

mEDUrare: Supporting Integrated Care for Rare Diseases by Better Connecting Health and Education Through Policy

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Rare diseases affect an estimated 6-10% of the Australian population, a prevalence similar to that seen in other regions worldwide. These multi-system conditions are often severely debilitating and affect multiple domains of a person's life. A salient necessity for effective care provision thus, is holistic care, achieved by appropriate and continual multi-disciplinary and cross-sectoral collaboration. Synonymous with this priority for collaborative care, is the need for increased partnerships between the health and education sectors. This partnership has the potential to benefit people with rare disease of all educational ages, but in particular, school-aged children and young adults. More than 70% of rare diseases affect children, and this population often experiences difficulties with overall well-being and functioning, including impaired school performance and confounding mental and social comorbidities. Ensuring adequate schooling needs and experiences along with provision of adequate medical care, is crucial in ensuring overall well-being for this population. For this, effective partnerships between the health and education sectors are paramount. This article highlights fundamental elements of health and education priorities, ingrained in current strategic documents, to build a policy foundation that informs and supports increased inter-sectoral partnerships between health and education services. Shared priorities identified in both sectors' guidelines, co-developed with those with lived experience of rare diseases, build a strong policy base for future advocative initiatives to mold better integration between the sectors, a partnership which is vital to improving the overall quality of life, experiences and journeys of people living with rare disease.

INTRODUCTION

Rare diseases are a vast group of chronic, often debilitating conditions that can affect every system in the body. There are over 6000 rare diseases (RD) currently described in literature, affecting approximately 6-10% of

the Australian population [1]. This prevalence is similar to other countries worldwide, with an 8.4% prevalence predicted in the US population, and 6-8% prevalence in Europe [1]. In addition to this high cumulative prevalence, there exists a marked disparity in health and other service requirements, quality of life and life expectancy

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Abbreviations: RD, Rare Diseases; PLWRD, People Living with Rare Disease; WA, Western Australia; ALTNHP, Australia's Long Term National Health Plan; SHR, Sustainable Health Review; NSAPFRD, National Strategic Action Plan for Rare Diseases; WARDF, WA Rare Diseases Strategic Framework 2015-2018; CAHS, Child and Adolescent Health Service; SDFPS, Strategic Directions for Public Schools 2020-2024; CAHSSP, CAHS Strategic Plan 2018-2023; SAHSR, School-Aged Health Service Review; CHN, Community Health Nurse.

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between people living with rare disease (PLWRD) and the general population [2].

Rare conditions have complex underlying pathophysiology, with multiple genetic abnormalities underlying the majority of rare disease presentations. This often results in a significant period of diagnostic delay, termed the “Diagnostic Odyssey.” The average diagnostic odyssey for a patient with rare disease is more than five years, due to limited awareness of rare diseases, their complexity and rarity [3].

Rare diseases are also often severely debilitating, with more than 50% of rare diseases causing some degree of debilitating fatigue, developmental difficulties, or neurological and intellectual disability [4]. The symptoms experienced, combined with frequent hospitalizations and visits to multiple treating physicians and specialists in the process of obtaining a diagnosis, disrupts life and limits independence, often also resulting in psychological and financial strain, and social isolation [4]. A rare disease can have an enormous impact on every aspect of a person’s life, not just their health. As such, individuals with RD require integrated, multidisciplinary care and support. While there are disease-specific considerations for care, many of the needs required are common across the diverse individual conditions. Effective care and advocacy for PLWRD therefore, must focus on effective interdisciplinary and intersectoral collaboration and knowledge-sharing.

Better integration of the health and education sectors is an important opportunity to improve the well-being of PLWRD across all educational ages. There is, however, a particular imperative in implementing targeted measures to address the well-being of school-aged children and youth with RD. Approximately 70% of rare conditions are exclusively childhood onset, and a further 20% are childhood or adult onset [5]. Children with rare diseases often face increased health concerns and lifestyle difficulties, including deficits in school performance, due to chronic fatigue, behavioral issues and learning or development difficulties. Significant psychological and social strain can also occur with frequent school absences, hospitalizations, and impaired school performance [6]. Growing up as a young person with a rare disease also has implications for identity due to the complex experience of prolonged medicalization.

In this way, the quality of life of children living with RD is significantly influenced by their schooling experience. Many describe positive school experiences and participation as an empowering and protective aspect to their illness [6]. Ensuring adequate schooling experiences, and effective intersectoral partnership and communications, is a quintessential necessity in supporting the growth and development of children living with RD.

This paper details the similarities in fundamental

elements of health and education advocacy, espoused in current Australian strategic documents, as a policy foundation to inform and support improved intersectoral partnerships, which are pivotal to improving the holistic care and journeys of people living with rare diseases.

