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## RARE METABOLIC DISEASE WORKFORCE WHITE PAPER

# Towards a Strengthened Rare Disease Workforce for Australia

Equity Economics and Development Partners
and Rare Voices Australia

February 2022

## About Rare Voices Australia

Rare Voices Australia (RVA) is the national peak body for Australians living with a rare disease. RVA provides a strong, unified voice to advocate for policy as well as health, disability and other systems that work for people living with a rare disease. RVA is dedicated to working with all key stakeholders to drive the best outcomes for all Australians living with a rare disease through collaborative leadership for the development and implementation of rare disease policy in Australia. Key stakeholders include people living with a rare disease, governments, key peak bodies, researchers, clinicians and industry.

In 2018, the Australian Government commissioned RVA to lead the collaborative development of the National Strategic Action Plan for Rare Diseases (the Action Plan).[1] The Action Plan is the first nationally coordinated effort to address rare diseases in Australia and was informed by an extensive multi-stakeholder consultation process led by RVA. The Minister for Health launched the Action Plan in February 2020, with bipartisan support. RVA continues to work with State and Federal Governments, as well as other stakeholders, in leading the collaborative implementation of the Action Plan.

www.rarevoices.org.au

## About Equity Economics and Development Partners

Equity Economics and Development Partners (Equity Economics) is an Australian economic consultancy committed to providing quality economic analysis and policy advice to the not-for-profit, corporate and government sectors. Equity Economics is uniquely focused on addressing issues surrounding inequality, particularly through inclusive growth, equality of opportunity and stronger bilateral and multilateral relationships. Equity Economics strives to bolster development and shared prosperity in Australia and internationally.

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# Acknowledgement of Country


We acknowledge Aboriginal and Torres Strait Islander peoples as the Traditional Owners of Country throughout Australia and their continuing connection to both their land and seas. We also pay our respects to Elders – past and present – and generations of Aboriginal and Torres Strait Islander peoples now and into the future.

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*The opinions in this paper are those of the authors and do not necessarily represent the views of RVA, Equity Economics members, affiliates, individual board members or research committee members. Any remaining errors or omissions are the responsibility of the authors.*

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# EXECUTIVE SUMMARY

Australia's first nationally coordinated effort to address rare diseases—the National Strategic Action Plan for Rare Diseases (the Action Plan)[1]—called for the development of a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics. This White Paper is an initial step to progress that priority—a pilot project focusing on the workforce challenges of the rare metabolic condition workforce. Many of the findings in this pilot will be transferable to other groups of rare diseases and will inform the development of a rare disease workforce strategy more broadly.

Around 2 million Australians are living with a rare disease (approximately 8% of the population). Ensuring these Australians receive the same level and quality of care, and comparable access to lifesaving and life-improving therapies and technologies as those living with more common conditions, is an ongoing challenge.

It is prominently cited that there are more than 7,000 different rare diseases. One subset of rare diseases is those that are 'metabolic' in nature, known as rare metabolic conditions—or inborn errors of metabolism. Rare metabolic conditions are often, but not always, genetically inherited—that is, individuals are born with the disease. Taking into account the variation in how some conditions are grouped or separately defined based on their genetic origins, rare metabolic conditions include over 1,600 disorders.[2]

Rare metabolic conditions are believed to have a global prevalence of approximately 50 per 100,000 people.[3] With a population of over 25 million, Australia is expected to have an estimated 12,700 individuals living with a rare metabolic condition. This is forecast to increase by approximately 150 per year with the current rate of 300,000 births annually and new conditions being defined every year.[4]

Early diagnosis and treatment are vital in slowing down or even halting the progression of many rare metabolic conditions. Rare metabolic conditions are often diagnosed in asymptomatic newborns through newborn bloodspot screening (NBS). However, some undiagnosed patients and older patients living with a rare metabolic disease may present to the healthcare system as chronically symptomatic, well-managed, or in a state of metabolic crisis.[5]

In recent decades, advances in testing methods have allowed for greater diagnosis capacity, yet many individuals do not reach a definitive diagnosis until later in life. At the same time, new treatments have dramatically increased life expectancy for certain rare metabolic conditions, meaning more people are living into adulthood with these conditions and require ongoing treatment and care.[6]

New and emerging cell and gene therapies offer the prospect of cures for some rare metabolic conditions; however, they will require additional healthcare resources and capacity in a system already stretched in providing patients with rare metabolic conditions the health care they need.[7]

The variability and complexity of rare metabolic diseases, together with the rarity of each condition, causes overwhelming medical and social burden on families.[8] This complexity means that patients require care from a variety of specialists. Nonetheless, there is an overall bias in health and social care systems towards more common conditions. Herein lies the significant challenge for people living with a rare metabolic condition and for the healthcare workforce striving to care for these individuals. Rare metabolic conditions

are inherently disadvantaged because they are a highly heterogenous group of complex, multisystemic conditions that are poorly understood due to low patient numbers.

This White Paper is focused on better understanding the current workforce of healthcare practitioners who dedicate their time and skills to treating rare metabolic conditions across healthcare services in Australia. It looks at the composition, distribution, capacity and capability of this workforce to provide high-quality care to the estimated 12,700 Australians living with a rare metabolic condition. The White Paper brings together the voices of healthcare practitioners, patients, carers, advocates and industry to look at what is needed to strengthen the Australian rare metabolic workforce so it can meet both current and future challenges.

Overall, we found underfunding and a lack of sufficient workforce mean there is inadequate quality and consistency of care available for Australians living with rare metabolic conditions. The existing workforce is not suitably defined, recognised or resourced to achieve optimal levels of care today or into the future. This is in no way linked to the workforce's dedication and commitment to patient care, but rather inadequacies and inconsistencies in resourcing, infrastructure and current models of care.

The following 5 key findings emerged from the research and will inform ongoing strategic work led by RVA.

**KEY FINDING 1**

The role of and need for a specialised rare metabolic workforce is often poorly recognised by medical peak bodies and administrators. This both underlies *and* exacerbates other serious challenges faced by the rare metabolic workforce, patients and their families.

**KEY FINDING 2**

There is a lack of clear and formalised clinical care pathways, models of care and care plans for rare metabolic conditions.

**KEY FINDING 3**

There is significant variation in the composition and maturity of metabolic services and the workforce both *within* and *across* states and territories, as well as *within* and *across* rare metabolic conditions. This creates a 'postcode lottery' for Australians with rare metabolic conditions.

**KEY FINDING 4**

Critical workforce shortages exist and are not being addressed due to poor alignment between 'activity-based' hospital funding models and the complex, multidisciplinary care that rare metabolic patients require.

**KEY FINDING 5**

Specialised metabolic services are not sufficiently resourced to contribute to innovation. This is a barrier to the development and trialling of emerging therapies and technologies in Australia, which could offer significant benefits to patients.

# ABOUT THE RESEARCH

## Motivation

The Action Plan[1] was published in 2020 by the Australian Government Department of Health with its collaborative development led by Rare Voices Australia (RVA). The Action Plan is the first nationally coordinated effort to address rare diseases in Australia and was informed by an extensive multi-stakeholder consultation process led by RVA.

The Action Plan pointed to the systemic reforms required across Australia's health and social care systems, highlighting both the unmet patient need and gaps in rare disease management. The Action Plan builds on other strategies, both national and international, including the National Aboriginal and Torres Strait Islander Health Plan 2013–2023[9] and the Asia-Pacific Economic Cooperation (APEC) Action Plan on Rare Diseases.[10]

**THE ACTION PLAN IS CENTRED ON THREE FOUNDATION PRINCIPLES**

1. Person-centred
2. Equity of access
3. Sustainable systems and workforce

The need to develop a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics, is a priority under the Action Plan.

This White Paper focuses on a subset of the rare disease workforce that manages rare metabolic conditions. It represents an important first step towards better understanding the current workforce and building the future workforce for all rare diseases.

## Key Contributors

The development of this White Paper was possible due to the significant contributions of a range of healthcare professionals, patient groups and individuals living with rare metabolic disease and their families. Important acknowledgement must go to all those who shared their time and information, data, views and/or experiences.

### Rare Voices Australia

Rare Voices Australia (RVA) engaged Equity Economics and Development Partners (Equity Economics) to undertake a comprehensive review and analysis of the Australian rare metabolic workforce and develop a White Paper to help move Australia towards a strengthened and sustainable future rare disease workforce.

The collaborative relationship between RVA and Equity Economics has been a key strength of this process. RVA's respected position within the rare disease space and existing relationships and networks contributed to both the reach and quality of the research process.

### Project Steering Committee

Development of this White Paper was guided by a Project Steering Committee comprised of the specialised metabolic workforce across Australia, including representation from members of the Australasian Society for Inborn Errors of Metabolism (ASIEM), who bring together the different healthcare practitioners involved in managing and diagnosing individuals with rare metabolic conditions. The knowledge and expertise of the Project Steering Committee, and the use of their peer and patient networks, was critical to defining the scope of this research and ultimately the realisation of key findings and strategic direction for this document.

## Methods

### Literature Review

In consultation with the Project Steering Committee, a literature review was carried out in May 2021 to understand workforce trends and directions internationally and help define the scope of the research methodology for this study of the Australian metabolic workforce.

### National Metabolic Workforce Questionnaire

A questionnaire with 55 questions was distributed to key diagnostic and specialist workforce professionals between August and September 2021. Individual healthcare practitioners, services and some major hospitals from all jurisdictions were invited to respond to the questionnaire. Targeted follow-ups were undertaken to collect data from every jurisdiction. The questionnaire was open for three weeks and intended to capture information at the service level, so recipients were encouraged to collaborate with their teams to provide a single response.

The questionnaire captured the following types of information:

- Service area;
- Number of services and service types (including telehealth and outpatient clinics), and number of diagnoses for the period 2018–2021 amongst infant, paediatric and adult patient categories;
- Access to diagnostic equipment/testing;
- Current staff profile of specialised metabolic roles;
- Strengths of the services and challenges faced (self-reported); and
- Available resourcing to meet current and future needs.

Thirteen different clinical and diagnostic services across New South Wales (NSW), Victoria (VIC), Queensland (QLD) and South Australia (SA) responded to the questionnaire. The data was used to understand the distribution and make-up of the rare metabolic workforce across Australia, available treatment pathways, as well as to draw out strengths and pressure points.

## National Patient and Carer Experience Survey

A Patient and Carer Experience Survey was disseminated by RVA and members of the Project Steering Committee to patients, carers and families living with rare metabolic conditions (Figure 1). RVA distributed the survey via their existing network of relationships with advocacy, support and representation groups for individual rare metabolic conditions. This was critical to the reach of the survey.

**PATIENT AND CARER SURVEY**

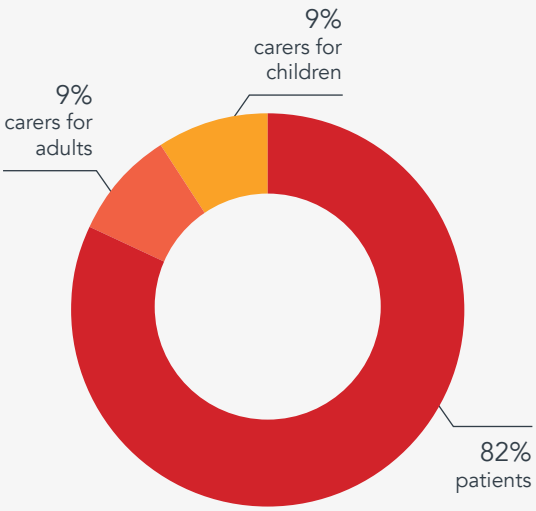
**310** patients and carers responded

**60** different rare diseases were represented

**70** patients reported being diagnosed with phenylketonuria

**29** rare diseases only had one respondent

**FIGURE 1: RESPONDENT BREAKDOWN**




The survey sought to understand where and how patients were accessing diagnosis, treatment and management services for their rare metabolic condition, as well as which health professionals they were engaging with. It picked up on interfaces with the National Disability Insurance Service (NDIS) and My Aged Care, and how service provision may have changed as a result of the COVID-19 pandemic and rise of telehealth. Respondents were also given the opportunity to reflect on how they would improve care for metabolic conditions in Australia.

