, meeting minutes, workshop notes).

We invited key stakeholders in the re-design for interviews, including clinicians who subscribed to invitations to the Expert Panel meetings ($N = 58$, at the time of the study), as well as key members of the genetics policy unit and partner institutions. In total, we interviewed 29 key stakeholders from across all 7 organisations involved in the program, including seven geneticists, thirteen non-geneticist clinicians (some of which were hospital executives), three laboratory scientists, two individuals from the private research facility, and four stakeholders involved in policy making. Each interviewee reflected on their experiences of the work system re-design across the design, implementation, and operation stages. For the full interview protocol, please see Appendix A.

To immerse ourselves in the program, understand the perspectives of the key stakeholders, and observe the work system re-design in action, we also regularly shadowed participants during the Expert Panel meetings (Quinlan, 2008).

The data analysis consisted of a combination of template analysis (King, 1998; see Waterson, 2014) and iterative thematic analysis (see Høyland et al., 2019). To answer the first research question about how the systems were configured, we compared the system components and their outcomes, before and after the re-design, using the SEIPS 2.0 as an STS template (Holden et al., 2013). We also drew on work design theory (Parker, 2014) to provide a more detailed account of the specific task characteristics of the clinicians' work, relying on the motivational, knowledge, and social dimensions from the expanded work characteristics model (Morgeson & Humphrey, 2006; Humphrey, Nahrgang, & Morgeson, 2007) to describe the nature of clinicians' tasks, responsibilities and relationships with others. Based on our regular field observations and interviews, we estimated the relative level of these work design dimensions in terms of "high" versus "low" levels (bottom of Figure 2).

To answer our research question about the re-design process, we organised our data into the stages of Design, Implementation, and Operation (using a model from Clegg, 1988, as a template). Following this, we identified *Barriers*, which we defined as factors impeding the design process, and *Enablers*, or factors that mitigate the negative impact of specific barriers. We also identify *Accelerators*, or higher-level system characteristics that have a positive, progressive impact on the re-design process, as well as *Motivators,* defined as person-level characteristics that have a positive impact on the re-design process.

For further detail about our data collection and analysis, please see Appendix B.

In what follows, we first present our findings regarding the structure of the work system before and after the re-design, to give the reader a detailed understanding of the nature of these systems. We then report on the nature of the re-design process that transformed the former system into the latter.

## 4. The effect of the re-design on the work system configuration

To address research question 1, we compared the configuration of the work system components before and after the re-design (Figure 2.).

***Insert Figure 2 about here***

### *4.1 Work system configuration before the re-design*

The work system for the diagnosis of rare diseases prior to the change was no different from that which supported the diagnosis and treatment of diseases more generally, which involved patients being seen by a specialist who then operated in relative isolation from other specialists. More specifically, in the current case of rare diseases, patients' symptoms often necessitated the involvement of a number of specialists, each of whom operated in isolation – often at different hospitals and laboratories, with their own records of patient information. When specialists communicated with each other, they did so via short e-mails or by sending physical files filled with sometimes hundreds of pages of historical patient information. The patient then moved from one specialist to the other, each of whom focused on their specialty area of expertise. Further, the specialists were often under a level of time pressure that did not allow them to take the time required to comprehensively review the patients' history, as well as the most recent scientific literature. Further, although rare diseases often have a genetic basis, this genetic expertise was also siloed and perceived as difficult to access by most specialists. Although this system of work functioned reasonably effectively for the diagnosis of patients with common diseases due to them requiring the expertise of far fewer specialists, with less complicated diagnostic work, the individual-specialist approach was rarely effective for patients with rare diseases.

In what follows, we describe each of the components of the work system in more detail.

For the *technology* component of the old system, digital information was often only available within each speciality. Clinicians mentioned that databases were highly specialised and poorly integrated, leading to miscommunication and duplication between specialties (*"[we are] lucky to see a few of the results that endocrine (another specialty) ordered; so there's a lot of doctors who do the same thing over and over again"* – Clinician).

With respect to *tasks*, in the old work system, some motivational job characteristics were characterized as high (i.e., *task significance*), and others as low (i.e., *task identity* and *feedback from the job*). High task significance (i.e., the extent to which a job influences the lives of others) stands out in this context; providing diagnoses for rare disease patients can significantly transform the quality of their lives. However, the separation of clinical experts across multiple hospitals meant that *task identity* (i.e., the extent to which the job reflects a whole piece of work) was rather low within the old system, and tasks were described by interviewees as disjointed. For example, diagnostic tasks were described as being split across multiple speciality silos: *"They'll see an endocrinologist for that problem, and they'll see a neurologist for that problem – but these two doctors don't talk to each other, so they don't get holistic treatment, because they're not seen as a whole person, they're seen as their individual body parts"* (Policy officer). Undiagnosed patients in the old system were *"managed*

*in an ad hoc way, so different specialties just get asked to deal with them as their problems occurred"* (Clinician). Furthermore, clinicians reported that they received very little (to no) *feedback from the job* of working with rare disease patients. One clinician reported that it could take months to get answers for these cases ("*There'll be a three to six month gap before you see them again and then you sit with your symptoms for another three to six months*" – Clinician) and that, in some cases, they never learn about the effect of their clinical contribution on the patient.

In terms of the knowledge characteristics, work of clinicians (in the old system) was characterized by extremely high levels of *knowledge demands*. For example, most clinicians pointed out that diagnosing rare diseases is an extremely difficult job with very high levels of *job complexity* (Edwards, Scully, & Brtek, 2000). We observed multiple instances in the Expert Panel meetings in which clinicians had to process excessive amounts of information (i.e., piles of documents documenting patient assessment histories). Finally, rare diseases were characterized as non-routine events within clinical practice that required clinicians to engage in *problem-solving* and to generate unique solutions (Wall & Jackson, 1995).

In terms of social characteristics, the old work system was characterized as *sequentially interdependent* (i.e., low levels of interdependence; Saavedra, Earley, & Van Dyne, 1993); that is, diagnostic activities flowed sequentially from one specialist to another (see Figure 2, top left) – and mostly in one direction. Due to this organisational segregation, the old system also provided lower levels of *social support*, whereby clinicians could not as easily ask others for advice or support (Karasek et al., 1998), and thus received low levels of *feedback* from other specialists.

Finally, the *coordination demands* for managing rare disease patients were very high; clinicians had to collate patient information (e.g., test results) from multiple locations (or technical systems), which they described as a significant administrative burden on their workload. Clinicians spoke negatively about this bureaucracy and suggested that much of their time and cognitive capacity was spent searching for patient information and test results, and doing general clerical work – lessening the time they had available to work on complex patient cases in-depth.

In terms of the *Organisation* of the old system, interviewees across both clinical practice and policy highlighted the 'fragmentation' of the health care system into specialty silos. This fragmentation also reflects the *internal environment* of the system, being dispersed within and across: multiple hospital sites, multiple interdisciplinary partners (including government services), private and public facilities, as well as the Department of Health more broadly.

This work system configuration resulted in a low diagnostic rate for rare disease patients (cf. Walker et al., 2017; Molster et al., 2016), which led to frustration among clinicians, substantial workflow inefficiencies, and little access for clinicians to informal professional networks (e.g., *"a lot of the human interactions and the efficiencies of the stuff you used to do in the corridor when you walked past someone, didn't happen anymore"*, Program Director).

### *4.2 Work system configuration after the re-design*

In stark contrast to the fractured, piecemeal nature of diagnostic work in the pre-change work system, the UDP system of work was a system that functioned in a more collaborative and integrative way. The key change in the social work structures to achieve this collaboration and integration was the introduction of regular Expert Panel meetings. In these meetings, a panel of interdisciplinary participants (specialists, generalist paediatricians, and laboratory scientists) have comprehensive, in-depth discussions of currently undiagnosed patients, to identify (or work towards) a potential

diagnosis, and decide on further clinical assessments. These meetings brought together the previously-siloed clinical expertise and laboratory capabilities, and were facilitated by the Program Director. The key change in the technological work structures was the introduction of an online knowledge platform ('Patient Archive') to manage scientific and patient-related information – which further served to integrate and streamline the diagnostic process. Finally, another key change – at the interface of the social and technical structures of this work system – was the introduction of a Nurse Coordinator, who provided administrative support (i.e., collating patient information, scheduling meetings), and who helped to co-ordinate patients going through the program (i.e., managing referrals, coordinating tests, counselling patients and their families). In doing so, the Nurse Coordinator mitigated some of the administrative demands of diagnosing patients with rare diseases (who often had long medical histories) that was previously placed on the clinicians.