OBJECTIVE AND METHODS

The objective was to review existing national and state health and education policies, to ascertain thematic commonalities as a policy basis for the advancement of health and education sector integration in rare disease care provision. These policies were analyzed for commonalities in advocacy priorities, with a focus to factors that would support improving the lives of PLWRD. A thematically grouped discussion of the common elements and priorities in both sectors’ documents has been discussed.

A further objective is the use of insights gained from policy analysis to inform the subsequent development of solutions that support more integrated care.

Inclusion criteria for policy selection included policy from each level of the health and education system (national, state, and local), which was the most current and foremost source of policy guidance in its respective sector. The policy search was conducted online, using state and national-level government websites, and all strategic documents ascertained are current leading policy sources.

In addition to review of the literature at the level of each jurisdiction, stakeholders representing youth living with rare diseases (AL), clinicians in training (HS), practicing clinicians, including those with rare diseases policy expertise (GB), and educators (CJ) were engaged. Herein, we present the initial results of this work.

POLICY FRAMEWORK AND SELECTION

Within the health domain, initial focus was placed on national health review and reform resources, of which the most current and foremost policy is *Australia’s Long Term National Health Plan* (ALTNHP) published in August 2019 [7]. This national reform plan was created by the Australian Department of Health with the aim of improving the national health system with a focus on preventative health and health promotion measures [7]. Policy analysis then proceeded to state-based reform resources, selecting the *Sustainable Health Review* (SHR), a 2017 review of the current health system in Western Australia (WA), with eight “Enduring Strategies” containing recommendations for future directions of the state’s health system [8].

Subsequently, specific rare disease policy guidelines were ascertained. Nationally, the current guiding policy on rare diseases is the *National Strategic Action Plan for*

Rare Diseases (NSAPFRD), published in February 2020 [9]. This was launched in 2020 following a review of local and international rare diseases policy, informed by Australia's peak RD advocacy group, Rare Voices Australia. It is the first nationally coordinated, comprehensive, and government-endorsed initiative addressing best possible health outcomes for Australians living with RD [7]. Coordinated, connected approaches and cross-sector approaches to support the life journey are key components of the Plan. The state-level equivalent, *WA Rare Diseases Strategic Framework 2015–2018* (WARDSF) [10], published in 2015 before the NSAPFRD, was also included for analysis, as it was the first RD strategy document developed in Australia. The framework's six objectives are directly relevant to this work, and collectively focus on coordinated and integrated care that facilitates access to cross-sector support systems and networks within and across sectors.

Education policy review commenced with the *Strategic Directions for Public Schools 2020-2024* (SDFPS) guideline, released by the WA Department of Education in 2020 [11]. This 5-year plan outlines five aspirations for every student in the WA public school system. Building on these, it outlines six "Improvement Drivers" to guide future strategic directions. The second guideline analyzed was the *Focus 2021* policy document, also published in 2020 by the WA Department of Education [12]. This document deploys the six "Improvement Drivers" from SDFPS into specific actions and desired outcomes [12].

In addition to the above state department resources, guidelines produced by the Child and Adolescent Health Service (CAHS) were also reviewed, as the service plays a key role in child health reform and provision of state-wide health and education services to school-aged children. The documents analyzed included the *CAHS Strategic Plan 2018-2023* (CAHSSP) which highlights five strategic objectives to achieve the CAHS vision of "healthy kids, healthy communities" [13], and the *School-Aged Health Service Review* (SAHSR) document published in 2019 which outlines nine "conclusions" to advance delivery of services that meet specific staff and student needs [14].

These policies were analyzed for commonalities in advocacy priorities, with a focus to factors that would support improving the lives of PLWRD. A thematically grouped discussion of the common elements and priorities in both sectors' documents is provided below.

PERSON-CENTERED CARE AND SELF-ADVOCACY

The term "patient-centered care" was first coined by the Picker/Commonwealth Program (now Picker Institute) in 1998 [15]. The term refers to a care model that

focuses primarily on the patient and their family during planning and execution of care [16]. Although many terms exist for this care model, the underlying meaning and importance is the same; to ensure care and support that focuses first and foremost on the individual person and their needs. For the purpose of this piece, the term "person-centered care" is used, as this term is transferable between both the education and health sectors.

As demonstrated in ALTNHP Pillar One, the need for primary care provision to be more person-centered, rather than disease-centered, is a fundamental one [7]. In the health field, person-centered care allows for personalized treatment and service provision, with increased patient satisfaction and more effective use of resources and funding.

In the education sector, person-centered care allows teachers to bring to attention, each student's individual priorities, with a focus on individual needs, aspirations and hopes [17]. This allows implementation of teaching strategies and support services to best suit each child's individual needs, and thus provide the best possible academic experience [17]. Accordingly, CAHSSP emphasizes the need for students and families to remain the central focus and concern for the service [13].