Through the survey dissemination process, we found that many patients or carers do not necessarily identify as having a 'rare metabolic condition' and that this sort of language is not well understood amongst patients.

## Industry Survey – Rare Voices Australia's Round Table of Companies

A small industry snapshot was taken from a group of industry representatives with an interest in metabolic disease therapies and health technologies. Seven organisations represented on RVA's Round Table of Companies that have therapies or health technologies approved or in the pipeline for the treatment of rare metabolic diseases responded to a short, anonymous survey.

The survey sought views from industry on the current capacity of Australian metabolic services around:

- Clinical management of emerging therapies or health technologies; and
- Facilitating clinical trials for emerging therapies or health technologies.

It also sought to understand what affects each organisations' abilities to develop, produce or trial new therapies or health technologies, as well as assess the effect of the COVID-19 pandemic on clinical trials, clinical management and/or access to current therapies.

### Workforce Focus Groups

A series of three 1-hour virtual focus group sessions were held with metabolic workforce representation from across Australia. These followed the distribution of a discussion paper that set out early findings of the study and offered themes to guide conversation.

Each group had a mix of approximately 5 to 8 different healthcare practitioners from across NSW, VIC, QLD and SA, including metabolic physicians, metabolic dietitians, metabolic nurses and nurse practitioners, biochemical geneticists and scientists. Workforce from all jurisdictions were invited to participate in the sessions.

The sessions were recorded and observations and quotes from the discussions used as qualitative data. This was particularly useful in understanding the cross-jurisdictional relationships between states and territories that had not participated in the National Metabolic Workforce Questionnaire (Northern Territory (NT), Tasmania (TAS), Australian Capital Territory (ACT) and Western Australia (WA)).

### Patient Group Leader Focus Groups

A single 1-hour virtual focus group session was held with 8 leaders or nominated individuals from different rare metabolic disease support groups from across Australia who responded to the call to participate. These followed the distribution of a patient/carer focused discussion paper, which set out early findings of the study and offered some themes to guide conversation. Leaders or nominated individuals were encouraged to share this within their groups to gain collective views and anecdotal experiences to bring to the session. The session was recorded and observations and quotes from the discussions were used as qualitative data.

### Written Submissions

Some individual members of the metabolic workforce, patients and carers, together with support group leaders provided written submissions via email. These emails usually contained additional views, not collected through the questionnaire, survey or focus groups, or pointed to existing literature or studies. Written submissions became part of the qualitative dataset.

### Input from International Metabolic Experts

Virtual meetings and email correspondence with overseas metabolic clinicians and others from the metabolic specialty facilitated the gathering of information around international practices in metabolic medicine. Information gathered contributed to the international best practice section of this White Paper. Overseas data was collected around the gaps and strengths of metabolic specialist services, the types of specialists involved in metabolic care, full-time equivalents for each role, the number of patients looked after and the approach to care.

# RARE METABOLIC DISEASE CARE IN AUSTRALIA: WHO IS INVOLVED?

Newborn screening has been identifying some rare metabolic conditions in Australia since 1966, when testing for phenylketonuria began. Since then, the number of conditions screened for through the newborn bloodspot screening (NBS) program has grown, as has the number of known rare metabolic conditions. As a result, diagnostic and clinical aspects of patient management have emerged together. Today, there remains a strong linkage between diagnostic and clinical services for rare diseases in Australia, including for rare metabolic diseases.

In Australia, like other countries, a multidisciplinary approach has been established as necessary for caring for metabolic patients. However, there is significant variation in the scale and make-up of services available across states and territories. The Australian rare metabolic workforce faces the inescapable geographical challenge of distributing and delivering health services to a dispersed population and small numbers of individuals living with rare metabolic disease.

## Metabolic Healthcare Practitioners

### Medical Doctors

Metabolic physicians have two key roles: 1) supporting other key healthcare practitioners such as dietitians, and 2) providing treatment and ongoing management to patients.[5] In Australia, accreditation of genetic metabolic physicians occurs through the Royal Australasian College of Physicians (RACP) Genetics Services Advisory Committee (GSAC). Infants, children and their families may engage most frequently with general paediatricians when navigating the early stages of rare disease diagnosis and management.[8] There are a number of metabolic physicians in Australia who have not been accredited by the GSAC but are still able to provide care.

### Dietitians

Specialist metabolic dietitians are vital to the management of many rare metabolic diseases, which respond to highly specialised diets. These diets require constant review and modification, based on nutritional requirements for age and metabolic control.[11]

### Nurses, Nurse Coordinators and Nurse Practitioners

Nurses with additional metabolic training have been consistently shown to be an important part of the metabolic workforce.[12] They play an essential role in communicating with, educating and supporting patients and families, as well as facilitating coordinated care through clinics and liaising with other specialists.

### Genetic Healthcare Practitioners: Clinical Geneticists and Genetic Counsellors

Genetic clinics often involve clinical geneticists, genetic counsellors, administrative staff and other support staff such as data managers.[13,14]

Clinical geneticists are a group of medical doctors with specific genetics training. In Australia, these doctors can become further subspecialists in inborn errors of metabolism (IEM) through additional metabolic genetics training under the GSAC.[13]

Clinical geneticists work closely with genetic counsellors. Genetic counsellors are allied health professionals with training in genetics and counselling, who communicate with patients and their families to help them adapt to genetic diagnoses.[15]

## Diagnostic Professionals

### Genetic Scientists

The work of healthcare practitioners is actively supported by laboratory staff, including cytogenetic and laboratory scientists. For an inherited metabolic diagnosis to be made, testing must occur.

### Biochemical Genetics Scientists

There are 6 laboratories in Australia that perform specialised biochemical testing to diagnose metabolic conditions. Many of the tests are not routinely available elsewhere and are not on the Medicare Benefits Schedule (MBS). Each laboratory works in close collaboration with metabolic teams.

### Newborn Screening Scientists

NBS tests are offered to every newborn baby in Australia. There are 5 laboratories in Australia that conduct NBS for babies across all jurisdictions. The newborn screening services work in close collaboration with Genetic Pathology and metabolic teams.

## Australasian Society for Inborn Errors of Metabolism

The Australasian Society for Inborn Errors of Metabolism (ASIEM) is a special interest group of the Human Genetics Society of Australasia (HGSA), which started in 1992. It aims to bring together different healthcare practitioners involved in managing and diagnosing individuals with rare metabolic conditions, including laboratory scientists, metabolic physicians, nurses and dietitians. ASIEM also play a vital role in educating other healthcare practitioners about inherited metabolic diseases, and have developed national guidelines, policies and patient information. Additionally, they provide expert advice to other organisations on behalf of the HGSA.

Significantly, ASIEM have created a series of national professional support sub-groups led by the rare metabolic workforce—including one for clinicians and one for dietitians—to share information, foster relationships and support those who are new to the metabolic space.

## Peak Advocacy, Support and Representation Groups

Rare Voices Australia (RVA) is the national peak body for Australians living with a rare disease. RVA partners with rare metabolic disease support groups to help strengthen and expand their influence. RVA and specific rare metabolic disease support groups have a role to play in the rare metabolic disease workforce. Australian rare metabolic disease support groups are involved in some or all of the following activities:

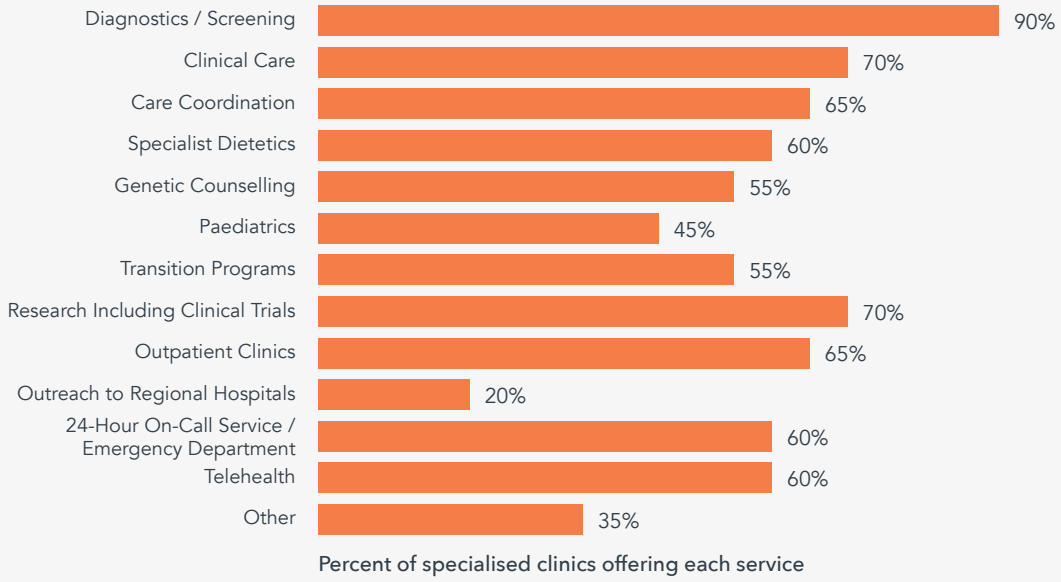
- Partnerships (state, national, international);
- Management of rare disease data and registries;
- Awareness/social/educational events, including organising conferences;
- Prevention and support advocacy;
- Facilitating communication between policymakers and the rare disease community;
- Care and support services;
- Transition services (including palliative care);
- Facilitating access to diagnostic and expert care;
- Offering support throughout patient and family journeys;
- Advocacy for equitable access; and
- Driving research.

# CURRENT SCOPE OF METABOLIC SERVICES IN AUSTRALIA

The National Metabolic Workforce Questionnaire surveyed 13 specialised metabolic clinical and diagnostic services across Australia, providing insights into the comprehensive nature of services offered. We received responses from most major services across NSW, QLD, VIC, and SA, which provide a range of specialist services (Figure 2).

More detail on these individual services is provided in the 'States and Territories: Establishing Jurisdictional Baselines' section below.

**FIGURE 2: SERVICES OFFERED BY SPECIALISED METABOLIC CLINICS IN AUSTRALIA**




*'Other' signifies services nominated by workforce respondents including education and training; biochemical genetics; pregnancy/maternity care; consultative advice or clinical support to other services; and social work support.*

## Specialised Metabolic Clinical Services

### Staffing

All specialised metabolic clinical services in Australia are small units within large tertiary hospitals.

The care team is usually very small and typically comprises 1-3 full-time equivalent (FTE) specialists, 1-2 FTE dietitians and 1-2 FTE metabolic nurse roles.

Across the 5 major centres surveyed, there were 3,900 patients with rare metabolic conditions receiving specialist care. Across these centres, there were a total of 11.3 FTE metabolic clinicians, 8.9 FTE specialist nurses, 7.3 FTE specialist dietitians and 6.5 FTE specialist social workers.

### Geographic Coverage

All specialised metabolic clinical services that engaged in our research (from NSW, VIC, QLD and SA) provide services to patients in another state or territory, in addition to their own jurisdiction.

NSW, QLD and SA do have formal agreements for specific elements of the metabolic service, for example NBS. However, of all the specialist metabolic services providing support to other jurisdictions, only VIC services have an overarching formalised agreement to provide metabolic care more comprehensively.

## Specialised Metabolic Diagnostic Services

Specialised metabolic clinical services are strengthened by close working collaborative relationships with specialised diagnostic services. Historically, clinical services grew from tertiary laboratory services, and there is a strong representation of disorders that the labs diagnose in the clinical caseload. This interface with laboratories remains crucial.

### Responsiveness

All diagnostic facilities testing for rare metabolic conditions undertake these tests daily or at least weekly. Testing turnaround time varies significantly based on the kind of test – from 2 hours (for routine tests in urgent cases) to 5-6 months for more complex testing performed infrequently.