In what follows, we describe the components of the work system that were most significantly changed in more detail.

With respect to *Technology*, the re-design introduced a novel digital repository for patient-related information, which centralised information that was previously spread across multiple I.T. systems. This tool was used in a way that reduced the excessive *knowledge demands* (in particular, information processing demands) of the diagnostic work, in addition to increasing knowledge sharing and interdependence (via an online forum which allowed clinicians absent from the expert panel meetings to contribute; see also the *Task* components).

With respect to the *Task* characteristics, the re-design affected multiple dimensions of the clinicians' work design (see the bottom of Figure 2). In terms of motivational work characteristics, the Expert Panel meetings increased the extent to which clinicians experienced *task significance* within their jobs. First, successes in diagnosing patients were frequently shared within those meetings. Second, since knowledge was pooled within the meetings, rather than only being involved in part of the patient's treatment, clinicians were able to see and participate in the entire piece of work (i.e., the case) from beginning to end. Third, the re-designed work system provided clinicians with frequent feedback about test results that individuals had suggested, thereby promoting higher levels of *feedback from the job.* For example, one interviewee mentioned that the re-design allowed almost *"real-time assessment"* (Biochemist).

With respect to the knowledge demands, even after the re-design, the diagnostic work was still highly complex and required clinicians to generate unique solutions to challenging patient cases. However, the new Nurse Coordinator role significantly reduced the magnitude of the information processing demands for clinicians. While high knowledge demands are usually considered a positive aspect of a job (Humphrey et al., 2007), the demands can be excessive. Reducing the high levels of knowledge demands in this context thus had beneficial consequences for the clinicians.

The re-design substantially affected the social characteristics of the work. First, the panel meetings created a form of *intensive interdependence* (i.e., high interdependence; Saavedra, Earley, & Van Dyne, 1993), because specialists were required to diagnose, problem-solve, and collaborate as a team – all of which was centred on one patient. In this context, multiple clinicians reported that the meetings provided *social support*. In our field observations, we noted multiple instances during which clinicians asked for and/or received advice from others (*"This is what I've seen in clinic and please, please help. I really don't know what's going on."*).

With regards to the *Organisation*, clinicians mentioned that the Nurse Coordinator minimised the clerical and bureaucratic work that would usually be performed by the clinician (hence, reducing *coordination demands*). This Nurse Coordinator took over much of this administrative work (e.g., scheduling the patient for tests at hospital, liaising with clinicians), as well as consolidated patient-related information (e.g., test histories) across various databases from various hospitals.

Finally, the *External environment* factors that shaped the work system both before and after the re-design were identical; they were unchanged by the re-design. Notably, health care funding pressures and staffing levels created time pressures on clinicians. The activity-based funding model also reinforced the 'siloed' or speciality driven approach explained above. In fact, the *organisation* and *task* components of the re-designed work system were conceived to counteract the pressures from the *external environment*, which were seen to be inhibiting the successful diagnosis and management of patients with rare diseases.

### *4.3 Outcomes of the re-designed work system*

The primary goal of the program (improving the diagnosis rate of patients) was achieved. After one year of operating, the percentage of families receiving a definitive diagnosis had nearly doubled, achieving a diagnosis rate of 55%. Patients receiving a diagnosis also had a *"clearer pathway"* through the system, and better care coordination. Examples of the full breadth of positive implications of the redesign are presented in Table 1, including examples related to: obtaining diagnoses, next steps, and/or management plans for program patients; finding answers and a community for patients and their families; generating certainty and satisfaction for clinicians; the production of scientific knowledge for the broader patient population; and service improvements as well as global recognition for the health system.

**Table 1.** Outcomes of the re-designed work system.

| **Outcomes** |
|---|
| **Diagnosis**<br>*"One of the successful cases has been two siblings, two patients of mine, where we have managed to work out ... they have a very rare disorder, so that's been really fantastic from my point of view."* – Dermatologist<br><br>*"And their rate of diagnosis is amazing. It's quite a force really."* – Paediatrician<br><br>*"The sense is that in the decade gone they would've been considered highly complex, and may not have had a genetic diagnosis made. Now there's more chance of a diagnosis being made by these sort of meetings."* – Rheumatologist |
| **Focused follow-up plan**<br>*"But in terms of the actual meeting, I think it's when after all the discussions, [the Program Director] goes away with three or four key things that have come out of it, you know we need to get an echo, do a blood test... That might get them a step closer to diagnosis. I think that's when it becomes really beneficial for the patient."* – Paediatrician<br><br>*"...in general terms of understanding a complex patient better and their needs, it's very useful for that."* – Rheumatologist |

*"...you feel like, we've got somewhere to go with this. Yeah, we're gonna try that."* – Nurse

**Quality of life; patient management**

*"Even if a diagnosis is not achieved, some patients have had benefits in other ways like they have other things have been identified, which have been managed well, and the quality of life has improved."* – Geneticist

*"So, it might flag something to be followed up that needs ongoing management."* – Genetic Counsellor

*"See, I think it's definitely a better management plan by the end of it, regardless of whether they think they'll reach…the diagnosis."* – Research Assistant

**Benefits for other patients**

*"And now when we put together finding the mutation with this response to this medication, that's quite big news, so we can publish that, which will potentially help other children."* – Dermatologist

*"It increases our scientific knowledge as a body certainly and hopefully with discoveries made that that will improve management of patients around the world."* – Endocrinologist

**Certainty, satisfaction for clinicians**

*"So, in the end, after every discussion, after every admission, I think all the clinicians go home happy that they have discussed everything, like there's nothing more ….that there's nothing more that they can discuss."* – Geneticist

*"Ultimately, when somebody has suggested some condition and it comes positive, it's very satisfying."* – Endocrinologist

**Publicity, reputation**

*"We've had interest from the minister as to how the programs going and how it can be sustained. So it's, it's garnered that attention from like, really high levels within government. So it's done, it's done very well, and it's obviously had a good success rate otherwise no one would have been that interested in it."* – Policy officer

*"I think it's a roaring success, given what it is."* – Hospital executive

**Impact on the system, on norms in Health**

*"We don't have the time when you have 30 patients in your clinic. You don't have the time to go and look into the details, but now because [the program] is there, if that doesn't have a diagnosis, what other issues this kid has? Probably we could discuss it in [the expert panel meeting]. Everybody has started thinking like that."* – Endocrinologist

*"I think this really makes people realise the importance of getting a diagnosis and just all the different ways that you can do that."* – Paediatrician

*"I think creating, I guess a little bit more of a norm within the health system, that say in the diagnostic phase, you are a specialist and you know that your patient is seeing several other specialists and you haven't hit upon a diagnosis, that it becomes the norm for at least one of those specialists to go, okay, we need a multidisciplinary team meeting - that doesn't seem to exist… think a culture of it just not necessarily been seen as, as the norm."* – Policy officer

*"You can translate that further into any area within this hospital that the key personnel need to be able to sit together at times and talk about the patient together."* – Rheumatologist

While the diagnosis *rate* of the program was indeed quite high, the UDP-WA sees approximately 12 patients per year, at each of its two sites. Thus, some may question whether the relatively small number of patients renders the UDP-WA a successful work system with regards to the diagnosis of rare diseases. It is important to note that the UDP-WA was intentionally established to be low throughput for a number of reasons. Firstly, to investigate whether the program would be successful and sustainable in its implementation in a paediatric hospital, a public health service, and within currently available resources. This success was critical to then expanding this program to involve adult patients and multiple hospitals. Secondly, the program was also established to be low throughput to allow the necessary attention these children and youth deserved on the basis of their need (as evidenced by their lack of diagnosis in the pre-redesign work system), in the knowledge that there were other pathways for those with less diagnostically intractable cases. Thirdly, implementing this program for these 'extreme', low-volume cases created a context in which innovations in clinical practise and system design could be tested and then translated to other clinical pathways (i.e. deliver a multiplier effect). Elements of these innovations have now been translated to other services to further scale the benefit. In recognition of this sustainable, innovative, and equitable service, the UDP was identified as the exemplar program of innovated for sustainability for the WA Health whole of Health System review, the Sustainable Health Review (SHR). The SHR particularly noted its equity of service delivery, including for Indigenous and remote patients.