The guidelines outline multiple ways to implement person-centered RD care, including a tailored and holistic approach. As the WARDSF highlights, patients want their treating physicians to understand their condition and the potential impacts it could have on all areas of their life, to ensure that treatment takes a holistic approach and addresses all patient needs, not just the disease [10]. Similarly, the first SDFPS "Improvement Driver" emphasizes the need to provide every individual student with a pathway to academic achievement and future career success [11]. The SAHSR also emphasizes additional service access for families who require greater support [14]. The guideline notes that Community Health Nurses (CHNs) in schools play an important role in recognizing health priorities in their schools relevant to their individual students, utilizing a holistic approach that focuses on all aspects of students' needs, not just academic [14].

Another aspect of effective person-centered care is the promotion of self-advocacy. That is, the active participation of individuals in their own care. Petri, Beadle-Brown, and Bradshaw (2020) describe five main types of self-advocacy practices; "inform and be informed," "speaking up," "support others," "using media," and participating in "bureaucratic duties" [18]. The first of these, the concept of "inform and be informed," is highlighted in the WARDSF. The guideline encourages the training of health professionals to assist patients living with chronic conditions to be able to self-manage and understand their disease [10]. Similarly, Pillar One of NSAPFRD encourages individuals living with rare disease to actively

participate in their journey by first educating themselves and then further educating others about their disease [9].

The second practice is that of patients “speaking up” as advocates of their condition [18]. Congruent with this, NSAPFRD emphasizes information and education access to promote active participation of PLWRD in care planning and execution [9]. Similarly, CAHSSP encourages children and their families to be active partners in their care plans [13]. It is important to note that students can face stigma and other potential barriers to self-advocacy. A supportive and non-judgmental environment is required if individuals are to advocate for themselves successfully.

The third aspect of self-advocacy highlighted is acting to “support others” [18]. Both sectors’ guidelines encourage partnership with communities to allow individuals to advocate in this way. SDFPS Objective Five aims to partner with communities to enhance the educational engagement of each student [11]. The NSAPFRD also encourages increased support of rare disease organizations to promote further advocacy and representation of PLWRD [9]. The guideline encourages enlisting multiple forms of advocacy, including written submissions, meetings, public hearings, and increased communication within the RD community [9].

The fourth method involves the “use of media” in the advocacy of RD [18]. Abundant online advocacy groups and support services already exist which enlist the use of social media and online communication to highlight the voices of PLWRD. The WARDSF encourages collaboration with establishments such as Rare Voices Australia to support personalized care for PLWRD [10]. The framework also aims to map self-management supports (including online initiatives) that are being, or could be, implemented for use by patients with RD [10].

The last type of self-advocacy practice described, is that of “bureaucratic duties” [18]. As outlined by the WARDSF, this is encouraging the involvement of individuals in the “development of policies and strategies, service planning, design/redesign, delivery and evaluation” [10]. In a similar manner, *Focus 2021* emphasizes using student data to plan for future improvement and teaching strategies, to ensure decisions within schools are made in the best interest of students’ academic opportunities [12]. Focusing on evaluation and implementation, the SHR encourages increased use of patient opinion and feedback to shape and improve services, as well as hold providers accountable in the evaluation process [8]. The SHR also calls for transparent reporting of patient and carer experiences and feedback [8]. This is also emphasized in the *Focus 2021*, which encourages the use of student voice to guide and shape teaching within schools [12].

CARE COORDINATION

Between managing symptoms, health-service needs, diagnostic difficulties and the lifestyle impacts of their disease, the rare disease journey is a complex one. Day-to-day, PLWRD require not only adequate medical care, but also coordinated support services for the many other aspects of life that are affected by their illness. To ensure day-to-day functioning is as smooth and proficient as possible, all care services must be able to coordinate effectively and appropriately. Patients, families, and service providers alike describe the need for less fragmented care in the rare disease journey, as well as support through all key transitions [19].

In addressing this need, the current strategic guidelines highlight three main types of care coordination.

Effective provision of services relies first on care coordination within sectors. As highlighted by Simpson et al. (2021), lack of coordination within sectors can result in inefficient communication and ultimately unnecessary delays in receiving smooth care [19]. Disruption in intra-sectoral coordination is primarily a risk to patients themselves, with health risks and increased symptoms due to care delay, financial losses involved in travel and time off work, and psychosocial strain due to potential disruptions to routine schooling or work activities [19].

