Submission No 34

IMPROVING ACCESS TO EARLY CHILDHOOD HEALTH AND DEVELOPMENT CHECKS

Organisation: Genetic Alliance Australia

Date Received: 7 March 2024

info@geneticalliance.org.au
Level 6, 384 Victoria Street
Darlinghurst NSW 2010

7 March 2024

Mr Clayton Barr, MP
Committee Chair
NSW Legislative Assembly Committee on Community Services

Dear Mr Barr,

Response to NSW Legislative Assembly Committee on Community Services Inquiry on Improving access to early childhood health and development checks.

Genetic Alliance Australia (GAA) welcomes the opportunity to provide a submission outlining the importance of this issue to our membership. GAA is an umbrella group for rare genetic conditions, many of which are so rare they do not have their own support group. We seek to connect families and individuals affected by the same, or similar condition, and/or provide information about relevant support groups both nationally and internationally. GAA is a member of the Clinical Genetics Executive Committee, NSW Newborn Screening Committee, NSW Register of Congenital Conditions, Consumer Health Forum, EURORDIS, Health Consumers NSW and the International Alliance of Patients Organisations (IAPO).

It is estimated that 7,980 NSW children are born each year with a rare genetic condition.[1,2] The clinical manifestations of these conditions are diverse, frequently involving the nervous system and giving rise to symptoms such as intellectual disability, neuropsychiatric disorders, epilepsy, and motor dysfunction.[3]

Many infants and children with rare conditions presenting to healthcare professionals with developmentally associated symptoms are going undiagnosed,[4] limiting opportunities for crucial early intervention, targeted treatment and support. This can dramatically affect a child's prognosis and can have a profound and lifelong downstream impact on their access to education, their potential for learning and growth, and ultimately their quality of life.

The bi-partisan approved National Strategic Action Plan for Rare Disease [NSAPRD][5] calls for the voice of people living with a rare disease, their families and carers to be embedded throughout structures and systems that impact rare diseases. This inquiry provides an important opportunity to highlight the lived experience of our membership and the importance of early childhood development checks within the construct of the often complex and challenging rare diagnostic odyssey. As such, we recognise that some of the issues raised are supplementary to the terms of reference and outside the direct jurisdiction of the Committee of Community Services, however understanding the vulnerability of children with rare genetic conditions, and in particular the intersection of families from minority communities and those living in remote and rural locations is critical for informed future decision making.

With this context, we strongly encourage the committee to consider how potential interventions raised throughout the process of this inquiry could be expanded to address the significant disparities experienced by the rare community.

We applaud the committee for undertaking this inquiry and warmly invite any future opportunity to discuss the concerns and recommendations outlined in our submission.

Yours sincerely,

[redacted]

Jan Mumford
Executive Director
Genetic Alliance Australia (GAA)

Funded by NSW HEALTH

Table of Contents

Executive Summary:

In New South Wales, nearly 8000[1,2] children are born with rare genetic conditions each year, many of which manifest complex symptoms during early childhood that can impact upon developmental milestones.[3] Many children remain undiagnosed for years, undermining the opportunity for age-specific multidisciplinary intervention that can dramatically alter a child's outlook, affecting their clinical prognosis,[4] capacity for learning and advancement and ultimately their quality of life.[7,8]

The existing system for early developmental surveillance (which is valuable in identifying symptoms of rare conditions) is hindered by disparities in access to and expertise of healthcare professionals and long waitlists for geneticists, genetic counsellors, developmental experts, and other multidisciplinary providers. This is compounded by challenges in navigating across complex streams of multidisciplinary care and disparate health, social and educational support and services. This has created a system, particularly in remote and rural communities, that is subject to vast inequities in clinical and developmental outcomes.

Evidence has clearly demonstrated that these challenges, combined with the chronic and debilitating nature of many rare conditions, can lead to heightened stress, anxiety, isolation, loneliness and grief,[13] yet less than half of families currently receive adequate psychosocial support.[4] The interdependent health and psychosocial needs of children and parents are critical and well-supported parents can effectively navigate their child's rare condition. Conversely, a lack of support for children with rare conditions and their parents can impede their emotional, social, and cognitive growth, preventing participation in society to the fullest of their unique potential.

The NSW Government's ongoing investment in children's physical, social, and emotional health during their initial 2000 days is encouraging, especially for those with rare conditions and associated developmental comorbidities. Consultations with GAA member organisations have shaped this response and the following recommendations across three key areas; **1) education and capacity building; 2) addressing access barriers and inequity and 3) health system navigation and care coordination.**

Key recommendations:

***Education and capacity building**: improving the capability and capacity of all providers participating in early developmental surveillance in identifying, appropriately referring and identifying support needs for children with rare conditions through education, skills building and incentives.*

- **Rare condition and genetic/genomic literacy:** Support multi-stakeholder collaboration to provide professional development (CPD) accredited short courses and micro-credentials to rapidly upskill NSW Health staff on genetics and genomics in areas relevant to their fields. Increase early and interdisciplinary training exposure to rare conditions and genetic counselling and support partnerships between medical institutions and rare disease organisations to offer clinical rotations and internships, making these specialties more accessible and appealing to emerging healthcare professionals.

- **Introduction of incentives:** Encourage more comprehensive early childhood checks through the introduction of adequate incentives, including advocating to Federal bodies for the introduction of Medicare Benefits Schedule (MBS) item codes where appropriate.

- **Early childhood and school educator capacity:** Enhance the training and support for early childhood educators and teachers to effectively manage children with rare conditions and their associated developmental and psychosocial challenges. This involves incorporating education on rare conditions, genetics, and practical disability management into tertiary education curricula to cultivate a deeper understanding and practical skills among educators.

- **Parental capacity**: Increase parent's access to more detailed and interactive developmental assessment tools, that better continually monitor progress over time and are easily shared across different providers. Improve parent education on the importance of developmental surveillance and the availability of resources in pre-natal settings.

- **Community capacity**: Support the development of Aboriginal and Torres Strait Islander and CALD community-led initiatives designed to drive awareness and understanding of rare genetic conditions and genetic screening in a manner that respects and integrates their distinctive cultural perspectives on genomics.

***Addressing access barriers**: reducing the diagnostic and management burden for rare conditions through better access to specialised providers and diagnostic technology so that all children can benefit equally from timely, targeted intervention.*

- **Screening, diagnosis and multidisciplinary care:** Prioritise investment into multimodal and mobile programs that bridge access gaps and build capacity in screening, diagnosis and early intervention for children with rare conditions and developmental co-morbidities, especially within remote and rural locations. Ensuring these services are designed in a way that enhances and supports families in navigating the healthcare system and addresses referral barriers.

- **Psychological care:** Facilitate better access to psychological and peer support for children with rare conditions and their caregivers and families at key milestones and transitions such as diagnosis and entry into early learning and school environments.