Further, patients seen by the UDP-WA must meet a number of criteria, including: having a multisystem and severe condition that has presented an intractable diagnostic problem (submitted with previous unsuccessful diagnostic work up, often including genomic sequencing). Thus, while they may arguably be small in number, they are some of the most extensive users of the health systems. Patients are prioritised through an expert panel of physicians, covering a breadth of medical and surgical specialties. A review of health system cost data (unpublished) prior to the commencement of the program, and during its first two years, showed the staggering health system costs of the selected patients, and that even a small reduction in costs due to diagnosis of these patients would generate significant savings – more than sufficient to pay for the program, and to also accelerate other rare diseases diagnosis initiatives. Thus, we are confident in judging the UDP-WA as a success, and an exemplar for future system redesign efforts.

## 5. Factors influencing the Design, Implementation, and Operation of the new work system

In this section, we focus on the process of the re-design (research question 2). Figure 3 (below) depicts the systematic differences in the nature and level of the BEAM factors across each stage. The four levels in the figure – *external environment, organisation, task & work design,* and *person* – were operationalised based on the SEIPS 2.0 component definitions (Holden et al., 2013), whereby

*organisation* in this context reflects the organisation of the work system for rare disease diagnosis (i.e. spanning multiple hospitals and other institutions in the local context).

***Insert Figure 3 about here***

### 5.1 *Design Stage*

In the design stage, decisions were made regarding how to adapt the structure of the U.S. program and implement in the Australian context. Specifically, the structure, content, and support roles of the Expert Panel meetings, as well as how to embed the program within the hospital and wider health care systems, were all considered. The design stage spanned several years – from the initial idea of having an Expert Panel for rare diseases within the state to the first Expert Panel meeting. Across this period, the number and breadth of stakeholders involved in the design snowballed as the Program Director progressively sought the input and/or support of clinicians, hospital executives, the genomics policy unit, the state's Health Department, and international rare diseases networks.

We identified three pairs of barriers and enablers, and three accelerators, in the Design stage (for examples of each, see Table 2). In what follows, we unpack each of these factors further, in turn.

**Table 2**. Barriers, Enablers, and Accelerators to the Design stage.

| Barriers | Enablers |
|---|---|
| ***Local context*** (i.e., the nuances of the local context having ramifications for the design of the new system)<br>e.g., *"Things like the fact that we're a public health system, and that's a private health system…So how would they need to be modified to make sure that they fitted in to the [state] context."* – Policy officer | ***Consultation*** (i.e., seeking input and feedback from local stakeholders, based on their experience in the local context)<br>e.g., *"We got people to run through questions about how might we make [the program] work in Australia, what would be the benefits, what would be the barriers to implementation?"* – Policy officer |
| ***Funding*** (i.e., a climate of scarce financial resources)<br>e.g., *"Because, in the financial setting that we're in, in our health system, where there is no money, and certainly no new money, and budgets are being progressively cut…"* – Program Director | ***Partnerships*** (i.e., co-funding through collaboration with partner organisations)<br>e.g., *"The [policy unit] put quite a bit of funding into the establishment of this program …I think without the policy unit, it would've been hard for the program to get set up."* – Policy officer<br>*"…it was just a timing thing, the relationships and the fact that there was some money [from the clinical organisation] that could be put towards the program which essentially funds [the Nurse Coordinator]."* – Policy officer |

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| ***Norms*** (i.e., attachment to existing routines and ways of working – explicit or implicit)<br>e.g., *"Because familiarity breeds a certain reticence to change. It also breeds a certain sleepiness about the way you approach things. When people started talking about there's these rare diseases program, I go, "I'm already working in rare disease, what more do I need to understand?""*- Biochemist | ***Reputation*** (i.e., positive renown that precedes the program)<br>e.g., *"...it's grown so massively internationally. [the Program Director is] great because he just would send me the emails of you know, the next one that started up in Japan or you know, Hungary or somewhere in South America..."* – Hospital Executive |
| **Accelerators** | |
| ***Model*** (i.e., not designing the system from scratch)<br>e.g., *"They're very engaged with what's happening overseas and they just thought why not bring it here... I think having those relationships really helped and they're just very keen to make things happen."* – Policy officer | |
| ***Genetic technology*** (i.e., pressure for change coming from other sources, such as advances in scientific information and genetic technology – and, often, an interest in adapting to that change)<br>e.g., *"Largely because working within the children's hospital, it's obvious that we use labels and often it's not those for children with chronic disease, chronic undiagnosed rare disease. Those labels are invariably wrong. It's based on something that we thought we understood 20 years ago. That understanding changes with the growth and the technology."* – Biochemist | |
| ***Patient advocacy*** (i.e., pressure for change, and better solutions, coming from patients [i.e., the end-users of the system])<br>e.g., *"People with rare and undiagnosed conditions want answers…There is a Rare Voices Australia consumer lobby group on behalf of people with rare diseases and they're pushing hard for this kind of approach to become the mainstream ones. So I think those are the three things."* – Hospital Manager | |

Specifically, the new work system was designed based on an existing international program for diagnosing and managing rare disease patients. This externally-derived *model* for design thus functioned as an *accelerator*. However, the program designers recognised inherent differences between the health system from abroad and the *local health system context*. To overcome this barrier (i.e., structural health system differences), the 'designers' conducted extensive *consultation* with program stakeholders (cf. *participatory ergonomics,* Hendrick & Kleiner, 2001), to ensure congruency between the *organisation* of the local re-design and the external environment (i.e., the state-wide health system; cf. Clegg, 2000).

Translating the model from the existing international program, which was funded by research grants, to the locally-contextualised program structure, which was embedded in clinical practice, raised the question of *funding* within the resource-scarce public health system. Two *partnerships* – with a government unit and a private organisation that were strategically and culturally aligned with the objectives of better care transitions – were critical in financially supporting the local program.

The program disrupted the way of working in the local health department, constituting a threat to the *norms* and culture associated with these existing work practices. To resolve this, the designers systematically used the *reputation* of the existing international programs (where existing work practices had been successfully re-designed), which added legitimacy to the local program for many clinicians, as well as supporting a sense of being part of a larger international movement.

In terms of additional *accelerator*s in the design stage, rapid changes in *genetic technology* aligned with the system re-design. These technology-driven disruptions from the external environment provided an opportunity for clinicians to adapt to this disruption and proactively learn about novel genetic technologies (within the Expert Panel meetings). Patient *advocacy* groups, which were lobbying for a better approach to treating rare diseases, also helped to accelerate the work system re-design.

### 5.2 *Implementation*

The Implementation stage focused on making the re-structured system functional by securing buy-in and eliciting engagement from the stakeholders that would be involved in the forthcoming Operation of the program – specifically, hospital executives, hospital department managers, and the clinicians who were expected to contribute to core elements of the program (e.g., Expert Panel Meetings). The Barriers and Enablers are outlined, with examples, in Table 3. In what follows, we unpack them in more detail.