As SHR Recommendation Ten highlights, in the health field, improved intra-sectoral coordination consists of improving communications at all levels of care, including between primary care, hospital services, and tertiary care specialists [8]. The guideline also recommends increased use of the “My Health Record” patient information system in populations with complex health needs, to ensure collaborative approaches to care [8]. WARDSF Objective Six also encourages information sharing between professionals and highlights the need to map out WA Health services which are most utilized, and how these interface [10].

In the education sector, intra-sectoral coordination comes in the form of whole-school and inter-school approaches to targets, strategy, and policy. Reiterating this need, *Focus 2021* encourages increased whole and cross-school collaboration to strengthen teaching practices [12].

The second type of care-coordination highlighted is cross-sectoral collaboration. Both health and education policy acknowledges the need for cross-sectoral coordination, including with parents and family. As NSAPFRD states, there is a need for “cross-jurisdictional, cross-sectional working part[ies]” [9]. The guideline encourages collaboration between key partners including education providers, professional institutes, and community support services [9]. The SHR also mirrors this focus on increased partnerships between different sectors [8]. The need for cross-sectoral partnership is also evident in edu-

cation policy, with the CAHSSP emphasizing the need to work with key support partners, including primary health, to increase ease of access to support [13]. *Focus 2021* Objective Five also aims to enhance collaboration with support partners to enhance support services for children with disability [12].

The third type of care coordination emphasized is coordination in times of service transition. Most commonly, this is transition due to age, be that from child to adult health services in the health sector, or from primary to secondary schooling or beyond. The 2016 Molster et al. survey describing healthcare experiences of Australian adults with RD, found that 52.8% of participants experienced significant issues during transition from pediatric to adult health services due to a lack of targeted support measures [20]. This is consistent with the Nieboer et al. study (2014) which demonstrated that application of specific measures in times of transitional care, such as interventional support programs, improved coordination, and patient experiences thereof [21].

The guidelines for both sectors recognize the importance of implementing transition-specific interventions. The NSAPFRD encourages provision of specific supports during life-stage transitions, including child to adult services transition, end-of-life care and relocating [9]. The CAHSSP also encourages a focus on creating methods to ensure coordinated journeys for individuals through the health and education system [13]. The SAHSR suggests development of a formal handover system between school and health services to ensure that children with higher needs have adequate continuation of care through major transitions [14]. Additionally, *Focus 2021* also encourages a process of early planning to support students throughout their whole education and beyond [12]. Partnership and systems between health and education exist to enable procedural enhancements for young people during transitions.

INFORMATION-SHARING WITH PATIENTS AND FAMILIES

In both the health and education sectors, the importance of providing timely and appropriate information and resources to individuals and their families cannot be understated.

In the health field, care can only be considered complete when all aspects of the patient's health journey and care plan have been discussed with, and understood by, the patient and (if and when appropriate) their families. The Molster et al. survey (2016) reports that at the time of diagnosis, almost one in five participants (19%) did not receive any patient information, and in those who reported receiving information, only half indicated that they understood all of the information that was given

[20]. Further to this, many participants felt they had not received sufficient information regarding available support services and resources, including written resources and information pamphlets [20]. In assessing how to address this, WARDSE notes that the information-sharing process relies heavily on effective links and partnerships between PLWRD and the professionals involved in their care [10]. The framework emphasizes that effective connections allow discussion of, and hence referral to adequate information, support services and networks [10]. Underpinning quality communication and support is documented consent by the parent and student to enable timely exchange of information between the student, family, treating team and enrolled school. The NSAPFRD Pillar One also emphasizes having discussions with patients regarding where to find information and support services when needed [9]. The guideline also encourages increased involvement of relevant RD organizations to raise awareness of the service pathways available [9]. Similarly, the SHR encourages the introduction of community and online pathways to navigate individuals to support services [8].

Similarly, in the education setting, it is critical that all students and their carers receive continual feedback regarding student progress and learning needs, as well as an opportunity to provide feedback and evaluation to school staff. This is especially important for students with complex care and learning needs such as those living with rare disease. This is emphasized in the SDFPS guideline, which encourages partnership with families to allow information sharing and highlight any priorities that need to be addressed [11]. The CAHSSP also emphasizes making every family's journey easier by connecting them to relevant, local and easily accessible support services [13]. Children with rare diseases, and their families need to be equipped with ways to discuss their rare disease and the likely impact of the condition on their learning with school staff. Schools need to have access to adequate training to support capacity building and student health care planning.

Of additional relevance, is support and education for the families and carers of PLWRD. Recommendation Five of the SHR highlights the need for education and support for carers, through early recognition and training to ensure overall well-being and strengthen resilience in care [8]. Similar to this, the SAHSR highlights the importance of ensuring all parents have access to high quality and locally available parenting programs and support services [14].

TECHNOLOGY AS AN ENABLER OF RD CARE

What future innovations and methods will be used to

improve access to, and integration of, effective RD care?