- **Funding disparities:** Evaluate current gaps in funding and resources required to support children with complex conditions (rare or common) and associated developmental comorbidities within educational settings. Revise procedures to reduce time to funding for educational support applications to ensure children with rare conditions are not left behind in critical early years.

- **Genetic services:** Improve access to genetic screening, diagnostic services and technology for children presenting with complex developmental delays to facilitate timely diagnosis and intervention. Explore alternative service delivery options such as the establishment of community-based genetic services to deliver accessible care for patients.

***Health system navigation and care coordination**: ensuring children with rare conditions and developmental comorbidities receive timely, well-coordinated care regardless of the complexity or rarity of their condition.*

- **Multidisciplinary care coordination:** Prioritise funding for services that facilitate coordination across needs assessments, referrals, early interventions and treatment, address barriers to accessing services, supporting families with complex needs (whether rare or not) in obtaining comprehensive, multidisciplinary care. Prioritise accessibility to navigation support amongst the most vulnerable.

- **Increase sustainability of peer support and navigation:** Provide funding support to non-profit organisations currently providing navigation services ensuring their continued operation and effectiveness in assisting families facing complex healthcare challenges.

- **Interdisciplinary collaboration**: Facilitate greater interdisciplinary collaboration between the healthcare and education sectors and stronger protocols and mechanisms for information sharing and coordination between healthcare providers, educators, and social service agencies to better meet the diverse needs of children and families living with rare conditions.

Genetic Alliance Australia: Response to NSW Legislative Assembly Committee on Community Services Inquiry on Improving access to early childhood health and development checks.

Genetic Alliance Australia (GAA)

GAA is an umbrella group for rare genetic conditions, many of which are so rare they do not have their own support group. We seek to connect families and individuals affected by the same, or similar condition, and/or provide information on relevant support groups both nationally and internationally. GAA is a member of the Clinical Genetics Executive Committee, NSW Newborn Screening Committee, NSW Register of Congenital Conditions, Consumer Health Forum, EURORDIS, Health Consumers NSW and the International Alliance of Patients Organisations (IAPO).

While this submission was developed in collaboration with representatives from GAA's peer support organisations, we acknowledge that it may not fully capture the diversity of challenges faced by our community. Rather we seek to share lived experience across a number of our contributing members to help illuminate areas of high and common unmet need.

Defining vulnerability in the context of rare presentations

Throughout this submission, we refer to the rare community acknowledging this is a diverse group. The rare community encompasses rare genetic conditions as well as rare genetic variances that should not be classified as disorders These conditions confound healthcare providers during early developmental surveillance and beyond due to their rarity, complexity in the way they present and associated comorbidities.

The unmet need within this community is substantial, and its members tirelessly advocate for their children and families within a system that struggles to comprehend their complex needs. An Australian survey revealed that only a minority of individuals with rare conditions felt supported socially, financially, and psychologically.[6] This highlights the vulnerability of the entire community and underscores a more significant issue: **the need for better access to comprehensive, multidisciplinary cross-sector care that many are not receiving.**

However, we respectfully acknowledge the important social, economic and cultural determinants that further impact the way the rare community accesses and experiences care. Those who face economic difficulties, live in remote or rural areas, identify as Aboriginal and Torres Strait Islanders, or as Culturally and Linguistically Diverse (CALD) or other minority communities. They face additional obstacles in navigating the healthcare system and securing essential health, social, and psychological support. Addressing the layered disparities experienced within these parts of the rare community is crucial to ensuring all children and families have equal access to outcomes.

The role of early developmental surveillance in identifying rare conditions and as a conduit to early intervention

In New South Wales alone, an estimated 7,980[1,2] children are born with rare genetic conditions each year and the majority experience an onset of symptoms during childhood.[2] Rare conditions often present with diverse clinical manifestations, commonly affecting the nervous system and leading to symptoms such as intellectual disability, neuropsychiatric disorders, epilepsy, and motor dysfunction which can all impact a child's ability to meet their developmental milestones.[3]

Every avenue of early developmental surveillance, from routine checks by general practitioners and early childhood nurses to observations by early learning and childhood educators, holds critical potential for

intervention for infants and children grappling with developmental challenges linked to rare conditions. While many conditions lack a cure, early, age-specific multidisciplinary intervention can dramatically alter a child's outlook, affecting their clinical prognosis,[4] capacity for learning and advancement and ultimately quality of life.[7,8]

This has important implications when considering:

- The current construct and ability of early childhood checks to identify and act on developmentally correlated symptoms of rare genetic conditions is hindered by a limited understanding of rare conditions and knowledge of emerging genetic diagnostics.[4] Addressing these factors could mitigate the cost and burden associated with the diagnostic odyssey for families and the healthcare system.[9]

- The opportunistic and non-mandatory nature of developmental surveillance in primary care settings, in the absence of specific MBS item codes or other appropriate incentives, can hinder a comprehensive approach to investigating developmental symptomatology.

- Disparities in access to and expertise of healthcare professionals, referral challenges and long waitlists for geneticists, genetic counsellors, developmental experts, and other multidisciplinary providers creates a system where access to care for children with rare conditions often depends more on luck than fairness. This perpetuates inequities in clinical and developmental outcomes and in some cases can result in children "aging out" of eligibility for important treatments.

- Poor access to early developmental intervention programs such as school readiness and inclusion programs for (but not limited to) children with rare conditions and associated developmental needs resulting in social and educational disadvantage.

In recognition of these considerations and in line with the UN resolution on rare diseases,[10] which asserts that every child with a rare condition has the right to a timely diagnosis, appropriate intervention, and the opportunity to participate in society to the fullest of their unique potential, our submission focuses on the following three areas:

1) **Education and capacity building**: improving the capability and capacity of all providers participating in early developmental surveillance in identifying, appropriately referring and identifying support needs for children with rare conditions through education, skills building and incentives.

2) **Addressing access barriers**: reducing the diagnostic and management burden for rare conditions through better access to specialised healthcare providers and diagnostic technology so that all children can benefit equally from targeted intervention. Mitigating important disparities resulting from economic, social, geographic and cultural barriers that prevent equity of access to the above or exclusion from important early intervention, social and educational opportunities that aid in development.

3) **Health system navigation and care coordination**: ensuring children with rare conditions and developmental comorbidities receive timely, well-coordinated care regardless of the complexity or rarity of their condition.

ToR 1. Changes needed to address gaps in outcomes for vulnerable children, including those in rural and remote communities, Aboriginal communities, and culturally and linguistically diverse communities.