**Table 3**. Barriers and Enablers to the Implementation stage.

| Barriers | Enablers |
|---|---|
| ***Resistance*** (i.e., opposition to change from existing work system actors)<br>e.g., *"I might admit, I was reasonably sceptical, because I hadn't really ... The rare disease aspect had been talked about for so many years. I worked day in day out in rare disease and we're doing certain things. Like most people, you think you're doing alright. You've got a reasonable understanding of what you're doing. Because familiarity breeds a certain resistance to change."* – Biochemist | ***Problem recognition*** (i.e., clear ineffectiveness of the existing system of work; consensus regarding the need for change)<br>e.g., *"Clearly traditional approaches…have failed."* – Biochemist<br><br>***Relationships*** (i.e., trust in the system designer translating to trust in the new system; active persuasion)<br>e.g., *"…buy-in was driven by [the Program Director's] conversations and his relationships with people…"* – Policy officer<br><br>***Seeing successes*** (i.e., evidence of the effectiveness of the new work system)<br>e.g., *"Clearly with some early successes, it changed my thinking about that. Hold on. This actually works…"* – Biochemist |

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| ***Resources*** (i.e., a lack of dedicated funding for the new work system)<br><br>e.g., *"…the lack of funding and having to take that time out of their existing budget and their existing workload would have been a discouragement."* – Policy Officer | ***Synergies*** (i.e., drawing on existing resources from within the existing/'old' work system)<br><br>*"It was more how do you create a new thing for essentially no new resources? And so it was: where are the synergies, where do things align."* – Program Director<br><br>***Humility*** (i.e., asking for permission, support from stakeholders)<br><br>e.g., *"He said actually I just need to bring clinicians together. And if you can support me doing that, if I could have access to meeting rooms and we can you know, that might be enough. And so you know, we didn't actually have to put a lot of effort, money or time. It was just actually about support, and saying this is a great idea."* – Hospital Executive<br><br>***Championing*** (i.e., continuous, strategic advocacy for the new system, personified by a key individual)<br><br>*"I mean it needed a champion, which was [the Program Director], but then it needed the buy in of the other clinicians, and without that it wouldn't have worked."* – Policy officer |
| ***I.T.*** (i.e., existing technology that is difficult to use and/or incongruent with the new work system)<br><br>e.g., *"The IT stuff was quite frustrating…Every system we come across we are like this would be really useful we can get some more information. We have to justify why we need the system to the people."* – Clinician | [nil] |

In term of barriers, some clinicians displayed *resistance* to the re-design (affecting their willingness to attend the panel meetings). The following enablers mitigated these barriers: a shared acknowledgement that the existing way of treating rare diseases was ineffective *(problem recognition)*; the Program Director's ability to build *relationships* with the individual clinicians; and the early success of the program (*seeing successes*).

A further barrier affecting the implementation of the program was access to critical *resources* for the program (e.g., physical infrastructure and clinicians' time). Enabling factors that counteracted these

barriers all originated from the Program Director's specific approach to the implementation of the program: rather than asking for additional resources, he emphasised the potential *synergies* with existing resources; he was a *champion* for the program's benefits; and he portrayed a strong sense of *humility* in his communication with hospital executives.

Another significant barrier to Implementation was the multitude of *I.T. systems* that were part of the existing work system, which lacked functionality and interconnectedness. Since these systems were deeply embedded in the existing healthcare work system (i.e. multiple systems were used extensively, and are costly to improve), we did not identify any enablers that were able to mitigate this barrier (thus I.T. integration remains an issue).

### 5.3 *Operation*

For the final stage, we identified factors affecting the *current operation* of the program, (Table 4), as well as factors affecting the future *sustainability* of the program (Table 5).

#### *5.3.1 Current Operation*

**Table 4**. Barriers, Enablers, and Accelerators to the Operation stage.

| **Barrier** | **Enabler** |
|---|---|
| ***Psychological Safety*** (i.e., the need for a shared sense of there being no negative repercussions for speaking up in a team setting – which conflicts with the traditional hierarchy in healthcare)<br>e.g., *"Some of the other meetings you come in with that implicit hierarchy…That hierarchy exists and is reinforced in lots of different ways, both obvious and not so obvious, that when you go into those meetings, you take all of that with you…So like, I would never talk in an MDT meeting, there are registrars who wouldn't either."* – Medical student | ***Leadership*** (i.e., leadership behaviours that, implicitly and explicitly, create a sense of psychological safety)<br>e.g., *"Whereas with [the program] I feel like everyone kind of comes in from lots of different places and that tone is set right from the beginning and it's reinforced throughout the meeting that, you know, the fact that we go around individually, we all get to talk or the way that [the Program Director] will sometimes break down a complex topic for a registrar level. There's lots of different ways that, that atmosphere and that culture is reinforced."* – Medical student |
| ***Specialisation demands*** (i.e., a requirement for knowledge specific to a narrow specialty domain)<br>e.g., *"I've been to quite a number of the meetings over the years, but not felt I've necessarily contributed…I rarely am going to say, "I think it's this condition." That's not going to be my role."* – Hospital Executive | ***Questions*** (i.e., the ability to make a contribution to the diagnostic process just by asking questions)<br>e.g., *"But I've been around long enough to ask a few questions, and feel like it's useful."* – Hospital Executive<br><br>***Teaching*** (i.e., opportunities for the development of the specialty knowledge in question)<br>*"…teaching for the team about tests that because they realised that people weren't particularly* |

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| | *aware of particular testing or how to go about certain testing."* – Medical student |
| ***Time demands*** (i.e., lack of time available for people to engage in the new work system)<br>e.g., *"That was a problem, getting people engaged, people offering feedback, people being prepared to participate in a way that was more than just being on the mailing lists, that was a big problem."* – Genetic Counsellor | ***Nurse Coordinator*** (i.e., a role solely dedicated to coordinating participation in the work system)<br>e.g., *"We know that there is a person responsible, who is collating all this. I just have to read and give my idea, there's somebody else doing that. Because you see everybody is busy in their work… Having a dedicated time for this complex cases and dedicated person responsible who is coordinating everybody."* – Endocrinologist<br><br>***Structure*** (i.e., dedicating time for briefly providing time-poor participants with an opportunity to catch up)<br>e.g., *"And it's kind of like, you know, it doesn't just jump straight into, doesn't expect everyone to have had read everything, because people are really busy, but it just kind of starts with that, you know, a good summary background, you know, which has been synthesized already for the [program] …"* – Research Assistant<br><br>***Effectiveness*** (i.e., the perception that the time that is devoted to the work is effective)<br>e.g., *"Do you think it's wasted time that for one patient you wasted 12 years until you get to the [program]? So I don't think it's a waste of time. And you do need that level of depth if you want to find an answer."* – Genetic Pathologist |
| ***Information processing demands*** (i.e., the volume and complexity of the information that needs to be digested, and the need to process it in counterintuitive ways)<br>e.g., *"So it's like ok, we've got all this information, we all have our cognitive biases - geneticists all think in a certain way, if you're a neurologist you'll think in a certain way, and whatever."* – Program Director | ***Technology*** (i.e., systems that collate and summarise information – and suggest potential solutions)<br>e.g., *"So let's also add in something objective as well, that gives us another opinion and another look, and it doesn't have that cognitive bias."* – Program Director<br><br>***Nurse Coordinator*** (i.e., a dedicated person to summarise and distribute information) |

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|---|---|
| | e.g., *"And they make the meetings relatively accessible in that the information is not voluminous. So you know, if you want to come to a meeting, they send out the information, they've got permission to send that information out to the participants. You look it up, it can take you half an hour, maybe an hour. It's not much to go through, if you want to attend a meeting."* – Hospital Executive |
| **Accelerators** | |
| ***Ongoing success/impact*** (i.e., visible, continued effectiveness of the new work system)<br>e.g., *"I think that's critical to the sustainability. If we were not having any successes, people would start to question why do this."* – Biochemist | |
| **Motivators** | |
| ***Answers / Caring for Patients*** (i.e., individuals with a strong internal drive to find answers for patients)<br>e.g., *"If you can find an answer, it's so important for the families, isn't it, to know the answer? I think that was a great idea, and that's why I jumped at that opportunity to be involved with that."* – Endocrinologist<br>e.g., *"…people care about these patients, they care about finding them an answer and getting them the best care they can possibly get, does seem to be ultimately the driving force behind it."* – Policy officer<br><br>***Challenging*** (i.e., individuals with a strong motivation to be challenged, stimulated)<br>e.g., *"And medicine has changed a lot, you know, back in the day it used to be all about the bed side and working out what was going on with the patient just from examining them and now, we're so quick to do tests and investigations and whatever, that you lose a lot of that, like the art of medicine, things where you actually put the puzzle pieces together and work out what's going on. And so I think everyone finds it really refreshing to actually do that, to have a real puzzle and to have to use all the medical knowledge that you've got to kind of piece it together.* – Paediatrician<br>e.g., *"It's kind of like we're all nerds deep down and you've got these like 15 years of training and all this medical staff that day to day you often don't get to use. So I think everybody loves kind of flexing that muscle a little bit and actually getting a chance to do that…Obviously for the benefit of the patient as well, but it's a nice little test to see 'can you crack the code' kind of thing."* – Paediatrician<br><br>***Development*** (i.e., opportunities for professional development, with broader implications)<br>e.g., *"It's not just about endocrinology, it improves my knowledge overall…it broadens our knowledge and our approach, and it will help our future patients as well if somebody else comes up with similar combinations, then we know. We've done so much reading that, as a clinician as well we improve in diagnosing complex cases."* – Endocrinologist | |

The first barrier within the operation stage was a lack of *psychological safety* in the Expert Panel meetings, originating from the culture of hierarchy in the broader healthcare context, which threatened the active participation of clinicians within the panel meetings. However, the Program Director's *leadership* style during the meetings helped to foster psychological safety for idea-sharing, independent of a panel member's formal seniority or rank.