### Location

In NSW, VIC, QLD and SA, all metabolic clinical services reported to be either co-located within the same facility as a specialised diagnostic service, or to have access to diagnostic testing or laboratory services within the same local area/city.

### Geographic Coverage

75% of specialised diagnostic services provide their services to another state/territory. Of this 75%, there was an even proportion of services provided under formalised arrangements, no arrangement or a mix of informal and formal arrangements.

# Living with a Rare Metabolic Condition

## Sally's* Story: Severe Methylenetetrahydro-folate Reductase Deficiency

### Long-term survival of acute life-threatening conditions comes with complex care needs

### Understanding the Condition

MTHFR stands for *Methylenetetrahydro-folate reductase*. *Homocystinuria* (increased amounts of homocysteine in the blood and urine) is a rare metabolic condition that results from poor metabolism of folate, due to complete lack of a working enzyme called MTHFR. The gene that tells our body how to make the enzyme is also called MTHFR.[16]

### Introducing Sally and John

*John** is a parent to *Sally*. Sally is 13-years-old and one of three surviving children in Australia with this condition. Sally also has hydrocephalus (a condition caused by fluid build-up in the brain) as a result of her MTHFR deficiency. Sally was diagnosed with MTHFR deficiency at 4 months of age after multiple tests, including skin biopsies and a blood test for homocystinuria.

### Life for Sally

Life for Sally is challenging. Sally's MTHFR deficiency affects her mental and social capabilities, has delayed her development and means even simple daily tasks can be difficult. With mild intellectual disability, Sally has difficulty learning and socialising with others. She comes across as emotional, so her decision-making is often impaired by emotional stress. Sally is bullied and often feels rejected. She is now also showing signs of anxiety and depression and is often fatigued.

### Caring for Sally

Coming to terms with Sally's diagnosis was hard for John and the rest of the family. As the condition is so rare, John undertook his own literature review to understand what his daughter was facing. Initially, he says it was difficult, but with time the family have accepted and tried to adjust.

John says the family spends a lot of their time and attention on Sally—in particular, keeping up with her therapy needs. Sally attends weekly physiotherapy and psychiatry appointments and, prior to COVID-19, she was also attending swimming sessions as part of her therapy schedule. She also regularly sees her general practitioner for check-ups.

With John working full-time and Sally's mother caring for other children, time management is a challenge. Sally's care is led by the metabolic team at Westmead Children's Hospital in Sydney, and she meets with them twice a year. Other specialities involved in her care team include a neurologist and an ophthalmologist. John is grateful to the metabolic team for leading Sally's care and connecting them with all the other health professionals in her care team. They have supported the family through Sally's development from infancy, and now into her young teenage years. John says the family still has challenges, including convincing Sally to take her medication, but can cope thanks to the help that's available.

**Pseudonyms were used to maintain patient privacy*

# KEY FINDINGS – BUILDING A NATIONAL PICTURE

The aim of this White Paper is to build a national picture of the current rare metabolic workforce, including strengths and weaknesses, to inform further work towards a sustainable workforce today and in the future.

In this section, we outline the 5 key findings from our research, drawing on findings from a literature review; a survey of patients with rare metabolic conditions; a workforce questionnaire of major specialist metabolic service providers; an industry survey; and 4 in-depth focus groups with patients and clinicians.

**KEY FINDING 1**

The role of and need for a specialised rare metabolic workforce is often poorly recognised by medical peak bodies and administrators. This both underlies *and* exacerbates other serious challenges faced by the rare metabolic workforce, patients and their families.

## Who Provides Care for People with Rare Metabolic Conditions?

The healthcare system is geared towards treating and caring for people with more common conditions, creating an inherent bias against those with rare conditions. Rare metabolic diseases are a highly heterogenous group of complex multisystemic conditions, each only affecting a small number of people. While they can be grouped together by their underlying cause, they differ in presentation and healthcare treatment needs.

Together, this combination of diversity, complexity and rarity has created significant gaps in the resources and treatments available to those living with a rare metabolic disease when compared with other conditions. For example, continuous glucose monitoring and ketone measurement treatments are subsidised for the most common and widely known of all metabolic diseases—diabetes—but not for patients with far less prevalent metabolic conditions who may also require a ketogenic diet or suffer recurrent hypoglycaemia.

The need for timely and accurate diagnosis and better integrated care to improve quality of life and extend life expectancy for those with rare metabolic conditions is well-evidenced.[12] However, inherent challenges in appropriately understanding, defining and treating rare metabolic diseases are responsible for an emerging, yet underdeveloped, specialised rare metabolic workforce in Australia, which continues to perpetuate slower than needed progress.

As rare metabolic conditions include a spectrum of diseases, some are better defined and understood than others.[17, 18] However, the lack of clear national criteria for what constitutes a rare metabolic condition, and the absence of guidelines for which conditions should be under specialist metabolic care, mean there are significant variations and inconsistencies in the types and mix of care received by patients living with the same rare metabolic conditions across Australia. This is the result of largely cobbled together, patchwork and informal care arrangements.

Historically, patients with rare metabolic conditions received care from specialists based on their symptoms, and not the underlying cause of their condition (which was often unknown). Engagement with the Project Steering Committee and metabolic workforce through focus groups identified a wide spectrum of medical specialist areas involved in the care of a person with a rare metabolic condition, including clinical genetics, neurology, cardiology, nephrology and endocrinology.

Even in an international context, metabolic medicine (adult metabolic medicine in particular) is a comparatively new sub-specialist area.[19] This is due to the relatively recent emergence of metabolic medicine, and because the complex and multisystem presentation of most rare metabolic conditions creates significant crossover with other, more established specialised areas of medicine. This overlap is important because multidisciplinary care that includes one or more of these specialities is often needed for a person with a rare metabolic condition, based on their unique diagnosis and clinical presentation. The lack of a clear classification system for rare metabolic conditions in Australia contributes to patients not always being managed by a metabolic specialist. Because metabolic conditions often involve multiple organ systems, a non-metabolic specialist, such as an orthopaedic surgeon, might be in a position to care for a particular patient; however, they may not consider specific issues like airway dysfunction in patients with mucopolysaccharidosis.[20] Unfortunately, fatalities have occurred because of poor underlying disease awareness.

## Fragmented Care Lacking Specialist Metabolic Oversight

There were many examples raised during the focus groups where haematologists, neurologists, nephrologists and endocrinologists are heavily involved in a patient's diagnostic journey, particularly where the condition has symptoms concentrated within a particular body system. These specialists continue to manage the patient beyond their diagnosis with a rare metabolic disease.

For example:

- Care for patients with *Cystinosis*, a rare genetic lysosomal storage disorder that results in the abnormal accumulation of the amino acid cystine in the body's organs, is often led by a nephrologist, because the organ most significantly impacted by the disease is usually the kidneys; and
- Care for patients with *Gaucher disease*, a rare genetic disorder caused by deficiency of the enzyme *glucocerebrosidase*, is often led by a haematologist because, although it causes disruptions in several cell types, the most significant affect is a reduction in blood cell counts.

In some cases, the lack of management and leadership from a metabolic specialist means patients are not receiving optimal multidisciplinary care for their condition. This can have very real impacts on disease progression.

Leadership from another specialty is absolutely preferable to a patient not being connected to any specialist care at all. Particularly in some jurisdictions where the appropriate specialised metabolic workforce does not exist or does not have capacity to support metabolic patient care.

In some cases, non-metabolic clinicians start to organically attract new referrals for patients with the same specific rare metabolic diseases, because they have built up expertise and familiarity. Word-of-mouth of a specialty services' experience or reputation become important where there is an absence of formalised referral pathways. There are a number of examples where individual clinicians with a special interest in a specific condition begin to take on a regional or even national leadership role for that condition.

This is the case for Fabry disease, a lysosomal storage disease affecting 300 Australians. Despite not having a specialised metabolic service or centre, a leading Western Australian nephrologist has become recognised as delivering specialist care for patients with Fabry disease and runs a dedicated clinic.

Personal professional interest from non-metabolic clinicians can lead to the establishment of a specialist dedicated clinic for a particular condition that delivers high quality care for those patients. This model is not usually formalised and is reliant on individual clinicians who may move on or retire, jeopardising continuity of care. The lack of sustainability around this type of service, together with the lack of systemic funding and planning for metabolic services, means many Australians with rare metabolic conditions are missing out on the care they require. This is especially true in states and territories without a dedicated metabolic service.

## Lack of Training Pathways

A major issue stemming from the relatively recent emergence of metabolic medicine as a specialty area is that it is not yet supported by the appropriate training pathways that other specialities benefit from. This has a huge impact on ensuring there is a sustainable pipeline of suitably trained and experienced medical specialists to meet current and future demand. This does not only apply to medical training pathways, but also to allied health professionals, including specialist metabolic dietitians and genetic counsellors. Therefore, increasing capacity for formal training is needed to ensure there is sufficient workforce expertise to support patients with rare metabolic conditions into the future.

## Views of Stakeholders

### Specialist Metabolic Care

*"I felt really supported having a place where someone fully understood my condition. Most doctors have never heard of it and don't understand it. I was recently pregnant and had a baby, and there wasn't much research on someone with my condition going through childbirth. But there was a plan put in place by the specialist, which made me feel at ease that I could have my baby at a hospital close to my home and they would be fully briefed on all of my needs."*

– Patient with Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency, Patient and Carer Experience Survey

*"Our metabolic team is brilliant. We are making it up as we go as [our son] is so rare and having been in a drug trial, and now with the drug, we all just do the best we can. Metabolics [our metabolic team] lead the way. I call them our 'project managers'. We have 16 or more teams at Children's Hospital Westmead, and they are the 'managers'."*

– Carer, Patient and Carer Experience Survey

### Fragmented Care Lacking Specialist Metabolic Oversight

*"We hear from families of children… and also adults with Mito[chondrial disease] that they often wait months and years to access specialist clinics. In other cases, they see the 'wrong' specialist for years before their case is brought to the attention of a specialist with Mito knowledge and experience. Often this specialist is a metabolic clinician. The impacts of these delays often mean a delay to a definitive diagnosis, delays in surveillance for emerging symptoms and delays to access available treatment and management. We know this impacts quality of life and speed of progression of the condition."*

– Mito Foundation Representative, Written Submission

### Training Pathways

*"A training pathway doesn't exist. We [adult metabolic clinicians] are currently considered under clinical genetics, but it isn't really [accurate to call us this]. We aren't really on the radar of the [Royal Australian] College of Physicians."*

– Adult Metabolic Physician, Focus Group

*"The metabolic workforce would definitely benefit from clearer delineation from other specialty areas. It would be good for the RACP [Royal Australasian College of Physicians] to make metabolic medicine a specialty in its own right, as in other countries. At the moment, 'metabolic medicine' comes under clinical genetics, and there is no set curriculum specific for metabolic trainees. This means that metabolic trainees may not all be at the same standard upon finishing training. Currently, we are considered clinical geneticists. However, in many centres, we have difficulty in ordering genetic testing, and still need to run this past the clinical genetics team. We could build this understanding by encouraging the RACP to make metabolic medicine its own specialty, and by having a defined curriculum. Trainees should also be required to spend time training in other metabolic centres to broaden their learning and knowledge base."*

– Metabolic Consultant Physician, Written Submission

## Lack of Recognition and Definition of Specialist Metabolic Roles

Many people are diagnosed with a rare metabolic disease during childhood, with a number of conditions now picked up via the NBS program shortly after birth. In general, metabolic disorders detected through NBS are more easily defined and broadly accepted as belonging to the umbrella of rare metabolic conditions, compared with those diagnosed later in life.[21] In addition to this more streamlined entry point into care, paediatric metabolic medicine is generally more defined in scope and, as a result, it is more recognised and established as a specialty area, both in Australia and overseas, compared with adult metabolic medicine.

Adult care has its own specific challenges, yet the role of the adult metabolic workforce is not sufficiently understood. Furthermore, there are fewer and less developed adult services than paediatric services. In some cases, adult patients continue to access paediatric services into adulthood because adult services do not exist, or where they do, they are not adequately experienced or resourced to manage their particular rare metabolic condition. This is further exacerbated by the lack of formal recognition and training pathways for adult metabolic clinicians.