Gaps in healthcare provider capacity and capability:

For many families, consulting a general practitioner (GP) or early childhood and family nurse (ECFN) regarding developmental concerns is a crucial step in the journey to a rare diagnosis. However, the management of these concerns frequently hinges on the individual provider's knowledge, attitudes, and experience with complex clinical cases. A study of 462 Australian children with rare conditions showed that nearly 40% consulted six doctors before receiving a diagnosis, highlighting poor awareness among healthcare providers as the most significant barrier to timely and accurate diagnosis and intervention.[4] This has profound implications when considering the effectiveness of the existing Personal Health Record (Blue Book) developmental surveillance is contingent upon provider skill in eliciting and interpreting developmental anomalies.

The current screening approach is further compromised by an over-reliance on the Parents' Evaluation of Developmental Status (PEDs), which, when presented, may often be incomplete and is predominantly composed of closed, self-assessed questions. This format can limit the full disclosure crucial for the diagnosis of rare conditions. Additionally, the early symptoms of such conditions often manifest non-specifically, necessitating that both GPs and families are provided with sufficient time during consultations to discuss any concerns, even those seemingly insignificant. The voluntary, time-constrained nature of these checks can obstruct early detection of risk factors, underscoring the need for a more thorough and appropriately remunerated and incentivised approach.

While time constraints may pose less of a concern for ECFN's, our community has observed a notable variance in experience and provider skill in managing rare presentations. This diversity has led to a spectrum of encounters, ranging from positive to negative. Some individuals reported instances of judgement, or developmental delays being incorrectly attributed to more common causes and therefore insufficiently referred and investigated.

> *"As a first-time mum presenting with a baby who was struggling to feed with a cleft, I was made to feel like I was intentionally not feeding my daughter, instead of identifying causes and connecting me with support, it was horrific."*
>
> ***Mother to a baby with SATB2 Associated Syndrome***

Given that our community noted that ECFNs were primarily utilised by first-time mothers, these factors can have enduring psychological effects on parents and directly influence the timeliness of intervention and assistance-seeking.

Dismissal, minimisation of parental concerns

Parents of children with rare conditions frequently report instances where the dismissal of early concerns has directly impeded diagnosis costing precious time, creating profound stress for the family and most alarmingly, altering the prognosis for the child.

> *"We were repeatedly told that we shouldn't compare, we need to let her develop in her own time. We instead took it upon ourselves to find therapists who could possibly help us understand why our daughter couldn't talk or walk yet. We wish we had known sooner about her ultra-rare genetic condition, it would have given her early intervention and reduce the gaps of her developmental needs and regression."*
>
> ***Mother to a baby with SATB2 Associated Syndrome***

Inconsistent or underutilisation of surveillance tools

In other instances, incorrect or approximate use of surveillance tools contributed to delays in early intervention. In the instance of rare growth hormone deficiencies, examples of poorly conducted height surveillance (for example shoes remaining on or measuring charts incorrectly applied to walls), coupled with inconsistency in approach has led to delayed diagnosis and referral. This has led to delays in qualifying for Pharmaceutical Benefits Scheme (PBS) funded medication where 12 months of correctly tracked data were required for eligibility.

> *"It was incredibly stressful trying to gain second opinions and then having to pay for medication privately because we didn't have all the correct measurements we needed, but seeing my child anxious and self-conscious and wondering what this means for them as they go forward through life was the worst part."*
>
> ***Mother of a child with a delayed growth hormone deficiency diagnosis.***

Inappropriate intervention and informed decision-making

Rare genetic differences that are not (and should not be) classified as conditions can further confound healthcare and service providers during early developmental surveillance and beyond. An important illustrative example is children with innate variations of sex characteristics. The significance of these variations is influenced by a number of factors including the presence of physical and developmental symptoms, how families assign meaning to these variances, implications on identity and wellbeing and the urgency of intervention (i.e. life-saving versus elective intervention). Families are often "unseen" within systems and inadequately supported to make informed decisions.[11,12] Better provider education, service accessibility and inclusive practices are required to ensure children with rare genetic differences can thrive emotionally, physically and socially.

> *"It still triggers an emotional response every time I replay what happened to me and my baby when he was born. I remember coming out of the anaesthetic with white-coated doctors surrounding my bed, telling me there was something wrong with my baby, and a simple operation would "fix" him, this is before I had even held him for the first time. I was given no support, no education, no other options. Now, I understand the "fix" was cosmetic in nature. I'm glad I didn't succumb to the pressure exerted on me by those clinicians. But many do, thinking they are making the right decision for their child with knowing all the factors."*
>
> ***Mother of a child with an innate variation in sex characteristics.***

Within the scope of this inquiry, we encourage the committee to consider:

Education and capacity building:

- Increase the capacity of all key stakeholders involved in assessing early developmental milestones in identifying developmentally correlated symptoms of rare genetic conditions and available genetic diagnostic services. *(Note, this is further discussed in ToR3).*

- Increase parent's access to more detailed and interactive developmental assessment tools, that better continually monitor progress over time and are easily shared across providers. Improve parent education on the importance of developmental surveillance and the availability of resources in pre-natal settings.

- Encourage comprehensive early childhood checks through the introduction of adequate incentives and enhancement of provider-patient dialogue in consultations, including advocating to Federal bodies for introduction of Medicare Benefits Schedule (MBS) item codes where appropriate.*

Gaps in psychosocial care for children and families:

The chronic, often degenerative and life-limiting nature of rare conditions can be devastating and life-changing and the onset of symptoms in childhood places parents at the centre of navigating the uncertainties and stress associated with rare conditions.[13] Evidence clearly indicates families dealing with rare conditions are at heightened risk of developing mental health issues including stress, anxiety, isolation, loneliness and grief.[13] Despite widespread acknowledgment that psychological support should accompany diagnosis, research indicates that less than half of families currently receive adequate assistance.[4]

The critical importance and interdependency of children and parent's psychosocial needs cannot be understated. Parents who are adequately supported themselves have a greater capacity to engage in effective communication with healthcare providers, seek out necessary resources and make informed decisions about their child's care. Additionally, parents who experience adequate psychosocial support for their children report greater satisfaction with care.[4]

At the child level, gaps in psychological care can significantly impact developmental outcomes by affecting their emotional, social, and cognitive growth. Inadequate support may lead to heightened anxiety and stress,[6,7] impeding learning, relationship-building, and emotional regulation. Furthermore, research indicates that school-aged children with rare conditions and genetic differences often encounter higher rates of health-related stigma, bullying, and difficulties in self-care and mental health within mainstream education systems.[7,11] The absence of timely, appropriate psychological interventions can worsen existing conditions or trigger new problems, affecting the child's well-being and limiting their participation and success in early learning, school, and other enriching community activities.

These findings emphasise early psychosocial intervention and care for children and their caregivers is essential in managing rare conditions and associated developmental comorbidities and in fostering early learning and school environments where children with rare conditions can thrive emotionally, socially, and educationally.