The second barrier within the operation stage was *specialisation demands;* in the Expert Panel discussions, many clinicians were reluctant to comment on issues outside their area of specialisation (e.g. a Cardiologist commenting on an Endocrinology issue). Many clinicians also voiced insecurities about understanding genetics concepts (including genetic tests). An enabling factor was a recognition that simply asking *questions* allowed clinicians to make 'non-expertise contributions'. Additionally, the Program Director incorporated *teaching* moments into the meetings to provide information about genetic testing to the clinicians.

The third barrier was the high workloads of the clinicians (*time demands*). This barrier was mitigated by: the role of the *Nurse Coordinator* ($P_c$ in Figure 2), who substantially reduced time demands; the meeting *structure*, whereby patient information was summarised at the beginning of the meetings for clinicians who lacked the time to read it beforehand; and a recognition that, while perhaps being a small addition to their workload in the short-term, the meetings saved clinicians time in the long-term (*effectiveness*).

The final barrier was the extent of *information processing demands* (Morgeson & Humphrey, 2006) inherent in rare disease diagnosis, which was still very high (even after the re-design), resulting from the high volume of (often inconclusive) test results that typically accompanies chronically-undiagnosed patients. This barrier was mitigated by the novel *Technology* ($T_x$ in Figure 2) that served to synthesise patient data in a simple way. Thus, while the technology was not the key to the success of the program per se, it was crucial in mitigating information processing barriers.

An *Accelerator* to the operation of the program was its *ongoing success* and *impact*. Being able to see the new work system having positive outcomes for patients and the healthcare system fuelled the ongoing operation of the program.

In terms of *Motivators,* the program served to enhance important motivational job characteristics of the clinicians' work. For example, it played to their desire to *care for patients.* This motivator surfaced through the re-design as it allowed clinicians to find *answers* for rare disease patients (which was often a struggle for the clinicians before the re-design). The *challenges* and stimulation of the Expert Panel meetings also motivated many of the clinicians to participate (e.g., some felt that the 'art of medicine' had been lost in their work, describing how their participation in the program allowed them 'flex their diagnostic muscles'). Finally, professional *development;* the program, and the complex patients reviewed within it, offered an opportunity for the clinicians to improve their diagnostic skills and broaden their clinical knowledge.

### *5.3.2 Sustainability*

In terms of the sustainability of the re-design, we identified (1) current threats and (2) foreseeable future threats to the sustainability of the program (Table 5).

**Table 5.** Threats to the sustainability of the program.

| Threat type | Quote |
|---|---|
| **(1) Existing threats** | **Time demands** (i.e., continued time pressure placed on clinicians – which conflicts with the idea that everyone can make a contribution to the case)<br>e.g., *"If it was expected that the whole panel attended at each meeting, then I would go to each meeting…Because then it's quite equitable* .– Paediatrician<br>e.g., *"Everyone's got limited time, so you've got to be ... Somebody needs to say, "Right, who are the relevant people here? And let's feed it to the relevant people." Otherwise, it's time wasting for everybody else, and nobody's got time to waste."* – Dermatologist<br><br>**I.T. systems** (i.e., unresolved issues with difficult-to-use and/or incongruent technological systems)<br>e.g., *"The IT stuff was quite frustrating…Every system we come across…we have to justify why we need the system to the people."* – Clinician |
| **(2) Potential threats** | **Case selection** (i.e., the meeting-to-meeting variance in the nature of the case at hand, and the consequent [un]importance of each area of specialty expertise in findings a diagnosis for the patient)<br>One paediatrician described how *"…you just learn just by being there… I think it's such a good learning experience and so interesting."* Yet, nonetheless, they recalled a particular meeting that was *"quite high level between about three of the specialists. And I think a few of us who were a bit more general kind of zoned out a little bit. And then just because I knew quite early on there was nothing I was going to be able to say that was going to contribute…"* Yet, as one Geneticist noted, *"getting the right patients to go through the meeting is important, but by and large, that's done well"*. It remains to be seen whether or not the existing case selection approach will continue to satisfy the motivational needs of the clinicians – as well as, of course, patient needs.<br><br>**Funding** (i.e., the need to secure ongoing financial resources for the program)<br>***(-) Need to secure more funding; funding unavailable***<br>e.g., *"I think we could do with a data entry person, an admin person, and then so Lauren can focus her attention more on the coordinating side of things because she's doing all of that, and as a full time job… It needs more resources. That's all I can say."* – Genetic Counsellor<br>e.g., *"Funding of health systems needs to better recognise multi-disciplinary team work. So they're not adequately funded at the moment. So if the patient's not present, you basically get paid for one clinician being there when there could be 20 there. So getting better recognition, I think better data capture of those meetings and funding for them is pretty critical for longer-term sustainability."* – Policy Officer<br>An Executive at the research facility commented that, although *"it seems like a no brainer",* the *"very resource-constrained high-service delivery* |

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|---|---|
| | *orientated health system"* it is *"very hard to see them creating these innovative service models on top of what they're really trying to do".*<br><br>***(+) Minimal concerns from high-level stakeholders***<br><br>e.g., *"If there is a really strong benefit that can be shown, the costs are likely to be very, very small…You could absolutely say you might well save yourself money there by getting to a more clear diagnostic pathway, okay. I'd say that really it's gonna make the argument for itself."* – Policymaker<br><br>One of the Hospital Executives concurred with the perspective of the Policymaker, commenting that *"to maintain it is not really costing people a lot"* and that, consequently, *"it's highly sustainable; it's not at risk of being cut as much as some other areas might be."*<br><br>**Impact** (i.e., concerns that the current scale of the program is insufficient to meet patient demand)<br><br>e.g., *"The [program] is fantastic for the rarer unsolved cases, and it makes a big difference to those individual lives, but it doesn't help the hundreds of referrals that are coming that maybe don't require the complexity of the [program]."* – Geneticist<br><br>e.g., *"The thing that concerns me is that's 12 kids a year that go through this program…[and you want to see it] benefiting as many people as you can, because there's a large number of children who have complex undiagnosed disease that aren't going through that program."* – Paediatrician<br><br>**Succession planning** (i.e., the need to be able to replace key program staff, if necessary)<br><br>e.g., *"I think that at the moment it seems to me that it is very centred around [the Program Director] and [the Nurse Coordinator] and so on coordinating this all…What happens when [the Program Director] can't make it? Can this whole team be not just about an individual but about a whole service and a whole function. I think that needs to be looked at."* – Gastroenterologist |

Despite the enabling factors already mentioned, the *time demands* of regular participation in the Expert Panel meetings continued to be a barrier to the program's operation– affecting the diversity of clinical specialties present in the meetings. This specialty diversity is crucial for diagnostic success – both with respect to pooling the necessary breadth of clinical expertise for each patient, as well as for motivating the clinicians to attend (i.e., to feel like it is 'equitable')

*I.T. systems* constitute another barrier to the program's sustainability since, despite mitigating barriers to the program's operation, the new technology ($T_x$) still requires access to multiple databases (currently accessed manually by the Care Coordinator). This issue could be resolved if the Coordinator was *"anoint[ed] to be part of the healthcare system. So she becomes an employee of the [Hospital Catchment Area]…and that she's got free access to the medical records. Take away that fight. That would be a big win"* (Research Facility Executive). However, how to achieve this solution remains currently unclear.