The rare metabolic workforce is further stretched as *complex non-metabolic* patients are often managed by metabolic teams because a more appropriate specialty 'home' cannot be identified. This occurs because the metabolic team is usually well-versed in creating bespoke patient-centred care for complex multisystem rare diseases, with a sparse literature and support base. In some cases, metabolic specialist teams are having to share resources with other clinical specialities.

> An in-practice example of this crossover of specialty areas can be seen at the Royal Adelaide Hospital in South Australia where funding for Clinical Services Coordination is being shared across the endocrinology and metabolic units, despite the strong need for a dedicated metabolic position.

In tertiary hospital settings where a specialised metabolic department does exist, there needs to be both clarity of roles and strong linkages to support multidisciplinary care with other specialty areas. This is critical to ensure patients receive the right mix of care, and that their care is appropriately led.

## Delays in Diagnosis

Specialist metabolic diagnostic expertise is necessary for timely diagnosis of rare metabolic conditions. Many rare metabolic conditions are progressive and life limiting, so diagnosis is time critical.[1] Delays in diagnosis and misdiagnoses prevent access to the best clinical care and treatment options, including access to participation in clinical trials.

Delays in diagnosis, coined the diagnostic odyssey (the time between presenting with symptoms to the time of a definitive diagnosis), are exacerbated by the lack of a suitably defined and recognised specialised metabolic workforce with specific knowledge of the vast array of rare metabolic conditions. Late or delayed diagnosis is often caused by challenges faced by healthcare professionals without the expertise to piece together an often-confusing array of multisystemic symptoms.

While the NBS program is an important pathway to diagnosis for some rare metabolic conditions, the journey to a definitive diagnosis is not so straightforward for many Australians; with some not receiving a diagnosis until well into their adult years (Figures 3 and 4).

**FIGURE 3: AGE AT DIAGNOSIS REPORTED BY RESPONDENTS IN THE PATIENT AND CARER SURVEY**

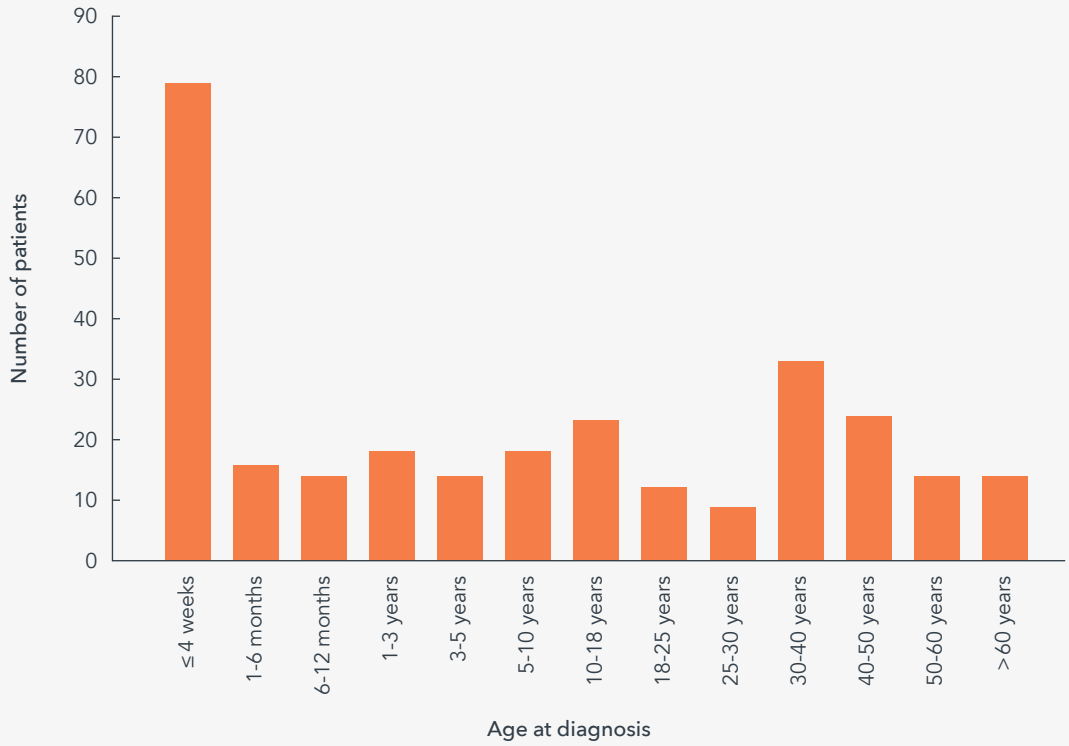
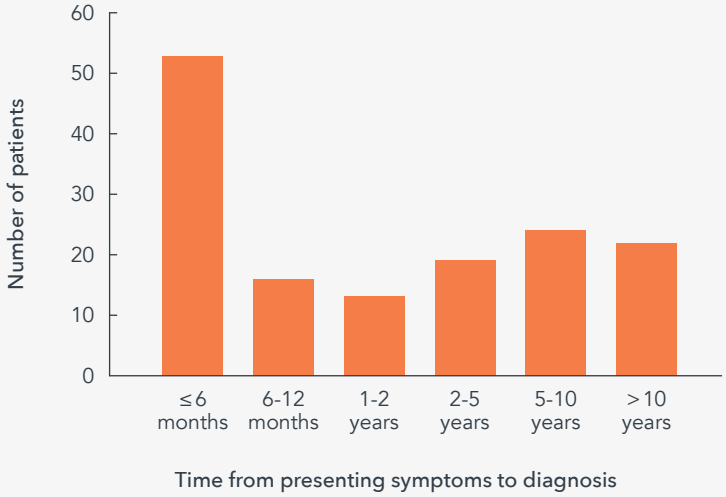



**FIGURE 4: DIAGNOSTIC DELAY REPORTED BY RESPONDENTS IN THE PATIENT AND CARER SURVEY**



**KEY FINDING 2**

There is a lack of clear and formalised clinical care pathways, models of care and care plans for rare metabolic conditions.

## Need for Multidisciplinary Teams

Rare conditions are inherently multi-systemic, with patients requiring complex, multidisciplinary care. Studies have emphasised the importance of a multidisciplinary team (MDT) approach including medical doctors, geneticists and genetic counsellors, researchers, nurses, dietitians and laboratory staff.[12] MDTs are vital in ensuring experts can work together to help their patients.

The need for multidisciplinary, coordinated and integrated care for the complex management of rare metabolic conditions came through strongly in our research across respondents in the workforce, patients, carers and industry. In particular, the lack of formal arrangements or systems to support this type of best-practice care was repeatedly raised by participants as a key issue requiring action.

All specialised metabolic clinical and diagnostic services report a high level of multidisciplinary involvement in care for patients with rare metabolic disease. These services named a total of **36** different health professionals/services that they were engaging with, at least monthly, *outside of their own service*.

The following were the most common external services/professions engaged with on a monthly basis:

- Paediatricians
- Clinical geneticists
- Diagnostic/laboratory scientists
- Dietitians
- General practitioners (GPs)
- Emergency department doctors
- Clinical trial researchers
- Neurologists
- Neonatologists
- Gastroenterologists

Patients also reported a range of healthcare professionals and supports, other than metabolic specialists, that they engaged with frequently. Figures 5 and 6 show the top 10 healthcare professionals and supports most frequently seen by adults (over 18 years) and children (under 18 years).

**FIGURE 5: TOP 10 HEALTHCARE PROFESSIONALS OR SUPPORTS MOST FREQUENTLY VISITED BY PAEDIATRIC METABOLIC PATIENTS (<18 YEARS) IN THE 12 MONTHS PRIOR TO AUGUST 2021**

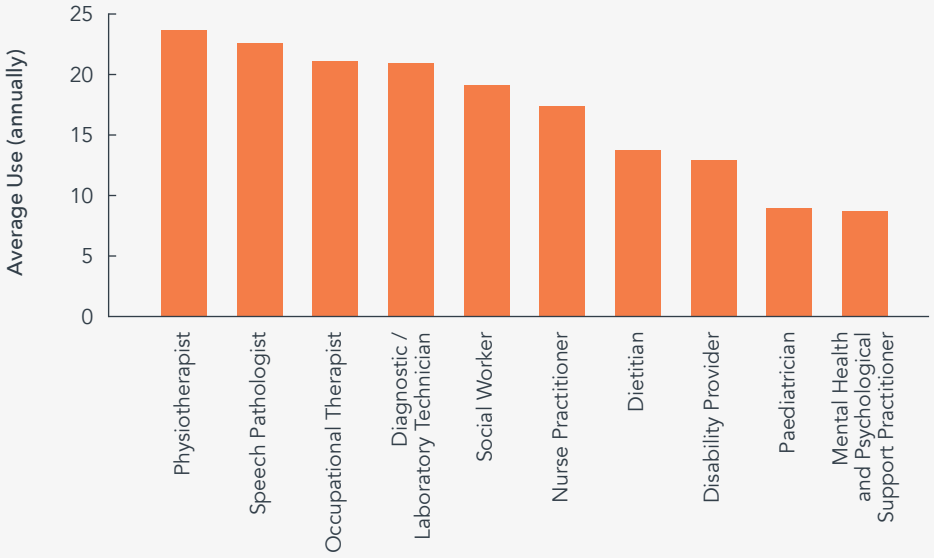
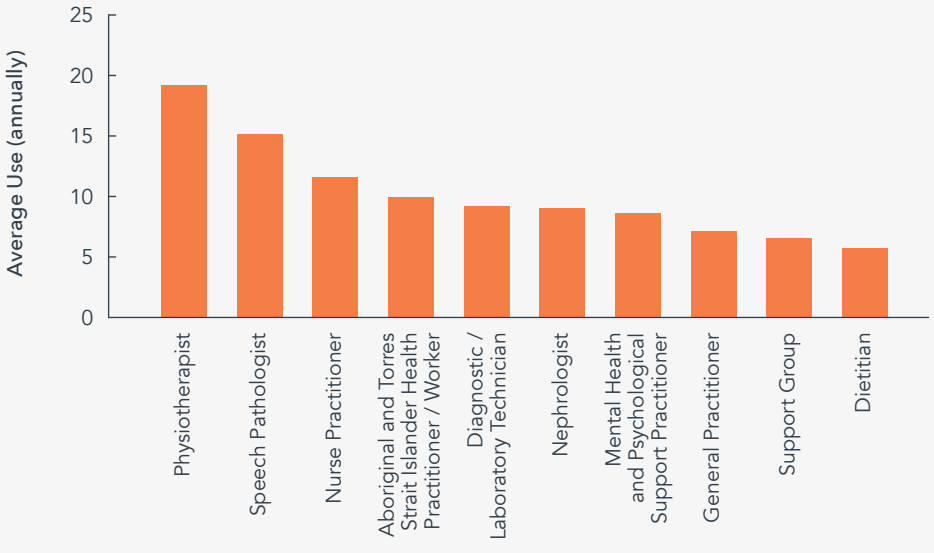


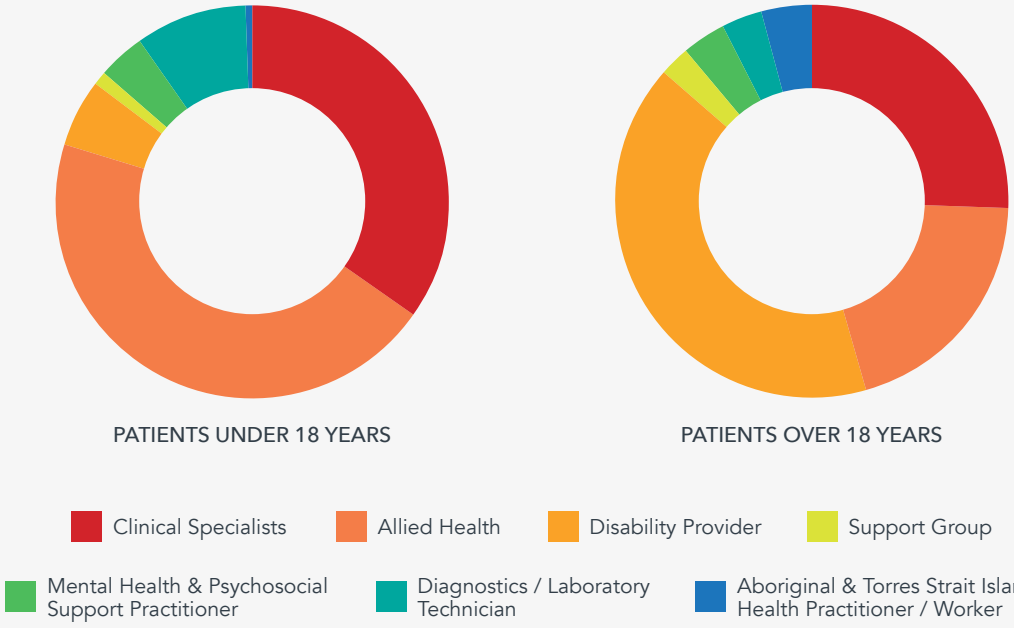

**FIGURE 6: TOP 10 HEALTHCARE PROFESSIONALS OR SUPPORTS MOST FREQUENTLY VISITED BY ADULT METABOLIC PATIENTS (>18 YEARS) IN THE 12 MONTHS PRIOR TO AUGUST 2021***




**This graph excludes disability providers, as the average frequency of access is skewed by some patients who access disability providers on a daily basis.*

Figure 7 shows the proportion of each service type accessed by adults and children over a 12 month period. Clinical specialists make up a significant portion of service types accessed by both adults and paediatric metabolic patients. Allied health is accessed most often by children compared to adults, whereas disability services become the most required service type for adult metabolic patients.