Gaps in care coordination and system navigation:

The above contextualises why navigating within and across the health system, social services, National Disability Insurance Scheme (NDIS) and education sectors for families living with rare conditions is not just complex; it's an emotionally draining and often disheartening journey, particularly for those in vulnerable circumstances.

Families frequently juggle multiple care streams due to comorbidities, navigate complex referral pathways, contend with inequitable access to specialist healthcare providers, shoulder high costs for diagnosis and intervention, and endure prolonged wait times for services, including critical NDIS resources. The genetic nature of rare conditions can create variability in disease progression, and life events such as unexpected illnesses or accidents can dramatically change a child's trajectory.

> *"If he gets a sudden or unexpected illness, he may require a cough assist machine or breathing machine because of his damaged muscles and lack of dystrophin to repair them, he may not have enough strength to go off the machine. If he didn't have an unexpected illness (like a Flu, COVID,*

*GAA recognises that Medicare and related MBS item numbers are subject to Federal jurisdiction but acknowledges the important role of provider remuneration in optimising consultation formats for surveillance discussions where appropriate.

common cold, lung infection, or pneumonia) it would be expected he may not have needed this equipment few years. But we need systems that are responsive and agile."

Parent to a child with Duchenne Muscular Dystrophy (DMD).

Health systems and the NDIS, alongside early learning and school systems, often lack the agility and flexibility to quickly adapt to these changing needs, which can adversely affect their health outcomes and prognosis, limiting their overall potential for recovery and quality of life. Despite the clear recognition of these challenges, only a small percentage of individuals have access to a dedicated care coordinator, which has been recognised as a critical unmet need within the National Strategic Action Plan for Rare Disease.[5]

Currently, not-for-profits play an important role in bridging across care coordination and health system navigation needs. For instance, national peak body, Rare Voices Australia is currently implementing a Rare and Complex Disease Telehealth Nurse Program[14]. Save Our Sons Duchenne Foundation[15] has financed neuromuscular nurses to meet the care coordination needs of children with Duchenne Muscular Dystrophy in major Australian hospitals and The Friendly Faces Helping Hands Foundation[16], based in Glen Innes, supports hundreds of rural NSW families navigate the complexity of accessing care away from home. These are only three of many examples and while incredibly valuable, the sustainability of these types of services is uncertain without sufficient fundraising and resources, underscoring the urgent need for wider access to comprehensive healthcare and social support services.

Addressing this gap more holistically within the health ecosystem would have broad reaching benefits, reducing the burden on families and ensuring more equitable and efficient access to necessary services for both children with rare complex conditions and those with more common but equally complex needs.

Within the scope of this inquiry, we encourage the committee to consider:

- **Health system navigation**: Facilitate better access to services, strengths-based and trauma-informed psychological and peer support at key intervention points across the diagnostic odyssey and important milestones and transitions such as entry into early learning and school environments.
- **Health system navigation:** Increase funding support to non-profit organisations currently providing navigation services, ensuring their continued operation and effectiveness in assisting families facing complex healthcare challenges while more sustainable options are developed.
- **Care coordination**: Prioritise funding for services that facilitate coordination across needs assessments, referrals, early interventions and treatment, address barriers to accessing services, supporting families with complex needs (whether rare or not) in obtaining comprehensive, multidisciplinary care.

Considerations for Aboriginal and Torres Strait Islander children and children from Culturally and Linguistically Diverse (CALD) families

Perceptions of rare conditions and their causes are influenced by societal, cultural values and beliefs. This is heightened by language barriers in already challenging exchanges between providers and families. [17,18] Consequently, complex decision-making regarding investigations and interventions that may have profound lifelong impact may not be fully understood. This may impact the use of medications, allied health interventions and available support mechanisms.

Social stigma surrounding certain rare conditions may discourage families from seeking medical attention or disclosing symptoms during developmental surveillance checks, thereby delaying diagnosis and subsequent support. The lack of cultural safety experienced within mainstream care settings further contributes to feelings of alienation and reluctance to seek medical assistance which in turn further hinders the ability to navigate the healthcare system.

While Aboriginal and Torres Strait Islander communities may not inherently experience a higher incidence of rare diseases, a lower median age suggests a higher likelihood of encountering rare diseases among this population group, given that such conditions typically manifest early in life.[17] Despite this Aboriginal and Torres Strait Islander children and their families face complex challenges in accessing equitable early developmental surveillance, diagnoses and referrals for rare genetic conditions.[17-19] Recent evidence suggests that even when developmental delays or other indicative symptoms indicate the potential presence of a rare genetic condition access to confirmatory genetic services is impacted by referral hurdles, limited access to specialists, racial biases, and socioeconomic and geographic barriers.[18,19] Confirmation of a rare genetic diagnosis is made more difficult as a reference genome and limited genetic data for this population group are not yet available.[5]

The biomedical medical model used within the NSW health system can result in inflexible consultation formats that may not accommodate family structures and decision-making.[18] A lack of consideration for important cultural beliefs and norms can exacerbate stigma and psychosocial stressors associated with heritable genetic conditions.[19] Additionally, as genomics intersects with traditional concepts of identity, kinship and country, any intervention for children with rare genetic conditions or differences must be culturally sensitive, respectful of Indigenous knowledge systems, and inclusive of community voices and perspectives.[17]

CALD communities likewise encounter similar issues in engagement with the health system. An understanding of other cultures' perceptions towards medical interventions may be unique to each cultural group. Therefore, tailored responses and solutions need to be co-developed depending on the needs of that community.

Within the scope of this inquiry, we encourage the committee to consider:

- **Education and capacity building:** Support the development of Aboriginal and Torres Strait Islander and CALD community-led initiatives designed to support awareness, identity and understanding of rare genetic conditions and genetic screening, fostering genetic literacy in a manner that respects and integrates the distinctive cultural perspectives on genomics.

- **Health system navigation:** Facilitate community co-design of multidisciplinary models of care for rare conditions and developmental co-morbidities for Aboriginal and Torres Strait Islander and CALD children.

ToR 2. Barriers that affect parents' access to routine health and development checks that track their child's progress against developmental milestones.

Barriers to specialists, allied healthcare and developmental support services

One of the most prevalent challenges affecting equitable screening and intervention for developmental comorbidities is the chronic shortage of paediatric developmental healthcare providers, genetic counsellors and clinical geneticists.

The shortage of public paediatricians across NSW, particularly those specialising in developmental challenges is a pervasive issue that is even more acute in remote and rural settings. Consequently, children can face delays of 2-3 years before being seen by a developmental unit following initial screening, and an additional 2-3 years before accessing genetics services. This leads to a cascade effect where children may be 6-8 years old before receiving a diagnosis, by which time they have missed crucial early intervention opportunities. This delay not only hinders their developmental progress but also places an undue strain on families and the healthcare system, emphasising the urgent need for systemic improvements to paediatric care access and coordination.