Given the voluntary nature of clinician participation in the Expert Panel meetings, and the program's dependence on engagement from a diversity of clinicians, sustaining the *motivators* that we identified (Table 4) is crucial. All of the motivators (i.e., finding *answers*, *caring about patients, challenges,* and *development)* depend in some way on the nature of the cases discussed in the meetings. As the

Program Director noted, *"Sometimes the case was too simple…so it didn't give people opportunity to buy in as much or feel like we're contributing".* Consequently, selecting the right case is important for clinician motivation, yet is difficult to achieve: the patient's symptoms must be diverse, such that clinicians are able to learn something outside of their specialty; yet the case must also *not* extend too far beyond the domain of the clinicians' specialties, so that they can still contribute meaningful ideas. Additionally, cases must be complex enough that each clinician finds it challenging; but not too challenging, such that the clinicians' feel like they cannot reach a diagnosis (and thus find *answers* for patients). As one Geneticist noted, *"getting the right patients to go through the meeting is important, but by and large, that's done well"*. However, it remains to be seen whether or not the case selection approach will continue to satisfy the motivational needs of all of the clinicians.

Many clinicians also identified the need to secure ongoing funding for the program to ensure its sustainability. Some, who had been more involved in the administration and coordination of the program, pointed to the need for the better funding of this type of cross-disciplinary work. One executive pointed to the scarcity of funding in the Health system; the Policy Officers also attributed part of this issue to the changing nature of the funding of clinical activity in the Health system. However, the Policymakers (who were more senior stakeholders), were not as worried by the impact of this funding trend on the sustainability of the program, perceiving it as highly sustainable due to its apparent success.

The funding environment also influenced the emergence of another threat to the program's sustainability: some clinicians expressed a concern (or curiosity) about the magnitude of the impact of the program on patients, given that *"it's resource heavy because of all the specialists are working on one case"* (Endocrinologist). Specifically, they were concerned about the structure of the program, whereby only 12 patients are seen per year. One clinician wondered: *"Is there a way to have more patients assessed without requiring the enormous amount of work that it obviously is?"*(Rheumatologist)

Finally, many clinicians identified the Program Director and Nurse Coordinator roles as strong enablers across all stages of the re-design. Some Expert Panel members perceived this as a potential threat to the program's sustainability, whereby they saw the individuals in the roles as being closely tied to the program's success – and thus wondered what would happen if they were to move on from the program. While the current study highlighted the benefits of introducing these *roles* to the new work system, further attention may need to be paid to the qualities that the *persons* in these roles must bring with them in order to maintain the operation of the program.

## 6. Discussion

Our study of the re-design of the system for diagnosing rare disease patients has yielded multiple important theoretical and practical learnings (see Table 6 for a summary).

### *6.1 Removing care transitions*

This special issue was conceived in light of the lack of Ergonomics and Human Factors (EHF) work exploring the socio-technical components of care transitions. In the novel context of rare disease diagnosis (cf. Waterson et al., 2018) – work that requires the integration of complex, high-volume, interdisciplinary knowledge and collaborative problem-solving – this case study demonstrates the benefits of effectively reducing the reliance on care transitions through a more integrated approach. The new UDP work system led to the doubling of the diagnostic rate, as well as improved patient management and quality of life, with implications for other patients and clinicians in the broader

healthcare system. Thus, while is it obviously important to continue studying how to improve the effectiveness of care transitions, we urge EHF (and other) scholars to consider how the application of STS design principles could be used to remove the need for care transitions, where necessary. Further, we urge scholars and practitioners to carefully consider the claims that technology will likely increasingly be used to mediate communication between people who collaborate and share tasks (e.g., Eason, 2014), as this was one of the characteristics of the old system that led to the abysmal rate of diagnosis (i.e., clinicians were not able to discuss the patient, face-to-face, in depth). It was the re-design of the social structures – the integration of specialty knowledge across the pre-existing siloes, the establishment of new cross-disciplinary norms for collaboration, and the power of face-to-face discussions – which were aided by the technology, that were the key to making this complex work system more effective.

### *6.2 Customisation and participation*

Challenging traditional assumptions in EHF theory, our findings empirically confirm Eason's (2014) proposition that, instead of systems being designed from scratch, systems likely evolve in a piecemeal way, whereby multiple existing sub-systems are recombined, and new sub-systems are added, in the design of 'new' STSs. Indeed, the basic structural foundation of the UDP social work system was adopted from the U.S. UDP; and the Patient Archive (i.e., the cornerstone of the technical sub-system) had already been built independently of the UDP, for use by geneticists in Australia. However, also in line two of Eason's (2014) proposed solutions, the 'designers' of the Australian UDP, unknowingly, applied the principles of (1) local design (i.e., "customize [the already-built sub-systems] so that the local sociotechnical system can be optimized" [p. 219]) and (2) user participation in local design (i.e., using human resources as designers in the system, utilising their knowledge of the existing work process; see Figure 3). Without these principles, Eason (2014) warns, system design will simply degenerate to "a re-engineering of the work process to exploit the capabilities of increasingly sophisticated new technology" (p. 219), or 'techomagic' (Wastell, 2011) – which is further reflected in the often technology-centric design changes that STS studies in healthcare focus on (e.g., studying the implementation of new I.T. systems; Carayon, 2006, Table 4), and in well-known case studies of system implementation failure, such as the National Programme for IT in the NHS (Waterson, 2014).

### *6.3 Designing trans-organisational work systems*

Our study supports the claim that it is crucial to proactively managing the process of STS design (Clegg, 2000; Carayon, 2006; Kleiner, 2006). Understanding the design process is just as important as understanding the configuration of a sociotechnical system. We provide an expansion on the nascent literature on the complexity of the STS design process, and the barriers and enablers to STS design in healthcare (e.g., Koma, Bergh, & Costa-Black, 2019). We extend this literature by offering a more nuanced framework of the contributing factors that classifies them as barriers, enablers, motivators, or accelerators.

However, parsing the BEAMs into a multi-level framework not only adds structure and practical utility (i.e., address barriers with enablers at the same level), it also answers calls to better incorporate principles and theories from organisational psychology into design science (Clegg et al., 2017). Many of the BEAMs that we identified are informed by psychological theory – for example: the role of norms in generating change resistance (See Oreg, Vakola, & Armenakis, 2011); the way in which team leadership cultivated a sense of psychological safety in the panel meetings (e.g., Walumbwa & Schaubroeck, 2009); and the way in which feedback about previous patients, perceived impact, and a sense of stimulation and professional development (cf. Hackman & Oldham, 1975) fuelled clinicians'

intrinsic motivation to participate in the meetings (e.g., Gagné, Senecal, & Koestner, 1997; De Cooman et al., 2013). Thus, by drawing from psychological theories, we were able to understand the people in the system as being "so much more than human resource components in a system" (Eason, 2014, p. 215), and unpack the effects of human behaviour in the system and in the design process – both of which are much needed contributions to design theory in the domain of human factors.

Finally, this study makes an important contribution to the ergonomics literature through our investigation of a successfully designed trans-organisational work system (Eason, 2014). Due to the requirement for multidisciplinary expertise, and the population of rare disease patients being spread across the entire state, for the UDP work system to be successful, it had to cross organisational boundaries. In fact, the new work system was designed to essentially remove organisational boundaries, and the care transitions associated with these boundaries, to improve the rate of diagnosis for patients. Many of the barriers that we identified reflect the particularly challenging nature of designing, implementing, and operating trans-organisational STSs in the public healthcare sector. The UDP work system requires active collaboration between time-pressured clinicians and scientists from across multiple healthcare organisations – a challenge that was mitigated by the use of both human and technical resources that served to coordinate these diverse actors. Further, changing existing, well-entrenched ways of working is difficult in any environment; however, in the cash-stripped public healthcare environment, in which values of expertise and hierarchy are historically dominant, changing the system of diagnostic work to a more collaborative approach was particularly challenging. Thus, for every organisation affected by this new work system, stakeholders at all levels needed to be convinced to engage with (and, at times, fund) this new way of working. This highlights that Clegg's (2000) view of the design process itself as a sociotechnical system is particularly pertinent to the design of trans-organisational work systems. The learnings from this case study of this successful trans-organisational work system are rich, and could greatly support future such theoretical and practical trans-organisational endeavours in the healthcare context. In doing so, we contribute to scholars' desires to develop "a design approach that can apply across organisations" (Eason, 2014, p. 215).