While it is evident that human factors are critically important to holistic and accurate care provision, increased use of technology could be a critical enabler in the provision of best care. The promise of technology is highlighted across the policy documents of both sectors.

A key focus of more coordinated and integrated care reflected in the reviewed documents, is the use of universalized and easily accessible personal health records. As described in the Macleod et al. survey (2015), many individuals with chronic conditions, such as RD, are having to maintain their own records, including resources regarding their condition, and provide these upon care initiation with different members of their treating team [22]. As the article suggests, the use of patient-centered information repositories, such as universalized personal health records accessible cross-sectorally, including by student services staff and school nurses, will permit more complete information sharing, and thus ease care continuity between sectors [22]. Increased accessibility, for every member of the care team, to comprehensive patient records will grant more efficient, more accurate and more personalized care.

This necessity is also highlighted among current guidelines. As the WARDSF describes, patients want their information shared effectively amongst all professionals involved in their care [10]. The guideline encourages development of methods to allow sharing of patient information across health and other professions [10]. The NSAPFRD expands on this, aiming to leverage existing health care records, such as My Health Record, to improve its integration across health and other services [9]. In support of this, the SHR Recommendation 22 prioritizes rollout of a functional and universalized electronic health record across the health system [8]. The CAHSSP also supports this need for streamlined access to information and services provided to all partners in care [13]. The SAHSR highlights that online health information access and electronic health records must also be easily accessible by CHNs within schools [14]. Complete and efficient care provision within schools relies on CHNs having access to all appropriate health and service utilization records prior to the child starting school, to ensure services can be requested and provided promptly upon student enrolment [14].

Macleod et al. (2015) also encourage the use of technology as an avenue for information sharing in chronic disease [22]. Similarly, WARDSF prioritizes the use of electronic information-sharing techniques to educate patients, families, and treating professionals about relevant online information and services [10]. Parallel to this, NSAPFRD Priority 1.2 is to implement an easily accessible, multi-purpose online repository containing information about RD support services and pathways [9].

Hirschhorn et al. (2020) also propose the idea of augmented artificial intelligence as a future forefront of RD care and treatment [23]. Considering this, and many other articles like it, which describe complex technologies as a way forward for integrated RD care [24], a digitally capable workforce is a necessity. A recognition of the importance of a technologically literate workforce is apparent in the guidelines for both sectors. The SHR recognizes the importance of digital health technology in empowering people through ease of information access [8]. Accordingly, Recommendation 11 of the guideline is to focus on working with patients, families, and care providers to increase the uptake of telehealth and virtual care services in all disciplines [8]. The NSAPFRD also aims to increase the utilization of digital health technology [9].

Similarly, CAHSSP highlights the need for responsible use of resources and suggests increased uptake of digital technology in care will ensure more responsible and personalized use of services [13]. Increased use of technology in schools also aligns with the SDFPS Objective Two of strengthening support for teaching in our classrooms [11]. The SAHSR also highlights the need for increased digital technology integration into student support services, such as increased use by CHNs in schools, not just in classrooms [14]. *Focus 2021* prioritizes increasing the capacity and confidence of school staff in information and communication technology services, with an aim to increase professional learning for all staff to acquire relevant skills [12].

A digitally capable workforce is a current need and may help to realize increasing opportunities for improved service provision for children with increased care needs. As highlighted, children living with RD have frequent school absenteeism due to hospital admissions and other health requirements. As *Focus 2021* highlights, technologically capable teachers will be able to continue provision of education programs and teaching during any disruptions to face-face learning [12].

EARLY DETECTION AND DIAGNOSIS

Early detection of potential issues allows for faster initiation of the diagnostic journey, and thus more prompt diagnosis. Obtaining a diagnosis is not only beneficial for patients in terms of earlier initiation of best medical care, but also for the patient's own peace of mind regarding an explanation of their symptoms and experiences [25]. The NSAPFRD emphasizes the need to train and encourage all health professionals to consider, investigate for, and refer, all potential RD diagnoses [9]. The guideline also aims to ensure all screening and testing programs evolve in time with new RD research, allowing for sustainable pathways of referral and diagnosis [9]. Building on this, the SHR aims to decrease diagnostic odyssey by creating

a system-wide approach identifying frequent users of emergency and inpatient health services, to enable referral and reduce repeat presentations [8].