This situation is further impacted by shortages of occupational therapists, speech pathologists, physical therapists, behavioural therapists, and psychologists, leading to additional delays in intervention and fragmentation of care. Navigating across different service streams, convoluted and confusing referral systems and overly bureaucratic processes detract from the delivery of crucial, time-sensitive services, such as child and maternal mental health services, that families critically need.

Moreover, the problem is compounded when public hospitals refuse patients for being 'out of area,' despite their obligation to serve all children, highlighting a significant systemic issue.

Geographic barriers and disparities

The above challenge is significantly amplified in remote and rural locations across NSW. Over 50% of children with rare conditions received their diagnosis only after being referred to a clinical specialist at a large metropolitan paediatric hospital.[4] These delays worsen outcomes for children but also leave parents feeling socially isolated and emotionally distressed,[20] and impacted financially by reduced employment, travel and accommodation costs.

For families that must travel frequently for care, maintaining regular follow-ups and managing ongoing treatment becomes more challenging, which can impact continuity of care, the effectiveness of treatment and overall health management. This is further compounded by the complexity of cross-border care and differing approaches to developmental surveillance for NSW residents of border towns accessing care interstate.

In consideration of the retirement of Royal Far West's Healthy Bus Stop initiative and subsequent planned transition to the government's Brighter Beginnings program, the following observations were made. Although perceived as highly valuable for its multidisciplinary approach to assessment and coordinated care plans, the capacity of local health services to follow up on referrals was limited, resulting in delays and insufficient support for identified children.

> *"The referral capacity of Allied health couldn't support the referrals that were being passed on and didn't pass them on to other service providers within the community, so you end up with kids who have a lot of learning difficulties, that could be attributable to diseases, who are never properly diagnosed and supported. Those are the kids who become the naughty kids who play up at school because they're always pushed down, they don't fit into society. And honestly, a lot of those kids are the kids who are suicidal."*
>
> ***Rural Not-for-Profit Support Provider.***

There is hope that the Brighter Beginnings initiative, which includes the implementation of health and development checks in preschools may help identify more children requiring intervention, however, these children will still face the same barriers to multidisciplinary physical and psychological care, which cannot always be sufficiently addressed through initiatives like telehealth alone due to the often-physical nature of assessments.

The lack of timely services impacts not only impacts early learning and social development. The impacts continue into secondary school and later years, necessitating funding into education programs, mental health programs and justice interventions [28].

Cost barriers and disparities

Gaining an etiological diagnosis for developmental symptomatology, particularly in the case of rare conditions leads to direct, indirect, and opportunity costs for rare families. Delayed diagnosis may shift healthcare spending away from treatment and supportive therapies towards unnecessary procedures that cost both the families and the healthcare system.

Direct costs of diagnosis and intervention, such as genetic testing, specialist consultations, and experimental or off-label treatments, may not be fully reimbursed by Medicare or private health insurance, resulting in considerable out-of-pocket expenses for families. Additionally, families often bear the brunt of indirect costs, including reduced employment engagement, caregiving burdens, home modifications, secondary treatment expenses, and travel and accommodation costs. For children from families with low resources to navigate these costs, this can mean delayed or foregone essential services, exacerbating developmental delays and reducing their chances of reaching their full potential.

Within the scope of this inquiry, we encourage the committee to consider:

- **Multidisciplinary care:** Prioritise the development of and investment into innovative mobile programs to facilitate frequent, multidisciplinary care for children with rare genetic conditions and developmental co-morbidities, especially within remote and rural locations.
- **Health system navigation:** Prioritise the development of and investment into dedicated care coordinators or patient navigators to help families manage the complexity of the healthcare system, including scheduling appointments with specialists, arranging necessary tests, and ensuring that care is continuous and coordinated across different healthcare providers. Prioritise accessibility to navigation support amongst the most vulnerable.
- **Address access barriers:** Improve access to genetic services for children presenting with complex developmental delays to reduce unnecessary spending in diagnostic odyssey. Consider measures to reduce financial barriers associated with healthcare costs for families affected by rare genetic conditions and their co-morbidities.

ToR 3. Recruitment and retention of health professionals to address workforce shortages.

Bridging the gap in the rare specialising and genomics workforce

The landscape of rare condition management in Australia is marked by a pronounced lack of awareness and workforce capacity. As genomic testing becomes increasingly prevalent, the demand for specialised professionals like genetic counsellors, clinical geneticists, and genetic pathologists has surged, creating a significant gap between supply and demand.[21,22] This gap is exacerbated by the concentration of expert genetics services in metropolitan areas, leaving those in rural and remote regions with limited access. [22] Furthermore, parents with adequate resources will travel to seek appointments in larger centres to ensure timely access and continuity of care. This places an additional burden on the metropolitan workforce and distorts the data on the regional rare health needs.

The breadth of rare conditions and complexity in the presentation of symptoms has resulted in often insufficient coverage of rare conditions as well as advances in genomic technology within existing medical curriculums and frameworks,[22] underscoring the pressing necessity for comprehensive training and support to meet the escalating demand for genetic services.

Mainstreaming of genomics and new technologies

In a recent review of the integration of genomics into the National Health Service (NHS) in the United Kingdom, it was emphasised that genomic education should be tailored to the specific roles of staff, delivered in a timely manner to align with technological advancements, and supported by incentivised engagement strategies.[23] An Australian review noted that education should vary across disciplines and health conditions, and include education on the translation of genomics into clinical practice, related governance requirements as well as changes to professional practice needed to implement the new technologies.[24]

Retention of healthcare professionals

GAA acknowledges this specific ToR will be better addressed by those working directly within service provision. However, we emphasise the complexity and time demands of investigating developmental delays or symptoms of rare conditions, which exacerbate burnout and strain on an already under-resourced workforce. Earlier access to genetic testing and improved care coordination for families beginning the diagnostic journey could help alleviate these challenges.

In line with ToR 1 recommendations, we encourage the committee to consider:

Education and capacity building:

- Collaboration with universities, medical colleges, and professional associations to access continuing professional development (CPD) accredited short courses and micro-credentials to rapidly upskill NSW Health staff on genetics and genomics in areas relevant to their fields.
- Increasing awareness and exposure to the field of genomics during early training and interdisciplinary training that includes aspects of rare diseases and genetic counselling to enhance skills and interest.
- Partnerships between medical institutions and rare disease organisations for clinical rotations and internships to provide hands-on experience, making these specialties more appealing and accessible to emerging healthcare professionals.

ToR 4. Funding for early intervention programs and screening to ensure children are given support for developmental issues, including telehealth and other models.