**Table 6**. Key learnings from the UDP work system design process.

| **Challenge** | **Learning/Recommendation** |
|---|---|
| Change resistance; challenging well-established norms and ways of working | 1. Seek examples of **similar, reputable programs** or systems of work to use as positive examples *(in the current study, the exact same program was already successful in the U.S.)*<br>2. Seek individuals who are well-networked, who have positive existing relationships with resistive individuals, to act as **champions**<br>3. Allow **user participation** in local design to create ownership and buy-in (cf. Work design theory)<br>4. Regularly **feedback** the success of the new work system to those participating in, or contributing to, it – 'show, don't tell' |
| Idiosyncratic features of the local context presenting a misfit with the designed work system | 5. Facilitate **local design**; "customize [the already-built sub-systems] so that the local sociotechnical system can be optimized" (Eason, 2014, p. 219)<br>6. Allow **user participation** in local design to draw on local knowledge; i.e., using human resources as designers in the system, utilising their knowledge of the existing work process (Eason, 2014) |

| | |
|---|---|
| Scant funding available in the public healthcare sector, and a funding system that does not reward multidisciplinary work | 7. Find **synergies** with existing facilities, and adjacent programs or organisations, rather than creating an entirely independent work system<br>8. Approach stakeholders with **humility**, not over-promising, asking for endorsement and selling the synergistic nature of the system *(i.e., highlight where existing resources can be used)* |
| Time demands placed on clinicians due to resource pressures in the public healthcare sector | 9. **Reduce** time demands of participation where possible (e.g., streamlining participation via collating or summarising patient information in advance)<br>10. Offer **flexibility** with regards to the method and timing of participation *(i.e., work methods autonomy [Morgeson & Humphrey, 2006]; e.g., via e-mail or other electronic submissions of clinical opinions)*<br>11. Give **feedback** to clinicians, demonstrating the output of the time they are investing in the work |
| The conflict between the hierarchical, expertise-driven culture of medicine and the openness and comfort (i.e., psychological safety) required for collaborative, creative work | 12. Create a culture of **psychological safety** through **leadership** behaviours that set norms of equality and non-judgement *(e.g., asking everyone's opinions in turn [regardless of expertise or seniority], breaking down complex specialised topics into simpler terms for non-specialists, friendly and welcoming demeanour, casual humour)* |
| [All of the above] | 13. Capitalise – but do not *rely* – on **natural accelerators and motivators** that are inherent in individuals or the external environment *(e.g., advances in technology, advocacy of end-users, vocational motivation [e.g., desire to care for patients], opportunities for professional development)* |

### *6.4 Limitations and Future Directions*

We recommend several future research directions to extend the findings of our study, with a vision to improve the quality of patient care transitions and the success of the design of collaborative, creative, multidisciplinary systems of healthcare work.

There are three primary avenues through which the robustness and validity of our findings could be tested.

First, a simple way to extend our study would be to follow-up with our sample at a future point in time, to examine the presence or absence of the proposed threats to the program's sustainability (Table 5).

Second, while the qualitative nature of our data provided a richness that was useful in elucidating the specific context of rare disease diagnosis – and an ability to capture the nuances of diverse stakeholders' perspectives – future studies using a quantitative methodology could extend our findings and develop predictive utility by quantifying the magnitude of the effect of each component of the work system on the patient and system-level outcomes. Similarly, quantitative longitudinal studies of the process of re-designing such work systems could uncover the magnitude of the effect of each BEAM on the progression and ultimate success of the STS re-design process, and perhaps

further specify the interdependencies between different BEAMs. Further, our interviews were conducted approximately 1.5 years following the implementation of the new work system. None of our interviewees appeared to have any issues recalling how the re-design process unfolded, which was also a very salient and profound moment in the careers of those who were extensively involve. Further, because the UDP-WA represents a small-scale prototype of sorts, many of our interviewees were still working in the pre-redesign work system (either for their patients with common diseases, or patients with rare diseases unable to be seen within the UDP-WA). Consequently, they were easily able to describe both work systems. Nonetheless, it is still possible that hindsight bias may have had an effect on our findings, which are partly based on our interviewees' retrospections. Future quantitative and/or qualitative studies could investigate such redesign processes as they unfold, in real time, and further explore the magnitude of, and causal relationships between, the factors we have identified.

Thirdly, whilst the richness of our study is undoubtedly a strength, future research could strive to 'cast a broader net' and explore the generalisability of our findings to other nations or other work systems – or, put differently, to explore the contextual variables that lead to different system outcomes. Perhaps this approach could entail studying multiples contexts at once, using a comparative, multiple case study paradigm (for an example, see Maitlis, 2005; as per Lee, 1999, and Yin, 1994).

### *6.5 Conclusion*

In this study we unpack the case of a successful STS redesign that essentially eliminated what were ineffective patient care transitions, consequently mitigating what was previously a massive emotional cost for patients, their families, and clinicians, and a massive financial cost for the public health system. By investigating this STS in the specific context of the work of rare disease diagnosis, we hope to simultaneously: (1) challenge and equip other scholars and practitioners working in similarly acute, complex, multidisciplinary areas of healthcare to redesign existing, inefficient work systems; and (2) inspire scholars and practitioners working in other areas of healthcare, however dissimilar, to consider how crafting collaborative and creative social structures in their work system could serve to improve work outcomes. We urge scholars and practitioners to use, and build upon, the findings and learnings from this study to unlock the potential that may be trapped within poorly-designed systems of work.

**Acknowledgements**

We would like to thank all of the participants in our study for their time, patience, and openness. We would also like to acknowledge Gareth Baynam, Alicia Bauskis, Lauren Dreyer, and Hugh Dawkins for their invaluable support. We would also like to sincerely thank our Research Assistants – in particular, Dale Long, a masters student and Research Assistant, who made a considerable contribution to the Analysis stage of this study, particularly in compiling some of the figures.

Funding: [Removed to retain author anonymity].

No other potential conflict of interest is reported. The views expressed in this paper are those of the authors and do not reflect the views of their employers.

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## Appendix A: Interview Protocol

### Semi-structured Interview Questions – Undiagnosed Diseases Program (UDP) Study

**Q1. Interviewee Background**
a. Can you please give me a brief overview of your career? (Prompts: Have you always worked in this occupation? Where have you worked? Where did you train?)
b. Would you say that having a career in [your field] an important part of who you are?
c. What is your current primary occupation and where are you currently employed?

**Q2. Motivation at work**
a. What do you like most about your work? What motivates you to come to work each day?
b. What frustrates you in your work? What do you like the least in your work?

**Q3. Current work design**
a. What does a typical day look like for you? What sorts of tasks do you do on a day-to-day basis?
b. Do you feel like you have a lot of autonomy in your work? (e.g. decision-making autonomy, work scheduling autonomy)

**Q4. Looking back (Antecedents of the UDP)**
a. How would you describe the diagnosis and management of patients with rare diseases in Western Australia prior to the inception of the UDP? (Prompts: Were patients being diagnosed effectively? How well were patients coordinated across multiple clinicians / specialty areas?)
b. From your point of view, what led to the inception of the UDP?
c. Can you please tell me a bit about how you initially got involved in the UDP?
d. What were your expectations about joining the UDP? What were your initial thoughts?

**Q5. Interviewee's Job Role on the Team**
a. How long have you been part of the UDP?
b. Do you have a specific role in the UDP? How was this role determined?
c. Do you have any other responsibilities outside of the team meeting? What are they?
d. Have you worked with anyone on this team before? In what context? For how long?

**Q6. Task and Team Structure**
a. Describe how work gets done in your team. What are the steps involved?
b. Do you share responsibility and accountability for outcomes (or do some persons have this responsibility and others do not)?
c. How much did the work or work requirements for the UDP team change over time?
d. Are certain members core to the project and others more peripheral? Please describe.
e. How much of the team's work gets done face to face?