**FIGURE 7: SERVICE TYPES ACCESSED* IN THE 12 MONTHS PRIOR TO AUGUST 2021**




**Clinical specialists include: paediatricians; clinical geneticists; clinical trial researchers; nurse practitioners; endocrinologists; neurologists; immunologists; haematologists; cardiologists; ophthalmologists; obstetricians; gastroenterologists; nephrologists; rheumatologists; orthopaedic specialists; respiratory specialists; dermatologists; dentists; general practitioners; and emergency department doctors. Allied health include: dietitians; genetic counsellors; speech pathologists; and occupational therapists.*

## Lack of Care Coordination and Standard Models of Care

Only **40%** of the specialist metabolic services surveyed (both diagnostic and clinical) have a lead position responsible for care coordination. Even in these cases, information provided through the National Metabolic Workforce Questionnaire and the Workforce Focus Groups suggests in many instances these designated lead positions are not a separately funded, discrete function in most services. Instead, care coordination is being performed by someone in the care team in addition to their clinical or diagnostic role. Of the remaining services that did not identify a lead care coordination position, one third indicated a collaborative, multidisciplinary approach to care coordination was still occurring within their service.

The absence of formalised and established models of care and care pathways for many rare metabolic conditions means that delivering the right specialist care—and coordinated care from across services/disciplines—is too often reliant on clinicians' awareness of the right services to connect patients with, as well as on good communication, and even goodwill.

The use of individualised care plans that clearly outline a patient's treatment or management goals and delineate the role and function of each member of their care team, are widely accepted as best-practice for complex and chronic conditions. However, individualised care plans are not routinely created for many patients living with rare metabolic diseases. In rare instances where they are created, their use is not being maximised by the patient's treatment or management team.

| MODEL OF CARE | CLINICAL CARE PATHWAYS | INDIVIDUALISED CARE PLANS | WHOLE-OF-LIFE MODELS OF CARE |
|---|---|---|---|
| Broadly defines the way health services are delivered. It outlines best practice care and services for a person, population group, or patient cohort as they progress through the stages of a condition. | Standardised, evidence-based multidisciplinary management plans, which identify an appropriate sequence of clinical interventions, timeframes, milestones and expected outcomes for a patient group. | A patient-centred health record that can be shared by many members of a care team, including the patient and their carer. Individualised care plans help improve relationships, understanding and efficiency. | Recognise the key transition stages across the life course of patients accessing care. For example, the Lifespan Model. |

# Service Model: Lifespan

## The 'Lifespan' Approach

A lifespan approach is a 'family-centred' approach to care. This is particularly important for rare metabolic diseases as many are inherited. Specialised metabolic staff work *across the lifespan*, supporting infants and children, young people, adults and families through the care journey. This approach minimises transition from paediatric to adult care clinics and the well-documented risk of 'losing' patients during transition. Parents feel reassured that their young adult will receive the same level of care throughout their life course.

This larger combined service approach, with pooled resources and staff, is also a cheaper model of care. It supports training and professional development, information-sharing, and it reduces the risk of staff-burnout with shared on-call duties. It also ensures patient safety and consistency of care, regardless of where they are placed in the service.

## How it Works at the Queensland Lifespan Metabolic Service

### Partnership between the Queensland Children's Hospital and Mater Hospital, Brisbane, Australia

Lifespan is a 'one-stop-shop' for metabolic care in Queensland (QLD). There is just one phone number to call for anyone in QLD seeking metabolic care. Staffing consists of metabolic clinicians, metabolic nurses, nurse practitioners, metabolic dietitians, metabolic social workers and psychologists.

The service runs multiple weekly clinics out of the Queensland Children's Hospital (QCH) for paediatric patients and the Mater Hospital for adult patients, which are conveniently co-located side-by-side.

While some roles are specialised in paediatric or adult care, the service works as one big team, with daily clinical handovers to all team members. Clinical practice is audited weekly—this means all patients seen each week are subject to a team review, to ensure optimal care planning and support the best clinical outcomes.

There is a formalised partnership between the diagnostic service and systems (such as IT) to support regular communication between scientific and clinical staff. Also, diagnostic service staff attend weekly education meetings with the clinical team.

The service produces multiple case publications and conference abstracts (national and international) each year. Both services are engaged in paediatric and adult research and clinical trials. Research projects around phase I and II clinical trials, clinical research, and quality improvement processes are streamlined within the clinical service; these are managed by a metabolic physician lead who coordinates research. The lifespan service also has a clinical research manager who facilitates Human Resource Ethics Committee and Governance applications and oversees the running of clinical trials and the distribution/publication of clinical trial results.

### How the Service Came About

The current Director of the QLD Lifespan Metabolic Service, Anita Inwood (BNurs, GradDipPaeds, MNP), won a Churchill Fellowship and, in 2013, travelled to the United Kingdom (UK) to explore the lifespan approach. While in the UK, Anita observed lifespan models/partnerships in place between the Royal Manchester Hospital and Royal Salford Hospital in Manchester, and between Evelina Children's Hospital and St Thomas' in London. Anita came into QLD metabolic services as a metabolic nurse 18 years ago, and the lifespan approach was her vision for QLD, along with Dr Jim McGill, the previous Director of the service. Finally, in 2015, a partnership was formed between two QLD Hospital and Health Services (HHS)—QCH and Mater Hospital. Initial seed funding helped set up and run the service for the first 12 to 18 months. Since then, the service has continued in good faith and comes with no additional funding, despite attempts to get a service level agreement approved. The service is at risk of being dismantled if either HHS choose to withdraw their support for this model of care, leaving no adult metabolic services in QLD.

### Looking to the Future

Need for the lifespan service is growing. This growing need is especially apparent in the adult space, and as new disorders are being discovered and the service is encountering milder presentations of metabolic disorders previously not described in literature. The services' focus is on continuing to provide excellence in metabolic care and securing funding that can embed the lifespan model of care into the QLD health system, so it is not at risk of future changes in hospital executive or government.

## Importance of Care Pathways

A yet to be released research paper (at the time of writing) on improving post-diagnosis management and communication for people with mitochondrial disorders, finds benefits from patients receiving a letter of diagnosis and general management plan to assist them in navigating generalist care.[22] While many services have developed their own care pathways, models of care or care plans, the consistent use and maximisation of these tools and standards is ultimately dependent on capacity. Resource challenges are covered later under Key Finding 4; however, it is important to note here that our research pointed to care coordination as being one of the first and most significant 'casualties' of services operating with critical workforce shortages.

The absence of these formalised systems and practices to support care coordination and care protocols, or the lack of capacity to fully implement them, is resulting in individual metabolic clinicians or clinics, patient advocacy groups and even individual patients and their families attempting to bridge this gap and coordinate and connect care. Examples of such attempts to coordinate and connect care included:

- Committed generalists (GPs or general paediatricians) taking on the navigation and coordination role between other specialities and allied health;
- An individual clinician trying to develop national clinical guidelines for Niemann-Pick disease type C;
- Fabry Australia initiating a Specialist Medical Advisory Committee that meets quarterly and shares information; and
- Sanfilippo Children's Foundation working with some jurisdictions to develop clinical guidelines.

Many of the fundamental components of delivering multidisciplinary, coordinated care—such as cohesive teamwork, strong collaboration and close linkages or relationships to other services—were self-reported **by over 80%** of specialised metabolic clinical and diagnostic services as existing strengths. Services also shared that these strengths needed to be further built on and formalised, but this requires dedicated funding. We also heard many positive examples of metabolic specialist teams effectively leading and coordinating complex care for patients and their families.

## Transitions from Paediatric to Adult Care

The establishment of consistent clinical care pathways, models of care and shared care plans would go a long way to addressing many of the challenges that patients and carers report in navigating care. Transitions from paediatric to adult services were consistently reported as an area of concern for workforce, patients and carers. At present, transitions occur at a vulnerable age for patients, when many are naturally disengaging from care—if not well managed, these patients are at risk of dropping off the care radar altogether.

Many clinicians pointed to best-practice transitions including a formalised 'shared-care' transition model from approximately 16 to 23 years of age but noted this is not occurring.

# Views of Stakeholders

## Lack of Care Coordination

*"Currently, the strength of care coordination is largely dependent on the success of the mix of personalities and relationships within the service — if this is right, it can work very well."*
– Metabolic Clinician, Focus Group

*"If we could have our metabolic workforce resourced enough to provide the care plan for the year, that gives the paediatrician for the child or adult a really amazing and hard-to-find GP that's taking that role—if they can actually give them the knowledge or the plan for the year, then I think that partnership can work really well. But there are a whole (lot) of barriers of resourcing and funding that make that difficult."*
– Patient Group Leader, Focus Group

*"We recommend [disease group] community to keep their GP or paediatrician involved in every step of their care to allow them to play a coordinating role. This is possible with motivated and collaborative GPs/paediatricians, but only if they are provided with guidance from metabolic teams."*
– Patient Group Leader, Focus Group

*"They come to us to ask: What should be their clinical pathway? We don't have those answers… What families are finding is they have to go and find the information and then go to their doctors and say, 'here's the information that you need in order to tell me how we best look after our children'. It's almost like it's a little bit back to front for [metabolic condition] families. They get a diagnosis and then are told 'come back to us when you know enough about it, for us to be able to help you'."*
– Patient Group Leader, Focus Group

## Standard Models of Care

*"The challenge for us now is that it's a model of care… that we feel is the best and we get the best outcomes. But what we're finding is that it is harder and harder to maintain that model of care going forward. We feel the biggest frustration is not being able to maintain that level of care… based on the level of funding and workforce that we have at the moment."*
– Metabolic Paediatrician, Focus Group

*"Current staffing is grossly inadequate to manage the growing service and requirements of our patient group who often require individualised plan management due to the heterogeneity of the conditions and the individual patient requirements especially for the dietitian, nursing, psychology, clinical and social work [staff]. We also need additional funding for access to allied health to support patients, such as speech pathology, occupational therapy, physiotherapy, additional psychology services and social work."*
– Patient Group Leader, Focus Group

*"This is a horrible thing for people to go through. You go back to a GP or paediatrician and you're probably the first family they've had with this condition. They're scrambling too. There is a really strong argument that we know that these things [standard models of care] work. We just need a workforce that can do them."*
– Patient Group Leader, Focus Group

### Individualised Care Plans

*"It's falling on the specialist to do the right thing. We'd like to see this more formalised… If you could have care plans developed, and that was the norm… like you would do with enhanced primary care plans for chronic complaints or musculoskeletal complaints, why can't something like that be formalised for the rare disease space? So that it is normal practice for a patient to have a care plan developed that is communicated across all avenues, all specialists involved, all allied health involved."*

– Patient Group Leader, Focus Group

*"We've seen that model [individualised care plans] in other conditions in the healthcare system — rare conditions are perfect for this approach because of the multi-systemic impact. This would be a positive way forward and is completely doable without reinventing the wheel."*

– Patient Group Leader, Focus Group

### Transition Support

*"While we are happy with the support and care we receive, I believe all clinics need more resources and am concerned that when she transitions to an adult service there will be less support at the time where she is likely to need more support because the high burden of care will be shifting to her while she also takes on other adult responsibilities."*

– Patient, Patient or Carer Experience Survey

**KEY FINDING 3**

There is significant variation in the composition and maturity of metabolic services and the workforce both *within* and *across* states and territories, as well as *within* and *across* rare metabolic conditions. This creates a 'postcode lottery' for Australians with rare metabolic conditions.