Telehealth for rare conditions and developmental comorbidities

Telehealth continues to be an important vehicle in diagnosis and ongoing care for children with rare conditions and associated developmental co-morbidities as it enables families to access specialised services without the burden of travel which is often difficult with a high-needs child. Additionally, telehealth can provide important insights into a child's behaviour within their home setting.

However, in the context of developing screening, early intervention and care provision it is important to note the following:

- Rare conditions (like many other more prevalent conditions) requiring a multidisciplinary approach can be difficult to coordinate without sufficient centralisation, leading to fragmented care for children and families disproportionately relying on telehealth services.

- Monitoring the progression of rare conditions, particularly those with neurological degeneration necessitates frequent in-person assessments over time. Telehealth may struggle to provide the consistency and accuracy required to track disease progression effectively. This is exacerbated in regions with limited healthcare resources.

- Some rare community members, particularly those in remote or disadvantaged communities, may face barriers to accessing or using telehealth technology due to factors such as limited internet connectivity, digital literacy, or language barriers. These challenges can impede their ability to engage effectively in virtual consultations for rare condition management.

- **Access to multidisciplinary care is crucial, yet the benefits are profoundly amplified when care is coordinated and rooted in a deep understanding of rare conditions and the specific care context, significantly improving the treatment experience and outcomes for families.**

Multidisciplinary outreach and mobile clinics

Mobile multidisciplinary allied health outreach clinics provide a dynamic approach to bridging gaps and overcoming access barriers in screening and care provision. These models can be designed to address common needs of both more prevalent and rare conditions requiring multidisciplinary care. Mobile outreach clinics can specifically target communities that are underserved or have limited access to healthcare services, reducing disparities and ensuring more equitable healthcare access across NSW.

Additionally, the following benefits are noted:

- **Education and capacity building:** By working closely with local healthcare providers, mobile clinics help build capacity for managing rare conditions within local healthcare systems. This includes training on rare conditions and managing developmental comorbidities, sharing best practices, and establishing referral pathways to specialised services when necessary. Mobile outreach can also support important capacity building within local schools that support children with rare conditions and developmental comorbidities in their catchments.

- **Health system navigation and care coordination:** Mobile outreach is a valuable conduit to information and integrated condition management strategies for children and families which helps to build capacity in navigating the healthcare system effectively.

- **Adress access barriers:** An integrated approach across telehealth and mobile outreach addresses many of the accessibility and equity gaps in healthcare system experienced across NSW.

- **Future intervention planning:** The programs can also collect valuable data on the prevalence of rare or more common complex cases across different regions, outcomes for children, and the effectiveness of various treatment approaches which is vital to inform future healthcare planning and policy across NSW.

School readiness programs for children with rare conditions and complex needs

School readiness programs (particularly those on-premises) significantly enhance the educational transition for children with rare conditions by systematically introducing them to their future learning environments. These programs directly contribute to effective early intervention by enabling schools to actively identify and plan for

each child's specific needs ahead of time. Through early familiarisation with the school setting, children gain confidence and adapt more easily, while educators gather essential information to develop tailored accommodations and support strategies. This preparation allows for a more immediate implementation of Individual Education Plans (IEPs) and other necessary adjustments as soon as the child begins their education.

GAA notes with interest, the Wellbeing and Health In-reach Nurse Coordinator program partnership between NSW Health and the NSW Department of Education that is currently being implemented within selected schools across NSW.[25] The remit and objectives of this program strongly align with the multifaceted unmet needs experienced by children with rare conditions specifically in coordinating early intervention, assessments and referral of students and families to services and programs, contributing to care coordination and working to overcome referral barriers.

In line with recommendations from ToR 1,2,3 and 5 we encourage the committee to consider:

- **Multidisciplinary care/health system navigation/addressing access barriers:** Prioritising investment into services that bridge access gaps and build capacity in screening and early intervention for children with rare conditions and developmental co-morbidities, especially within remote and rural locations.
- **Multidisciplinary care/health system navigation/addressing access barriers:** Ensuring these services are designed in a way that enhances and supports families in navigating the healthcare system and in transitioning to the school system, including where relevant capacity building and resourcing for schools.

ToR 5. Any other related matters.

The importance of investment in advancing Newborn Screening capability

Many rare conditions can lead to significant disabilities and comorbidities through prolonged diagnostic delay. The NSW Newborn screening (NBS) allows for the early detection of some rare conditions, often before symptoms appear. This is critical in reducing the burdensome rare diagnostic journey and facilitating early intervention strategies that aim to manage symptoms, prevent complications, and optimise developmental outcomes. Universal screening at birth can help mitigate the diagnostic disparities and barriers discussed throughout this submission for example in Pompe disease (PD), which is a rare, autosomal-recessively inherited deficiency in the enzyme acid α-glucosidase. In affected infants' prognosis is poor, and without treatment most infants die within the first year of life. [26] Anecdotally, support organisations have described significant geographical disparities in age of diagnoses ranging from 3 months in urban centres to 6-7 months in remote and regional locations, primarily due to paediatrician access challenges.

> *"To lose a baby in their first year of life to a rare disease causes much regret, guilt, and loneliness to parents, family, and friends. To lose a baby needlessly when there is an effective treatment amplifies this sadness."*[26]

Furthermore, technological advancements in genomic sequencing and the development of gene and RNA-based therapies present significant opportunities to enhance Australia's Newborn Bloodspot Screening (NBS) program.[25] The ability to efficiently extract DNA from dried blood spots has allowed Australia to become one of only nine countries to recommend screening newborns for conditions like spinal muscular atrophy (SMA).[27] Since the inclusion of SMA screening in NSW and ACT's NBS programs in July 2022, there has been a marked improvement in identifying at-risk newborns and facilitating timely care.[27] Coordinated expansion of genomic NBS (gNBS), aligned with the advent of new and cost-effective treatments, will enhance the healthcare

system's capacity to improve outcomes for newborns with rare genetic conditions, therefore, strategic investment in gNBS is not only a medical imperative but also a step towards equity in healthcare for all Australian newborns.

We encourage the committee to note:

- The importance of NBS in reducing the diagnostic odyssey for rare conditions presenting with non-specific developmental delays and supporting existing efforts to improve the increased sustainability, equity, and transparency within the NBS program in NSW.
- The time sensitivity of listing of conditions on NSW NBS Program once federal endorsement is made, including Pompe Syndrome which is currently listed for review by MSAC 1775.
- Importance of investing in genomic newborn screening capabilities, workforce and resources to position NSW Health as a leader in the implementation of gNBS and newborn genomic health.

Equitable access to genetic services and genetic counsellors

Despite advancements in the diagnosis of rare conditions and the ongoing progress in genetic and genomic research, many Australian families continue to face significant barriers in accessing genomic testing.[4,22]

Efforts to improve outcomes for children with genetic rare conditions necessitate early, rapid, and precise genetic diagnosis. Such diagnoses offer multifaceted benefits, allowing for the implementation of targeted interventions, personalised treatment strategies, and informed family planning decisions.