Early detection of potential issues that have become, or may become, barriers to a child's education, is also a responsibility of education staff. The guidelines demonstrate the shared priority of detecting issues early, to negate any possible consequences of late detection. SDFPS Outcome Two is to ensure students are on track in their early years, to ensure continued success through their schooling [11]. Detection of early difficulties in the classroom, as well as screening by student support services, is fundamental to this aim. The WARDF outlines that referral from CHNs to the state-wide child development service plays a crucial part in initiation of many children's diagnostic journeys [10]. Recognizing this key role of CHNs in the early detection of potential difficulties, SAHSR aims to provide CHNs with services for increased screening and thus earlier detection of students who require further assessment for developmental or other health issues [14].

Importantly, the WARDF emphasizes that CHN assessments of development and function will also be made upon referral from teachers [10]. Hence, enhancing RD awareness and training for teachers is necessary to support early detection and timely referral. Additionally, if teachers are trained in recognizing early symptoms and developmental problems, timely changes in teaching strategies within the classroom can be made to best suit the individual student, and negate any difficulties faced. This includes provision of effective teaching and learning, despite frequent absences and symptoms that may impair school performance. As discussed previously, continuation of schoolwork despite absences and other difficulties improves student experience and quality of life [6].

WORKFORCE CAPACITY-BUILDING

Rare diseases workforce capacity building across health professionals, teachers, student services staff and all other professionals involved in caring for PLWRD and their families, can support improved health and education journeys.

The WARDF highlights that patients want health and other professionals involved in their care, to have adequate knowledge of their condition and have a good understanding of the services and specialists available for referral [10]. The framework acknowledges the need to provide health professionals with ongoing access to RD training and education, and suggests enhanced collaboration with RD organizations to expand on current education and awareness programs, including public awareness sessions and advocacy in professional conferences and newsletters [10].

The NSAPFRD acknowledges patient difficulties in finding practitioners who are educated in their condition and recognizes the urgent need for focused education of the health and support workforce to increase its ability to meet specific care requirements, including knowledge of referral pathways [9]. The guideline identifies the need to create a national RD workforce strategy to conduct RD awareness and education activities and address the current gaps in the RD workforce in a targeted manner [9]. Parallel to this, SHR acknowledges that this training must be provided to educational institutions and professional colleges, as well as to hospital-based staff and junior doctors [8]. It encourages education of the workforce that includes a sound knowledge of mental health and social care system navigation, as well as training for a digitally-literate workforce [8].

Workforce capacity building is also prioritized in CAHSSP, with the need for continual professional development and life-long learning for school staff a priority of the service [13]. Concordantly, *Focus 2021* aims to ensure that all professional learning is up-to-date and contains training regarding pathway planning resources [12]. An actionable step towards this priority of workforce development, outlined by the SAHSR, is the creation of a state-wide, nurse-led training program for school staff, with targeted professional learning on how to adequately care for students with complex health needs [14]. As suggested in the SAHSR, this will be in collaboration with all key support partners, including disability and tertiary health providers [14]. The guideline also emphasizes training of CHNs within schools to ensure they have appropriate skills to work with vulnerable students and families [14]. This is further supported in SDFPS, which also emphasizes the need for focused education of allied professionals and student support staff to ensure holistic student well-being [11].

MENTAL AND SOCIAL COMORBIDITY IN RARE DISEASE

The prevalence of mental health disorders is disproportionately higher in people living with rare disease, compared to the general population, be that due to mental health being a known manifestation of the individual condition or as a comorbidity [26]. The mental health burden that often exists with RD may also complicate the diagnostic journey, as the symptoms experienced with RD are sometimes initially incorrectly diagnosed as a psychosomatic disorder [26]. PLWRD often also face increased social strains, and children living with RD experience these issues at a higher rate, manifesting as issues such as bullying, fear of being judged, and social isolation [27]. Integrated RD care and support must therefore address all domains of life, including physical, psychological, and

social.

NSAPFRD Priority 2.1 focuses on policy that meets the full range of individual requirements, including disability and education [9]. This includes making sure patients and their families receive community, clinical and digital mental health support services [9]. The guideline also encourages training for RD organizations to raise awareness of available mental health support services [9]. Similarly, SHR Recommendation 26 focuses on enhancing the teaching curriculum of health and social services to include a sound understanding of how both health and social care systems operate, and the referral pathways between them [8]. The guideline also recommends increased contractual engagements with community mental health services [8].

Similarly, SDFPS Aspiration Three is for every student to develop the personal and social attributes required for overall, holistic well-being [11]. In alignment with this, *Focus 2021* aims to provide further professional development training to all school staff to be able to better manage student mental health [12]. The guideline also wishes to promote alcohol and drug education programs to support high-risk students and families [13]. Importantly, SAHSR encourages formal training for CHN's in schools, which focuses on adolescent mental and sexual health issues [14].

A FOCUS ON PRIORITY POPULATIONS

People living with RD have been identified by the United Nations Office of the High Commissioner for Human Rights (OHCHR), as a priority under a human rights banner for the delivery of Universal Health Care [28].