Our research highlighted variation and inconsistency across Australia in the knowledge of, resources for, and approaches to, managing rare metabolic conditions. What this means for patients is a 'postcode lottery'. Put simply, patients living with a particular disease in the right location receive appropriate, high-quality care, leaving many patients with that same disease living in another location with suboptimal care. The following section takes a closer look at the specific composition of specialised metabolic services and the workforce across jurisdictions.

## Gaps in Coverage

Some Australian states and territories do not have established specialised metabolic services. Instead, patients may receive care from individual clinicians or through formal or informal inter-jurisdictional arrangements with other centres. For example, for patients in:

- **TAS:** diagnostic testing is referred to SA and care occurs under a formal agreement with VIC specialist metabolic clinics.
- **WA:** there is capacity for diagnostic testing, but no specialist metabolic clinics.
- **ACT:** testing and care are referred to NSW under a formal agreement.
- **NT:** diagnostic testing and care are referred to SA under an informal agreement.

Where a dedicated metabolic service does not exist within a state or territory, or even where regional referral pathways to tertiary hospitals are not sufficient, there is an increased risk that patients are not connected with any appropriate metabolic care. Often, it takes a formalised diagnostic pathway, like the newborn screening program, or an acute presentation in a hospital setting for patients to be connected with care, leaving a risk that patients who might be 'sub-crisis' status are slipping through the cracks.

## Variation in Access to Care

Australia's geography and dispersed population creates a tyranny of distance for most patients living with a rare metabolic condition outside capital cities with specialised metabolic centres. While access to local services for all patients is not practical, the consequence is a high reliance on telehealth, even prior to the COVID-19 pandemic. Telehealth is a good option to fill some of the gaps and for rural/remote patients; however, some patients and carers surveyed said that while telehealth was 'better than no care', it has significant limitations.

## Impact of COVID-19 Pandemic on Care of Rare Metabolic Conditions

**41%**

of patients and carers surveyed saw a decrease in the frequency in which they accessed health professionals as a result of the COVID-19 pandemic.

**72%**

of patients and carers surveyed experienced an increase in the use of telehealth during the COVID-19 pandemic.

**31%**

of patients and carers surveyed felt that the increased use of telehealth decreased the quality of their health care.

**45%**

of those patients that experienced an increase in telehealth during the COVID-19 pandemic felt that it decreased their access to healthcare professionals.

*"[telehealth] decreased the quality of assessments, access and relationship building between children and doctors."* – Patient or Carer Experience Survey

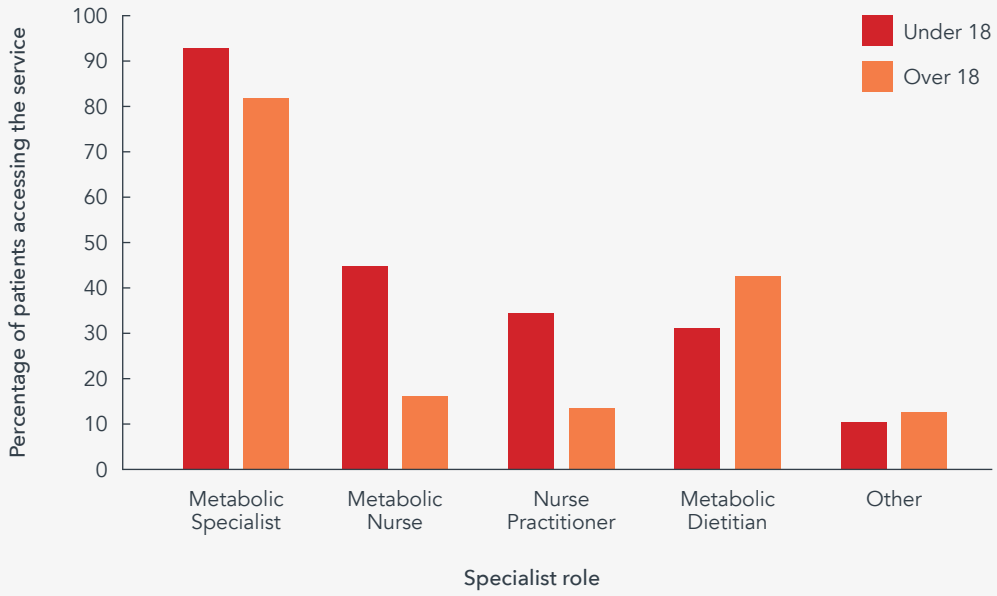
Patients and carers also expressed the view that beyond the COVID-19 pandemic, patients should have more opportunities to periodically access face-to-face specialist care.

Notably, variation in access to care is not limited to clinical services and the workforce. There are also variations in access to genetic testing for timely diagnosis, as well as in the access to and costs of medicines and therapies.

The data also showed extensive variation in the specialised metabolic clinical roles that patients accessed outside specialist metabolic physicians, and this changed depending on patient age.

Specialised metabolic care for paediatric patients is more established than that for adults, and it involves a number of large tertiary hospitals with well-known and well-developed clinical services. This is reflected in Figure 8, which shows that children typically engage more with specialised metabolic roles, such as dietitians and nursing staff, than adults do. Despite this gap, some clinical services caring for adults or for patients of all ages in major hospital settings are maturing and starting to offer team-based care to adult patients.

**FIGURE 8: TYPES OF SPECIALISED METABOLIC CLINICAL ROLES ACCESSED BY PAEDIATRIC AND ADULT PATIENTS SURVEYED**




*Note: This reflects which specialists a patient engages with and may not reflect the frequency of care.*

## Features of a Mature Metabolic Service

As much as capacity will allow, the more mature metabolic services located in major hospitals have also organically started to take on a leadership and education role in the care of rare metabolic patients. Leadership and education are taking place through the provision of consultations and peer support or advice to other clinical or diagnostic services or individual clinicians, both within and across jurisdictions.

Westmead Hospital in Sydney supports outpatient community care and management of rare metabolic patients, for example, for those who may be living in group home settings. This hospital has frequent cross-sector interactions with the National Disability Insurance Scheme and other community and social support services. Staff in the Adult Genetic Metabolic Disorders Service now undertake education sessions, which are also available for new, non-specialised staff within community and disability care, as well as for carers. This service is looking for innovative ways to make this work go further. They will be videoing upcoming sessions and offering their recordings to other services for training purposes.

Effective management of transitions between services is another hallmark of more established, and perhaps more sustainably resourced, metabolic services. There are examples of some services/jurisdictions where transitions involve at least one handover between teams and prioritisation of transitioning patients for engagement with the intake service. In other services/jurisdictions there is no capacity for this. In that case, transitions occur via a letter from the outgoing service, and the transitioning patient may wait more than one year to see the new intake service.

While it is possible to make some general assertions about the maturity of specialised metabolic services based on the capacity and resourcing of metabolic teams, it is important to make the distinction that clinical capacity, expertise and experience does vary across rare metabolic disease groups and sub-groups. This is a natural consequence of the thousands of unique rare and ultra-rare conditions, together with low patient numbers and the geographically dispersed population in Australia.

## Views of Stakeholders

### Gaps in Coverage

*"Sadly, it is the lack of knowledge or awareness of my disease that has created a gap in my healthcare plan. Access to some services would be great but I'm sick of 'doctor shopping' for the right advice and help, so I go without. It's just easier. I'm so tired of asking for help, and not getting anywhere or [being] given the wrong advice."*

– Patient, Patient or Carer Experience Survey

### Specialist Clinicians Involved in Care

*"We have had a number of families, with patients at the early stages of their disease with minimal clinical signs, and then one of their earliest consultations will be with a palliative care team to talk about end-of-life-care. There is often no step with the specialist to actually talk about the disease. There wasn't a lot of empathy or support. They were basically just advised to take their child away and enjoy their life. Every time they go back, they are told the same thing. I've had about five families approach me about this, and say the communication is where it breaks down the most."*

– Patient Group Leader, Focus Group

*"Families do give benefit of the doubt to these clinicians that it is lack of education and lack of understanding about the disease itself, rather than lack of empathy, but it does come across as being unempathetic, unfortunately."*

– Patient Group Leader, Focus Group

### Variations in Access to Care

*"Continuity and making sure loss of person-to-person care is not left behind because we can do telehealth. I am happy to travel to see my doctors… being over 500km away from the hospital sometimes you feel a bit forgotten. Local service for treatment is great, but their commitment is more 'babysitting' than care, and communication between the two [is] not always good. My concerns [are] not reported to Melbourne and locally they are not really followed up as they are only 'care taking'."*

– Patient, Patient or Carer Experience Survey

### Transition of Care Between Providers

*"More communication between jurisdictions would also be good: things may have changed now, but when I first moved to the ACT from NSW it took a long time to transfer my treatment."*

– Patient, Patient or Carer Experience Survey

**KEY FINDING 4**

Critical workforce shortages exist and are not being addressed due to poor alignment between 'activity-based' hospital funding models and the complex, multidisciplinary care that rare metabolic patients require.

## Services Operating Beyond Capacity

A consistent and recurring theme emerging nationally from the research was that current capacity of diagnostic and clinical specialised metabolic services for rare metabolic conditions is stretched.

**75%**

of diagnostic services are regularly undertaking testing outside of standard business hours to meet desired timeframes.

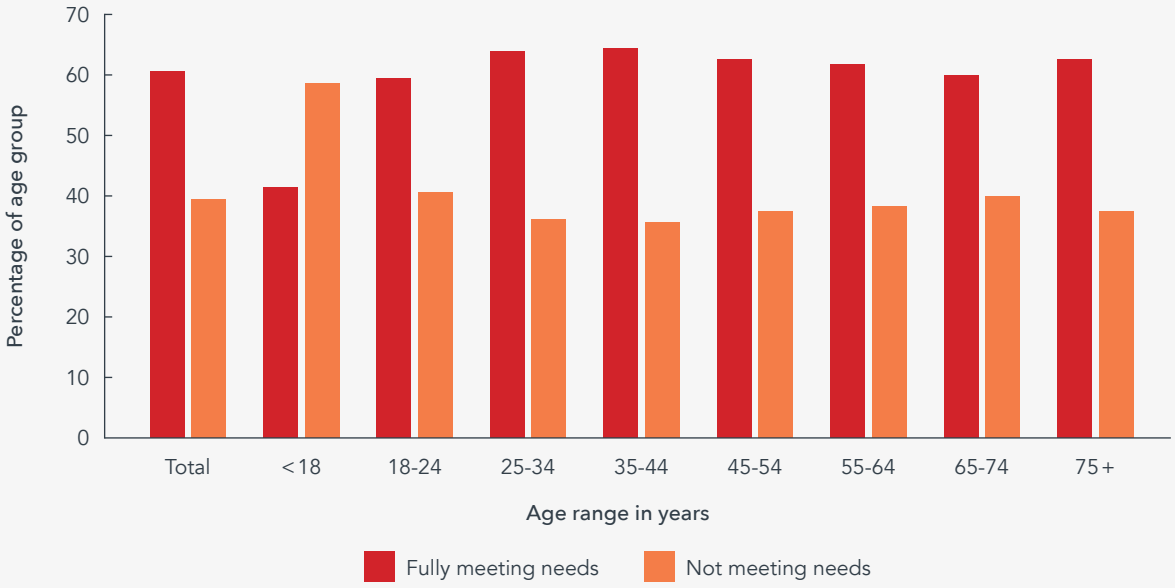
***All clinical services reported critical workforce shortages, indicating that current staffing levels were not sufficient to meet existing patient load.***

All but one diagnostic service felt their service was *inadequately* resourced to meet current and future needs in their service area.