Early developmental checks provide important intervention points to trigger referrals to genetic and genomic services. However, the process is impacted by low awareness amongst some referring healthcare professionals, cost to access, and prohibitive waitlists for some genetics/genomics services (including up to 4 years for non-urgent attendance) and a lack of available staff. This is further compounded by the low genetic expertise for General Practitioners, sub specialists and allied health providers.[22] Additionally, this landscape is further affected by a lack of MBS item numbers for genomics and genetic counsellors, as without item numbers to cover the cost of services many of these barriers cannot be sufficiently addressed.

Within the scope of this inquiry, we encourage the committee to consider:

- **Education and capacity building**: Investment into increasing genetic literacy within healthcare professionals undertaking early developmental checks and increasing awareness of genetic services and timely referral for etiological genetic diagnosis.
- **Addressing access barriers:** Explore alternative service delivery options such as the establishment of community-based genetic services, to leverage the capabilities of Genetic Counsellors, GPs, allied health providers and pharmacists to deliver accessible care for patients. This could include telehealth capabilities to triage requests and manage capacity demands.

The transition to school – developmental surveillance and support deficits

Many rare conditions will manifest symptoms in school-aged children. Early childhood and primary school settings are uniquely placed to help identify the developmental co-morbidities that accompany rare conditions but gaps in understanding, capability, surveillance mechanisms and resourcing can result in missed opportunities for early intervention, impacting the child's educational and developmental outcomes.

Additionally, children transitioning into the mainstream school system with a rare condition are often disadvantaged by the inequitable availability of support units, previous experience and varying investment into supporting children with developmental complexities. Research highlights that while accessible, inclusive education is a primary concern for parents, many struggle to articulate their child's educational needs to schools and require assistance in explaining their child's condition to their educators and peers.[6]

The creation of effective Individual Education Plans (IEP) tailored to a child's abilities, curriculum alignment, and access to funding for necessary accommodations or adjustments varies widely among schools and often involves a protracted process. This variability can lead to considerable delays in obtaining timely support and intervention, with some community members reporting waits of 6-12 months. Such delays during this foundational period can have profound downstream implications on a child's learning trajectory. In extreme cases, these challenges have resulted in children with developmental vulnerabilities exiting the school system altogether.

A survey of parents with school-aged children with rare conditions attending mainstream schools in WA revealed significant gaps in psychosocial care.[7] Findings indicated high levels of health-related stigma (75.6%), notable mental health difficulties (43.9%), and bullying (46.4%) highlighting the need for support to address the unique psychological and social challenges faced by these children.[7] The findings echo those of the Disability Royal Commission, which also highlighted that mainstream schools require major reforms to overcome the barriers that prevent students with disability, especially those with intellectual disability, from accessing safe, equal, and inclusive education.[8]

> *Children with rare conditions have a right to equitable access to educational opportunities that accommodate their unique needs, foster their development, and enable their full participation in academic and social activities, ensuring that they can reach their full potential despite the challenges they may face.*[10]

As discussed in ToR 4 initiatives such as the Wellbeing and Health In-reach Nurse Coordinator program[25] partnership between NSW Health and the NSW Department of Education strongly align with the multidisciplinary needs experienced by children with rare conditions. In particular this program can coordinate early intervention, assessments and referral of students and families to services and programs, contributing to care coordination and working to overcome referral barriers. The expansion of these services to support developmental surveillance would bridge an important and valuable gap for children and families experiencing the potential onset of rare conditions in these early school years. Most importantly, a person-centred approach to learning for children with complex needs necessitates an integrated strategy, combining the efforts of informed and skilled teachers, health in-reach services within educational settings, and timely access to allied health services.

We urge the committee to consider the following:

- **Education and capacity building:** Early childhood educators and teachers require more comprehensive training and support in managing children with complex developmental challenges stemming from rare conditions, including comorbidities and psychosocial needs. At a practical upstream level this requires working towards integrating rare condition and genetics education and practical disability placements into tertiary-level educational curriculum frameworks to foster deeper understanding and practical skills.

- **System navigation and coordinated care:** Greater interdisciplinary collaboration between the NSW healthcare and education sectors with robust protocols and mechanisms for information sharing and coordination. This requires a multi-pronged approach, including capacity building and upskilling of educators (as above) and expanded access to targeted programs such as the Wellbeing and Health In-reach Nurse Coordinator program and better access to developmental allied health services to help bridge the health and education resources to ensure children feel safe, supported and are able to reach their fullest potential. This multipronged approach also needs to encompassand social service agencies required to meet the holistic needs of children and families living with rare conditions.

- **Addressing access barriers:** Reducing time to funding for educational support applications and resources outlined in Individual Education Plans to ensure children with rare conditions are not left behind in critical early years.

Conclusion

In line with the National Strategic Action Plan for Rare Disease, which calls for the voice of people living with a rare disease, we have emphasised the importance of the lived experiences of our membership and their engagement with early childhood developmental checks. We also acknowledge the complex journey of diagnosis, intervention, and support for all families and children with rare conditions. It is imperative that the voice of families and carers be embedded throughout the structures and systems that impact those with rare conditions in early childhood and their development to enable them to reach their full potential.

While recognising that some issues may extend beyond the committee's direct jurisdiction, we urge the committee to explore all avenues for addressing disparities faced by the rare community, especially among minority and rural populations.

Through any potential interventions arising from this inquiry and Genetic Alliance Australia invites further opportunity to engage with the committee on these important issues.

References:

1. Australian Bureau of Statistics (ABS). (2022) Statistics Births and fertility rates for Australia, states and territories, and sub-state regions. Accessed: https://www.abs.gov.au/statistics/people/population/births-australia/latest-release

2. Murdoch Children's Research Institute (MCRI) (n.d) Rare Disease Flagship Program. Accessed: Australian Bureau of Statistics (ABS). (2022) Statistics Births and fertility rates for Australia, states and territories, and sub-state regions. Accessed: https://www.abs.gov.au/statistics/people/population/births-australia/latest-release

3. Kvarnung, M., & Nordgren, A. (2017). Intellectual Disability & Rare Disorders: A Diagnostic Challenge. Advances in experimental medicine and biology, 1031, 39–54. https://doi.org/10.1007/978-3-319-67144-4_3

4. Zurynski Y, Deverell M, Dalkeith T, et al. Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays. Orphanet J Rare Dis. 2017;12(1):68. Published 2017 Apr 11. doi:10.1186/s13023-017-0622-4