Within this recognition of PLWRD as priority for advocacy and care provision, it is to be noted that there are further priority populations within the RD community. As described by the NSAPFRD, these include individuals with undiagnosed RD, those with an increased risk of developing RD, Aboriginal and Torres Strait Islander populations, people living in regional, remote, or rural areas, those from Culturally and Linguistically Diverse (CALD) backgrounds and those living in socio-economic disadvantage [9]. The guideline highlights a need for targeted intervention to improve the health and well-being of these priority populations, suggesting targeted approaches to maximize the reach and appropriateness for educational materials and supports to Aboriginal and Torres Strait Islander people, those from CALD backgrounds and other priority populations [9].

In alignment with this, SHR Recommendation Three aims to focus on the expansion of compulsory, system-wide cultural learning to gain an understanding of Aboriginal health, with the aim of building a culturally competent health system [8]. The guideline's Chapter

One: "Courage to Take Action," describes in detail the inequalities faced by priority populations including Aboriginal and Torres Strait Islander people, people living in regional regions and those from CALD backgrounds [8].

This focus of priority populations is also demonstrated in the education sector. SDFPS Success Outcome One describes the priority of "enabl[ing] Aboriginal students to succeed as Aboriginal people" [11]. Further to this, *Focus 2021* prioritizes utilization of evidence-based approaches to improve Aboriginal student outcomes, as well as working with local communities to determine aspirations and directions for individual students [12].

Building on these recommendations, the Department of Education has also created the "Aboriginal Cultural Standards Framework," which comprises five cultural standards and a continuum, to guide staff about best care for Aboriginal students, their families and local community, as well as provide a structure to reflect on individual and whole-school strategies and progress [29]. Similarly, WA Health has created the "WA Aboriginal Health and Wellbeing Framework 2015-2030," an actionable framework with targeted measures to advance the education outcomes for every Aboriginal student [30].

CONCLUSION AND NEXT STEPS

Rare disease care requirements are fundamentally multi-disciplinary and multi-sectoral. An integrated approach to support PLWRD to live their best lives possible, needs to be informed from a whole-of-life perspective. Health and education are fundamental determinants of well-being, perhaps no more so than in childhood and youth compared to other life stages. Given the prevalence of rare diseases is greatest in children and youth, it is particularly important in this population to best integrate between the health and education sectors.

Following the policy review and stakeholder discussions described herein, a number of solutions are being developed. These include first, a Rare Disease Teacher Resource, a module to be distributed to educators that highlights red flags indicating that a child may have a rare disease, identifies commonalities in the academic needs of children and youth living with rare diseases, and provides linkages to further disease specific information, including specific education resources where they exist, and second, a video diary series of PLWRD highlighting aspects of their educational experiences.

These approaches will also be coordinated with the RArEST (Rare diseases Awareness, Education and Support) initiative funded by the Australian Commonwealth Health Department. The RArEST initiative aims to deliver a national approach and resource for RD awareness, education and support for a range of stakeholders. The team delivering RArEST includes patient advocacy

leaders, PLWRD, clinicians, researchers, educators, and policy experts. The initiative is informed by, and tightly aligned to, the NSAPFRD nationally, and the WARDSF on a state level. Collectively, these measures are the continuation of a journey, and additional approaches will likely also be required.

The future of rare disease advocacy and care is holistic, complete care that focuses on individual needs and requirements in all domains of life. Especially important is this partnership between healthcare and educational services.

As this review highlights, partnership between these sectors already exists, and is fundamentally intertwined, at a policy level. Both sectors have similar priorities detailed in current strategic policy, with similar recommendations of action to ensure best care. Future advocative initiatives can be confident that enhanced inter-sectoral collaboration between these services is in line with the core priorities of both sectors.