The top challenges cited by the diagnostic workforce that should be addressed to develop a sustainable workforce were ***critical staff shortages*** (in particular, the need for pathologists with biochemical genetics experience, as this is a new obligation under updated National Pathology Accreditation Advisory Council (NPAAC) requirements), ***equipment and lab space***.

When patients and carers were asked if the specialised clinical services they were engaged with were fully meeting their care needs, almost 40% said they were not, or, only partly doing so. Interestingly, for paediatric patients (< 18 years) this percentage was much higher, with almost 59% of respondents (who were mainly the carers of these patients) reflecting that their needs were not being fully met (Figure 9).

**FIGURE 9: EXTENT TO WHICH SPECIALIST SERVICES ARE MEETING PATIENT NEEDS**




When considering the cause of workforce shortages, it is not as simple as inadequate funding for required staffing roles. Both the short and long-term sustainability of the metabolic workforce are at risk because of an inadequate pipeline of suitably trained professionals to replace the existing workforce. This links to Key Finding 1, which cautioned the lack of recognition for the role of, and need for, a specialised metabolic workforce.

# Case Study: Daily Activity of a Metabolic Dietitian

## 'Non-counted' activity-based tasks performed daily to monthly as part of routine patient care

### Clinical Management

The number of different conditions and their spectrum of severity require frequent updating and development of clinical management protocols. This is time consuming, as usually a comprehensive literature review and consultation are needed to ensure accuracy.

Educational resources are multidimensional and individualised, needing to be tailored for each condition and for the chosen dietary therapy. Different resources are created to target different age groups, as well as parents or carers. These are updated and reviewed constantly.

### Communicating and Reporting Results

Monitored blood results are communicated to patients/carers by entry into a patient portal in an electronic medical record, email or phone and/or recording entries into an additional hospital database. This usually involves communicating dietary adjustments to patients/carers in response to results. This is particularly resource intensive when it comes to pregnant patients, as pregnancy often requires at least weekly dietary review, monitoring and adjustment.

### Specialised Product Knowledge

Some metabolic conditions require knowledge of complex and highly specialised food and formula/supplement products. To maintain this knowledge requires regular product updates and reviews. This includes meeting with representatives, updating records/diet composition charts and reviewing food and formula/supplement product suitability with team members. Before providing guidance to patients and their families on the use of a new specialised product, dietitians will often need to undertake actual food preparation. Dietitians are also responsible for education around the use of these products within the hospital setting for inpatients. Therefore, they also work with other staff, including ward nurses and catering, to ensure safety and quality.

### Other Consultation and Coordination Tasks

- Managing outpatient bookings to ensure patient follow-up needs are met.
- Scripts and pathology ordering through the electronic medical record.
- Maintaining patient lists and databases.
- Liaising and collaborating with other individual metabolic dietitians on individual cases, as well as with the professional network of dietitians that exists within the Australasian Society for Inborn Errors of Metabolism.
- Supporting staff from other hospitals to provide specialised food and formula/supplement advice for their patients (e.g. pre-diagnosis, management of ill health).
- Education for carers, as well as for National Disability Insurance Scheme dietitians providing support to clients living in group homes.
- Attending and presenting at conferences — given the complexity and number of different conditions, time must be invested in professional development.
- Supporting high quality research in the field, including peer reviewing.

### Service Coverage

Metabolic dietitians do not have cover from other generalised dietetic services due to the specialised nature of rare metabolic conditions. Without sufficient specialised capacity or coverage, any leave (i.e. annual or sick) must be absorbed by the rest of the team, including 24/7 on-call arrangements. In single-dietitian services, this means urgent results are still reported while on leave.

## Meeting Future Demand

To meet future demand, current expertise must be maintained and also expanded. Diagnostic services reported serious concerns related to a major expertise cliff on the horizon, with the retirement of senior laboratory scientists and pathologists, representing a risk to current service levels. Clinical services also reported a need to think about the supply of future physicians and specialised allied health roles. Three clinical services surveyed had single outstanding vacancies for longer than three months.

Where suitably trained professionals do exist, there are concerns that there are not always enough funded positions for them to move into due to a preference in some centres for generalists.

## Lack of Funded Positions

While there is clearly a need to train more staff to address the impending retirement of key personnel, our research indicates that staff vacancies do not appear to be the issue leading to shortages in clinical services. Instead, as already highlighted, it is the lack of funded positions.

Challenges in justifying the need for more specialised metabolic physicians, dietetics and nursing roles were discussed at length in workforce focus groups, with the shared frustration that not enough 'countable' activity is generated. Funding does not come close to matching up to the actual activities undertaken (the 'behind the scenes' work in preparing for clinics and extensive follow-ups), because many activities are not considered countable. These activities typically include phone calls to patients, preparation of dietary guidance, as well as applications for patients to receive access to treatments or therapies, particularly through the Life Saving Drugs Program (LSDP), which has specific data collection and administrative requirements.

Clinicians reported that administrators told them to record these 'other' activities to demonstrate need. However, clinicians also reported that there was no time to do so. There was a shared understanding that recording the activity would be futile as hospital administrators were limited by the current funding model.

## Inadequate Funding Models

Services receive funding for positions based on measures of service activity. A theme that came through strongly in the National Metabolic Workforce Questionnaire and all Workforce Focus Groups was the misalignment of activity-based funding models and the 'true' activity of medical and health professionals providing complex, multidisciplinary care to patients with rare metabolic conditions.

In particular, activity-based funding fails to capture activity associated with coordination tasks that are essential to deliver complex multidisciplinary care. There was a view that multidisciplinary, integrated care was best-practice. However, funding supported a more outdated, overly simplified transactional approach where time spent in face-to-face or telehealth services with patients was all that tallied. This was one contributing factor to metabolic services not being seen as 'profitable' and, therefore, not a priority for additional investment within a hospital setting. That is, despite it being clear that investment in these models avoids hospitalisations and can help reduce length of stay.

The other factor was that hospitals with specialist metabolic services also care for patients from other Local Health Districts, meaning that any savings from reduced hospitalisations and reduced lengths of stay tend to be shared with other hospitals. While from a system perspective these specialist services lower costs, for individual hospitals, the overall cost of services may be higher.

Because patient load and need so greatly exceeds funded positions available, our research uncovered many examples of services being forced to find creative ways to fund required roles, particularly specialised nursing and nursing coordinator roles. But this is not secure or sustainable. These nursing roles largely provide the coordination and integration that so many services are lacking, so an unstable, piecemeal approach to funding these positions is a major concern.

## Lack of Mental Health Support

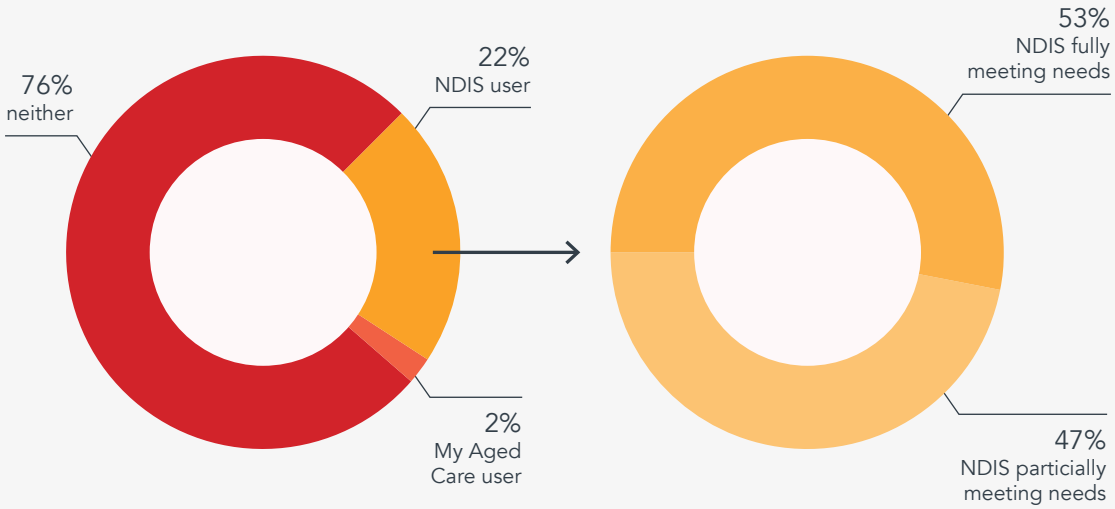
Psychology, social work and social support is not typically part of formalised multidisciplinary metabolic care and the impact of this is being felt by patients. Despite well-documented evidence of the psychological and psychosocial impact of rare metabolic conditions[23, 24], patients engaged with many metabolic services are not receiving mental health support as part of integrated care. A number of patient advocacy group leaders spoke about their own internal research showing the alarmingly disproportionate mental health burden faced by patients and families living with rare metabolic conditions.

To build a sustainable rare disease workforce in Australia, there must be understanding of urgent gaps in workforce. Specifically, understanding and urgently addressing gaps in mental health support, and the overall growing pressures due to the increase in patients diagnosed with rare metabolic conditions and their improved life expectancy.

## Disability Supports

While not directly within the scope of this research, it was highlighted through the data that there is a significant cohort of Australians living with rare metabolic conditions that are engaged with the NDIS. Figure 10 shows the proportion of respondents to the patient and carer survey accessing disability supports (NDIS, My Aged Care or neither). Of those receiving NDIS support, only a little more than half reflected that the NDIS was fully meeting their needs. This is consistent with findings reported in 'Disability & Rare Disease: Towards Person-centred Care for Australians with Rare Diseases'.[25]

**FIGURE 10: NDIS AND MY AGED CARE USE AMONG RESPONDENTS IN THE PATIENT AND CARER EXPERIENCE SURVEY**



# Views of Stakeholders

## Services Operating Beyond Capacity

*"[The major challenge is] Providing enough medical and nursing FTE to meet current and then future demand. Significantly greater resourcing for adult metabolic services [is needed], given the growing nature of the area as paediatric patients survive and transition."*
– Clinical Service, National Metabolic Workforce Questionnaire

*"Lack of trained metabolic specialist positions… lack of formal training program for nursing and allied health staff; workforce is below optimal benchmark by international standards."*
– Clinical Service, National Metabolic Workforce Questionnaire

## Meeting Future Demand

*"Staff numbers are unsustainable to cope with current numbers and expected growth over the years to come. This is across the board for scientists, doctors, nurse practitioners, nurses, dietitians, social workers, psychologists. We desperately need administration support, [a] clinical research manager and a genetic counsellor. And access to speech pathologists, occupational therapists and physiotherapists."*
– Clinical Service, National Metabolic Workforce Questionnaire

*"A paediatric hospital is the lowest on the ladder and we [the metabolic service] are the smallest unit within the lowest rung of the ladder. So, we are at the very, very bottom of the pecking order and so it's meant that we've really struggled over the years – it's an inequity for ourselves in terms of how we are seen across the health system. But we can sit here and complain as much as we want, but I think what we want is to do something about it, so we can make sure we can provide for our patients into the future."*
– Metabolic Physician, Focus Group

## Lack of Funded Positions

*"Improved funding for trainees to allow future workforce development."*
– Clinical Service, National Metabolic Workforce Questionnaire

*"Education of clinicians in adult presentations of inborn errors of metabolism."*
– Clinical Service, National Metabolic Workforce Questionnaire