5. Commonwealth of Australia. Department of Health. National Strategic Action Plan for Rare Diseases. Canberra; (2020). https://www.health.gov.au/sites/default/files/documents/2020/03/national-strategic-action-plan-for-rare-diseases.pdf

6. Pelentsov LJ, Fielder AL, Laws TA, Esterman AJ. The supportive care needs of parents with a child with a rare disease: results of an online survey. BMC Fam Pract. 2016;17:88. Published 2016 Jul 21. doi:10.1186/s12875-016-0488-x

7. Adama, E.A, D. Arabiat, D. M. J. Foster, E. Afrifa-Yamoah, K. Runions, R. Vithiatharan & A. Lin (2023) The psychosocial impact of rare diseases among children and adolescents attending mainstream schools in Western Australia, International Journal of Inclusive Education, 27:12, 1273-1286, DOI: 10.1080/13603116.2021.1888323

8. Australian Government Royal Commission into Violence, Abuse, Neglect and Exploitation of People with Disability. Final Report - Executive Summary, Our Vision for an Inclusive Australia and Recommendations. Published 2023. Accessed: https://disability.royalcommission.gov.au/publications/final-report-executive-summary-our-vision-inclusive-australia-and-recommendations

9. Cost of Delayed Diagnosis in Rare Disease - a health economic study (2023) https://everylifefoundation.org/wp-content/uploads/2023/09/EveryLife-Cost-of-Delayed-Diagnosis-in-Rare-Disease_Final-Full-Study-Report_0914223.pdf

10. United Nations General Assembly (2022). Resolution on Addressing the challenges of persons living with a rare disease and their families Accessed: https://www.rarediseasesinternational.org/wp-content/uploads/2022/01/Final-UN-Text-UN-Resolution-on-Persons-Living-with-a-Rare-Disease-and-their-Families.pdf

11. Victorian Department of Health and Human Services (2018) Health and wellbeing of people with intersex variations: information and resource paper. Initially prepared by T. Jones and W. Leonard; revised and edited J. Rostant on behalf of Department of Health and Human Services 2016–2018 Intersex Expert Advisory Group. Department of Health and Human Services: Melbourne. Available at Health of people with intersex variations

12. Carpenter M. Bodily integrity, and eliminating harmful practices. Genomics and genetic selection. Healthcare pathways. In: Intersex Human Rights Australia [Internet]. Accessed: https://ihra.org.au/health/

13. Atkins, J.C., Padgett, C.R. Living with a Rare Disease: Psychosocial Impacts for Parents and Family Members – a Systematic Review. J Child Fam Stud 33, 617–636 (2024). https://doi.org/10.1007/s10826-024-02790-6

14. Rare Voices Australia (RVA) (June 2023). Rare and Complex Disease Telehealth Nurse Program. Accessed: https://rarevoices.org.au/grant-update-rare-and-complex-disease-telehealth-nurse-program/#/

15. Save our Sons Duchenne Foundation: https://saveoursons.org.au

16. Friendly Faces, Helping Hands Foundation: https://www.friendlyfaces.info

17. Baynam G, Molster C, Bauskis A, Kowal E, Savarirayan R, Kelaher M, Easteal S, Massey L, Garvey G, Goldblatt J, Pachter N, Weeramanthri TS, Dawkins HJS. Indigenous Genetics and Rare Diseases: Harmony, Diversity and Equity. In: Posada de la Paz M, et al. (eds.), Rare Diseases Epidemiology: Update and Overview, Advances in Experimental Medicine and Biology 1031. Springer International Publishing AG; 2017. Chapter 27. https://doi.org/10.1007/978-3-319-67144-4_27

18. Dalach, P., Savarirayan, R., Baynam, G. et al. "This is my boy's health! Talk straight to me!" perspectives on accessible and culturally safe care among Aboriginal and Torres Strait Islander patients of clinical genetics services. Int J Equity Health **20**, 103 (2021). https://doi.org/10.1186/s12939-021-01443-0

19. Luke J, Dalach P, Tuer L, et al. Investigating disparity in access to Australian clinical genetic health services for Aboriginal and Torres Strait Islander people. Nat Commun. 2022;13(1):4966. Published 2022 Aug 24. doi:10.1038/s41467-022-32707-0

20. Anderson M, Elliott EJ, Zurynski YA. Australian families living with rare disease: experiences of diagnosis, health services use and needs for psychosocial support. Orphanet J Rare Dis. 2013;8:22. Published 2013 Feb 11. doi:10.1186/1750-1172-8-22

21. Industry Genomics Network Alliance (InGeNA) Delivering precision health by 2030 Outcomes of the Precision Health Roundtable held in Canberra 2022 March 2023 – https://ingena.org.au/wp content/uploads/2023/04/Precision_health_roundtable_reportv1.pdf

22. Long JC, Gaff C and Clay C. (2022). Deeble Perspectives Brief 20. Transforming the genomics workforce to sustain high value care. https://ahha.asn.au/sites/default/files/docs/policy-issue/perspectives_brief_no_20_genomics_workforce.pdf

23. Carpenter-Clawson C, Watson M, Pope A, et al. Competencies of the UK nursing and midwifery workforce to mainstream genomics in the National Health Service: the ongoing gap between perceived importance and confidence in genomics. Front Genet. 2023;14:1125599. Published 2023 Jun 16. doi:10.3389/fgene.2023.1125599

24. White S, Jacobs C, Phillips J. Mainstreaming genetics and genomics: a systematic review of the barriers and facilitators for nurses and physicians in secondary and tertiary care. Genet Med. 2020;22(7):1149-1155. doi:10.1038/s41436-020-0785-6

25. NSW Government. NSW Health (2023). Wellbeing and Health In-reach Nurse Coordinator Program. Accessed: https://www.health.nsw.gov.au/kidsfamilies/youth/Pages/whin-coordinator-program.aspx

26. Saich R, Brown R, Collicoat M, Jenner C, Primmer J, Clancy B, Holland T, Krinks S. Is Newborn Screening the Ultimate Strategy to Reduce Diagnostic Delays in Pompe Disease? The Parent and Patient Perspective. International Journal of Neonatal Screening. 2020; 6(1):1. https://doi.org/10.3390/ijns6010001

27. Ji C, Farrar MA, Norris S, Bhattacharya K, Bennetts B, Newson AJ, Healy L, Millis N, Kariyawasam DS. The Australian landscape of newborn screening in the genomics era. Rare Dis Orphan Drugs J 2023;2:26. https://dx.doi.org/10. 20517/rdodj.2023.30

28. Snow PC. Speech-Language Pathology and the Youth Offender: Epidemiological Overview and Roadmap for Future Speech-Language Pathology Research and Scope of Practice. Language, Speech and Hearing Services in Schools. 50: 324–339, April 2019. https://doi.org/10.1044/2018_LSHSS-CCJS-18-0027