REFERENCES

1. Knight AW, Senior TP. The common problem of rare disease in general practice. *Med J Aust.* 2006 Jul;185(2):82–3.
2. Walker CE, Mahede T, Davis G, Miller LJ, Girschik J, Brameld K, et al. The collective impact of rare diseases in Western Australia: an estimate using a population-based cohort. *Genet Med.* 2017 May;19(5):546–52.
3. Dudding-Byth T. A powerful team: the family physician advocating for patients with a rare disease. *Aust Fam Physician.* 2015 Sep;44(9):634–8.
4. Kirby T. Australia makes up for lost time on rare diseases. *Lancet.* 2012 May;379(9827):1689–90.
5. Nguengang Wakap S, Lambert DM, Olry A, Rodwell C, Gueydan C, Lanneau V, et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet.* 2020 Feb;28(2):165–73.
6. Paz-Lourido B, Negre F, de la Iglesia B, Verger S. Influence of schooling on the health-related quality of life of children with rare diseases. *Health Qual Life Outcomes.* 2020 Apr;18(1):109.
7. Australia's Long Term National Health Plan. Canberra (NSW): Department of Health (Australia); 2019 Aug 14.
8. Sustainable Health Review: Final Report to the Western Australian Government. Perth (WA): Department of Health (Western Australia); 2017 Jun.
9. National Strategic Action Plan for Rare Diseases. Canberra (NSW): Department of Health (Australia); 2020 Feb 26.
10. WA Rare Diseases Strategic Framework 2015–2018. Perth (WA): Office of Population Health Genomics, Public Health Division, Department of Health (Western Australia); 2015 May 1.
11. Strategic directions for public schools 2020-2024. East Perth (WA). Western Australia: Department of Education; 2020. 1 pp.
12. Focus 2021. East Perth (WA). Western Australia: Department of Education; 2020.
13. Child and Adolescent Health Service Strategic Plan 2018-2023. Nedlands (WA): Government of Western Australia; 2020.
14. Child and Adolescent Health Service School-aged Health Service Review: key findings. Nedlands (WA): Government of Western Australia; 2019.
15. Gerteis M, Edgman-Levitan S, Daley J, Delbanco TL, editors. *Through the Patient's Eyes: Understanding and Promoting Patient-Centered Care.* 1st ed. San Francisco: Jossey-Bass; 1993.
16. Barry MJ, Edgman-Levitan S. Shared decision making—pinnacle of patient-centered care. *N Engl J Med.* 2012 Mar;366(9):780–1.
17. Swan S. The importance of a person centered approach in special education. *DKG Bulletin.* 2017;83(4):34–6.
18. Petri G, Beadle-Brown J, Bradshaw J. Redefining self-advocacy: a practice theory-based approach. *J Policy Pract Intellect Disabil.* 2020 May;17(3):207–18.
19. Simpson A, Bloom L, Fulop NJ, Hudson E, Leeson-Beevers K, Morris S, et al. How are patients with rare diseases and their carers in the UK impacted by the way care is coordinated? An exploratory qualitative interview study. *Orphanet J Rare Dis.* 2021 Feb;16(1):76.
20. Molster C, Urwin D, Di Pietro L, Fookes M, Petrie D, van der Laan S, et al. Survey of healthcare experiences of Australian adults living with rare diseases. *Orphanet J Rare Dis.* 2016 Mar;11(30):30.
21. Nieboer AP, Cramm JM, Sonneveld HM, Roebroek ME, van Staa A, Strating MM. Reducing bottlenecks: professionals' and adolescents' experiences with transitional care delivery. *BMC Health Serv Res.* 2014 Jan;14(1):47.
22. Macleod H, Oakes K, Geisler D, Connelly K, Siek K. Rare world: towards technology for rare diseases. *Proceedings of the 33rd Annual ACM Conference on Human Factors in Computing Systems*; 2015 Apr 18–23; Seoul, Republic of Korea. New York: ACM; 2015 Apr. <https://doi.org/10.1145/2702123.2702494>.
23. Hirsch MC, Ronicke S, Krusche M, Wagner AD. Rare diseases 2030: how augmented AI will support diagnosis and treatment of rare diseases in the future. *Ann Rheum Dis.* 2020 Jun;79(6):740–3.
24. Groft SC, Paz MP. Preparing for the future of rare diseases. In: Paz MP, Taruscio D, Groft SC, editors. *Rare diseases epidemiology: update and overview.* New York: Springer-Link; 2017. pp. 641–8.
25. Baynam G, Pachter N, McKenzie F, Townshend S, Slee J, Kiraly-Borri C, et al. The rare and undiagnosed diseases diagnostic service - application of massively parallel sequencing in a state-wide clinical service. *Orphanet J Rare Dis.* 2016 Jun;11(1):77.
26. Nunn R. "It's not all in my head!" - The complex relationship between rare diseases and mental health problems. *Orphanet J Rare Dis.* 2017 Feb;12(1):29.
27. Adama A, Arabiat D, Foster MJ, Afrifa-Yamoah E, Runions K, Vithiatharan R, et al. The psychosocial impact of rare diseases among children and adolescents attending mainstream schools in Western Australia. *Int J Incl Educ.* [Ahead of Print]. 2021 Feb 22 [cited 2021 May 21].
28. United Nations human rights body stresses the need to address rare diseases within universal health coverage [Internet]. *Rare Diseases International*; c2021 [cited 2021

- May 21].
29. Aboriginal Cultural Standards Framework. Perth (WA). WA: Department of Education; 2015.
 30. WA Aboriginal Health and Wellbeing Framework 2015-2030. Perth (WA). WA: Department of Health; 2015.