**Q7. Information Technology and Applications** (Role of technology, e.g., Genome sequencing technologies and the Patient Archive system)
i. Do you feel that these technological changes (e.g. patient archive, genome sequencing) do/will radically affect your job or you role as a clinician? If yes, how?

**Q8. Social dynamics within meetings / meeting process**
a. Describe the process of how the team comes to a patient diagnosis within the expert panels.
b. How do you know if a meeting was effective?
b. How would you describe the most successful UDP meeting that you ever had? What elements or factors do you believe contributed to its success?
*(Prompts: design of the meeting, behaviour of participants, facilitation, preparation, mutual understanding of the case)*

c. How would you describe the most unsuccessful UDP meeting? What elements contributed to it being unsuccessful?
*(Prompts: design of the meeting, behaviour of participants, facilitation, preparation, mutual understanding of the case)*

**Q9. Performance Management and Human Resources**
a. How are the contribution and performance on the team and individual members evaluated?
b. What kinds of behaviours and performance are rewarded?
c. Is there a team performance reward and recognition system? If so, what is it?
d. How aligned is the team reward and recognition system with the overall goals of the UDP?
e. Are there any other organizational or human resource practices that we haven't discussed so far that have an impact on team effectiveness? Describe positive or negative impact.

**Q10. Impact on Work Design**
a. What impact did the UDP meetings have on the nature of your work (as a clinician)? Please describe some improvements that the UDP had on your workplace.

**Q11. Lessons Learned**
a. What lessons has the team learned since it began operating on how to make an interdisciplinary team effective?

**Q12. Sustainability**
a. What are the three factors that have the highest impact on in ensuring sustainability of the UDP?

## Appendix B: Further information on data collection and analysis

**Data collection:**

1. **Expert Panel Meeting observation:** For each Expert Panel Meeting, we had 1-2 researchers, who were familiar with the program, taking unstructured notes. This served multiple purposes: understanding the general format and flow of the meeting, and the nature of diagnostic work in this context; starting to think about how different individuals from different specialty backgrounds and levels of seniority participated in the meetings; building famiilarity and rapport with the Expert Panel members. Most importantly, these observations informed the design of the interview protocol.
2. **Semi-structured interviews:** During the first year of the study, we first conducted a round of preliminary interviews with key UDP stakeholders (e.g., the Program Director and the Nurse Manager), in order to further develop our understanding of the context and the program, and to refine the interview protocol further. Following this, we invited all members of the Expert Panel, and others involved in the implementation of the UDP, to participate in an in-person, semi-structured interview about their experiences with, and perceptions of, the UDP. These interviews were conducted in a private location – typically the interviewees' office – outside of the Expert Panel Meeting. The interviews were audio-recorded and then transcribed by professional transcription service. These interviews were conducted approximately 1.5 years following the implementation of the UDP, which was approximately 6 months into the duration of this study.

**Data analysis:**

The interview transcripts were first imported into NVivo. Two iterations of template analysis (King, 1998; cf. Waterson, 2014) were conducted:

1. ***Template analysis using SEIPS 2.0 as sociotechnical system template***: We created nodes for each component of the system, nested under two higher-order nodes (pre-UDP STS and post-UDP STS). Subsequently, we sifted through the transcripts to find evidence for what each component of the pre- and post-UDP STS looked like*, coded at the relevant node (based on definitions given in SEIPS 2.0).
2. ***A combination of template analysis using Clegg's (1988) model as template:*** We created nodes for each stage of STS design (i.e., Design, Implementation, and Operation), coding factors that affected the success of each stage under the relevant node. Additionally, to further refine this analysis, we engaged in a semi-inductive process of identifying different *types* of factors; we expected to find Barriers and Enablers, but we also found Accelerators and Motivators (which functioned differently to the Barriers and Enablers). Further, this inductive approach led us to notice that these 'BEAMs' existed at different levels of the system – hence we also nested the BEAMs in a multi-level framework, which describes how they interact to affect the re-design (e.g., Enablers mitigating Barriers).

In both (1) and (2), each of the three authors of this manuscript, in addition to one research assistant, were involved in the analysis. The first author conducted a first round of preliminary analysis with a representative sub-sample of the interview transcripts, to confirm that the templates were suitable for the data. Following this, the transcripts were split evenly between the first author and a research assistant, the latter of whom was taught how to use the templates (including given examples of which data would fit with which node in the templates). Once they had collectively analysed half of the transcripts, the first author and the research assistant met to discuss their progress, and resolve any discrepancies – and then repeated this after analysing the second half of the transcripts. Next, the first author and the research assistant met with the second author to discuss their findings. The second author listened to, and reviewed, these findings with the intention to challenge any assumptions made, offer alternative readings of the data, and to also see if the analysis fit with their own expectations and experiences of observing the Expert Panel Meetings. The third author, who had not attended the Expert Panel Meetings, but who was familiar with the context, also reviewed the analysis, to provide an informed, yet somewhat ‚outsider' perspective on the analysis in general, and on the second

author's suggestions. Following this, the Findings were revised based on the second and third authors' suggestions (e.g., adding the work design assessment to the system analysis; clarifying the exact difference between an Enabler, Accelerator, and Motivator; outlining the multi-level nature of the BEAMs).

***Measuring the job characteristics (from Figure 2)**

While work design research often relies on self-report surveys administered to job incumbents to measure job characteristics (i.e., Morgeson & Humphrey, 2006), the use of external observers to assess the work characteristics is an alternative approach (which is sometimes even considered more objective – see Tomaschek et al., 2018, for a more extensive discussion).

In terms of the assessment, we applied both the concept definitions and the items from the well-validated work design questionnaire (Morgeson & Humphrey, 2006) to our analysis of the interview and observation data. To illustrate, the concept definition for *task significance* is "the degree to which a job influences the lives or work of others, whether inside or outside the organization (Hackman & Oldham, 1975). People in jobs that have a significant effect on the physical or psychological well-being of others are likely to experience greater meaningfulness in the work (Hackman & Oldham, 1980)." (Morgeson & Humphrey, 2006, p. 1323) and items for the self-report scale include "The results of my work are likely to significantly affect the lives of other people" and "The job itself is very significant and important in the broader scheme of things". These items can be answered by job incumbents on a 5-point scale 1 = *strongly disagree* to 5 = *strongly agree* scale.

However, to apply an independent observer rating perspective that allowed us to infer task significance based on what participants reported in the interviews and based on our shadowing observations from meetings, we adapted these items accordingly both with respect to the focal referent (i.e., "my work" was changed into "the clinicians' work" and with respect to the time points ("Before the re-design" and "After the re-design" (e.g. 'Before the re-design, the results of the clinicians' work is likely to significantly affect the lives of other people'). We kept these items in mind when we analysed the interview material.

In terms of assessing the work design **after the re-design**, we paid particular attention to the clinicians' responses to the interview questions that referred to their current work design (i.e., Q3; e.g., What does a typical day look like for you?), as well as what clinicians reported with respect to Q5 (Interviewee's Job Role on the Team), Q6 (Task and Team Structure) and Q10 (Impact on Work Design). For example, one Endocrinologist said: "*If you can find an answer, it's so important for the families, isn't it, to know the answer? I think that was a great idea, and that's why I jumped at that opportunity to be involved with that",* which we interpreted as high task significance.

In terms of assessing the work design **before the re-design**, we paid particular attention to Q4 (Looking back [Antecedents of the UDP]) and also Q10 (Impact on Work Design), as the latter often included a direct comparison with the previous work system. For example, clinicians desribed the way in which they were often unable to find diagnoses for their complex patients, which left them feeling like they were not having a positive impact – we coded this as medium task significance, which exhibited a relative increase after the re-design.

Taken together, we rated the work design for the collective of clinicians (before and after the UDP). We did not use a *statistical aggregation* approach; we decided to adopt a *consensus approach* (i.e., we triangulated these ratings to come to a consensual rating; Quigley, Tekleab, & Tesluk, 2007). Since we used a consensus approach, we decided against using numerical scales when reporting these results in Figure 1. Instead, we use qualitative descriptors, such as "high level" and "low level" in Figure 1. In other words, these ratings reflect "gestalt ratings" based on the observational and interview data collected by work and organisational experts (cf. Tomaschek et al., 2018).