*"Ongoing funding for metabolic trainees."*
– Clinical Service, National Metabolic Workforce Questionnaire

*"I am also concerned about a likely 'bottle-neck' with jobs in the next few years. We have a number of metabolic trainees currently, many of whom are worried about getting a job in the future. Many centres employ general paediatricians, which has been useful to fill job shortages. However, space needs to be made for metabolic trainees."*
– Metabolic Consultant Physician, Written Submission

## Inadequate Funding Models

*"Daily activity is not captured, particularly for work which occurs outside of the hospital admitted system (for allied health and nursing follow-up); phone follow-up not captured and yet this leads to prevention of admission."*
– Clinical Service, National Metabolic Workforce Questionnaire

*"The metabolic service is bottom of the pecking order because we're not a big unit. We're not providing an income for the hospital. So, we're not considered to get additional funding."*
– Metabolic Physician, Focus Group

*"Often relying on 'other' funding to fund nurse coordinators… funded primarily from industry funding. We don't think that's the ideal, but we don't have funding coming from any other source. We would like to see government funding because it's not sustainable. Often, it's registry funding, or research funding… it's not reliable and it's a real concern."*
– Specialist Metabolic Nurse, Focus Group

*"To upgrade… my role into a clinical nurse consultant, which I don't have at the moment because of funding. The position hasn't been regraded. I need to find the funding to pay for upgrade myself. I have managed to find it… we get paid funds for entering data into international registries. With the agreement with others, I can use those funds to upgrade it [my role]. I am essentially setting that in place myself, with the agreement of my hospital, of course. But that's where the funding is coming from. That's what I was directed from above – 'where are you going to find the funding?'"*
– Specialist Metabolic Nurse, Focus Group

*"I am in a catch-22 where I need to provide more support to my patients, but I am working at a higher level [than I am being paid]. I do every day. You look at the database funding and think I could supplement my income with that, but then all the time it takes to enter the data, then you're not seeing your patients. I feel like my time is better spent on the patients, so do I sit here and enter data just to be paid for my role?"*
– Specialist Metabolic Nurse, Focus Group

## Lack of Mental Health Supports

*"Clinical care I experience at present lacks the multifaceted nature of the rare metabolic condition I am diagnosed with. A metabolic dietitian and genetic specialist covers a small proportion of the affected population's need. Treatment with an extremely limited dietary therapy and high mental health comorbidities requires psychological care."*
– Patient, Patient or Carer Experience Survey

*"More services attached to the clinic that can help patients, for example, psychologist, mental health services, counselling."*
– Patient, Patient or Carer Experience Survey

*"A 2018 study found a large proportion of patients with mitochondrial disease were experiencing mental health issues. The study found that 42% of respondents were suffering from anxiety and sleep issues; and 41% were experiencing depression. This is significantly higher than the Australian national average of 16% suffering from depression and 25% suffering anxiety."*
– Mito Foundation Representative, Written Submission

*"They [clinical services] have advised that they do not have access to other services such as psychology to assess functional outcomes of patients regularly."*
– Industry Representative, Survey

**KEY FINDING 5**

Specialised metabolic services are not sufficiently resourced to contribute to innovation. This is a barrier to the development and trialling of emerging therapies and technologies in Australia, which could offer significant benefits to patients.

The industry survey results supported the view that Australia is falling behind other countries as an attractive location for clinical trials, and in the development and approval of emerging medicines to treat rare diseases. Patients are also acutely aware that living in Australia is not affording them access to treatments that are available and subsidised internationally.

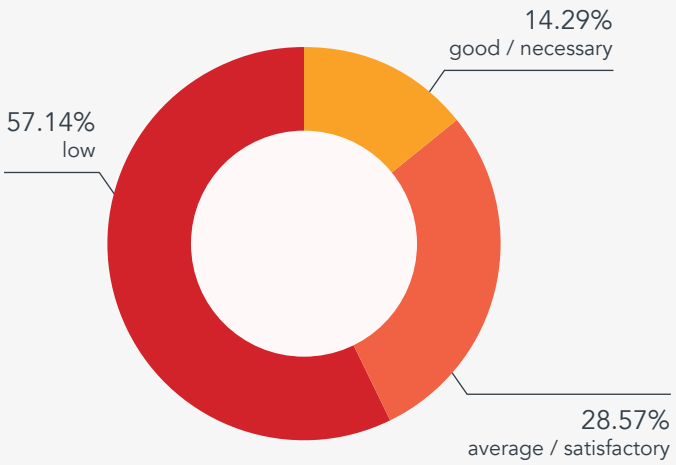
## Inadequate Workforce and Infrastructure to Support Clinical Trials

While critical workforce shortages compromise the level and coordination of care, insufficient staff and other resources also limit the capacity of clinics or services to participate in clinical trials, or to provide the clinical management necessary for promising new therapies, such as gene and cell therapies.

It is challenging to maintain the necessary ongoing requirements, including senior staffing supervision and instrumentation, to meet the needs of sporadic clinical trials. Similarly, it is difficult to meet the time critical requirements of setting up a clinical trial without an accessible workforce and flexible staff funding arrangements.

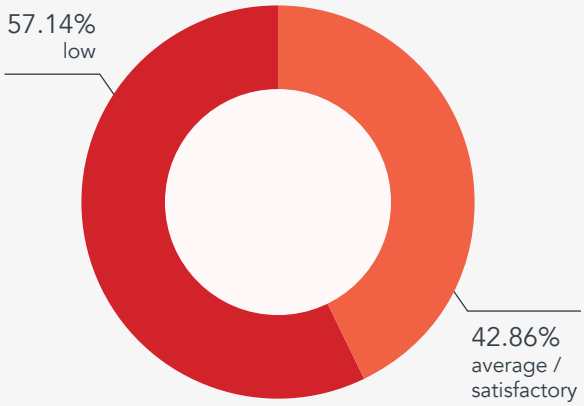
Of the small sample of companies surveyed that develop therapies or technologies for rare metabolic conditions, most had a bleak view of the current overall capacity (resource levels, skills and infrastructure) of Australian metabolic services to *facilitate* clinical trials (Figure 11).

**FIGURE 11: INDUSTRY VIEW OF AUSTRALIAN METABOLIC SERVICES' CAPACITY TO FACILITATE CLINICAL TRIALS**



There was a similar response when it came to the capacity (resource levels, skills and infrastructure) for Australian metabolic services to *provide the clinical management* necessary for emerging therapies or technologies (Figure 12). Industry representatives pointed out that it is not the capability or motivation across the specialised metabolic workforce that is lacking, rather it is simply a shortage of resources.

**FIGURE 12: INDUSTRY VIEW OF AUSTRALIAN METABOLIC SERVICES' CAPACITY TO PROVIDE NECESSARY CLINICAL MANAGEMENT FOR CLINICAL TRIALS**




## Access to New Treatments

While there are exciting developments on the horizon for new therapies and technologies, particularly with a move toward more personalised approaches like cell and gene therapies, the need for and complexity of clinical management is going to increase. Australians will not benefit from these new treatments if the workforce is unable to keep up with advances in skills and capabilities or allocate the time to appropriately support their use.

One interesting observation from industry was that most Australian metabolic services do not have psychology as part of their MDT. This becomes a barrier to participation in clinical trials, as close assessment of the impact of therapies and technologies on a patient's mental health and capacity is necessary.

# Views of Stakeholders

## Inadequate Workforce and Infrastructure to Support Clinical Trials

*"Even a single trial would put enough strain on a service, let alone consideration of running multiple at once."*

– Senior Scientist, Screening Service, Focus Group

*"Ideally, once set up, the site needs an ongoing flow of studies to make it sustainable, rather than dipping in and out of clinical trial work, and this requires dedicated FTEs (e.g. permanent Fellow position), and/or support from a co-located clinical research centre. Having a Fellow position one year and then having it taken away the next does not support trials or succession planning for clinical services."*

– Industry Representative, Industry Survey

*"On several occasions our organisation has tried to recruit and establish trial sites in Australia. However, due to the burden of current case load many opportunities for clinical trials are passed up."*

– Industry Representative, Industry Survey

*"Ambition is there, but the infrastructure is lacking. The willingness from clinicians is there, but the support is not… Resources are likely strained due to cost of services and budget pressure."*

– Industry Representative, Industry Survey

*"The intention is there, however the challenges lie in the complexity and challenges that each site has for opening a trial, as well as the burden on investigators on top of clinical management workloads. Most often, investigators will find a way as they recognise that for some patients, a trial is their only treatment option. Our clinicians want to trial new medicines and are close to scientific advance; however, [it is] likely they have to make a trade off on their workload and site limitations."*

– Industry Representative, Industry Survey

## Access to New Treatments

*"High incidence of bone density issues and white matter changes in adults [with phenylketonuria] requires better care and access to pharmaceuticals available overseas to improve the management and quality of life of those living with the condition."*

– Carer, Patient or Carer Experience Survey

*"[We need] to improve patient access to potential new treatments and cures for the metabolic conditions. There seem to be many more options available overseas than locally."*

– Carer, Patient or Carer Experience Survey

*"While my personal metabolic team do a wonderful job, I find it disheartening to see that other countries in the world have access to multiple forms of ongoing treatment."*

– Carer, Patient or Carer Experience Survey

*"Some of the forthcoming therapies require significant clinical resources to manage adverse events and dose titration over an extended period until the immune response is overcome and the patients reach efficacy, with subsequent dose adjustment and significant dietary management required. The clinics have advised that they do not have the resources for this additional workload, and it may hamper their ability to start patients on this life changing therapy."*
– Industry Representative, Survey

*"I also think lack of resources limits the potential for clinical trials and requires existing staff to do additional hours (example–we participated in a research project that required attendance at the hospital after hours). Some of the new medicines that are coming would require increased surveillance compared to current surveillance levels."*
– Carer, Patient or Carer Experience Survey

# LESSONS GLOBALLY: A SNAPSHOT OF THE INTERNATIONAL RARE METABOLIC DISEASE WORKFORCE

## A review of the international landscape reveals that in countries with comparable health systems to Australia, the rare metabolic workforce is typically more developed and defined.

All comparable countries in an international scan undertaken to inform the development of this White Paper recognised that an MDT approach represents the best care to support metabolic patients. The significance of MDT care is recognised for patients at all life stages, including during metabolic crisis or pregnancy, and for those with mental health issues.[12]

The United Kingdom (UK) and a number of European countries have formalised Centres of Excellence (CoE).[26, 27] In the UK, key characteristics to define CoE have been identified, and are also highlighted in the Action Plan. These characteristics include:

- Coordinated care;
- Adequate caseload to ensure expertise to support sustainability of care provision and facilitation of research;
- Not being dependent on a single clinician;
- Arrangements for transition from child to adult services;
- Information hub and location for peer interaction;
- Engagement with people living with a rare disease;
- Research active;
- Education and training for medical professionals; and
- Membership of international networks of excellence.

These collaborative centres are integrated with adaptive infrastructure, including databases and information technology systems, to strengthen clinical and research work. Internationally, these networks allow for research and clinical capacity to grow together with appropriate resources. Three international examples and a case example are presented below to demonstrate the variability of how rare metabolic patient care is provided.

## United Kingdom

The UK's approach to rare metabolic disease management focuses on specialist centres providing services to a local area, with some networking to broader European CoE. A 2006 study found that the ratio of 500 rare metabolic patients to one doctor, one nurse and one dietitian was not enough to meet demand for services.[12] While resourcing improved in 2006, there are no current workforce figures available.

### Key Statistics

- 24 specific metabolic disease centres across 16 cities.
- 6 specific adult or paediatric services.
- 10,000 patients looked after.

### Strengths

- MDT approach – specialist physicians, metabolic nurses and metabolic dietitians in teams with networks reaching out to other specialists when required.
- Some of the centres are part of the European Union's MetabERN (metabolic European Reference Network), which helps with access to relevant researchers and novel treatments.
- Laboratory connections are prevalent for many of the centres.

### Gaps

- Gross inequality between urban and regional centres, with only four regions, including London, having access to a centre scoring highly, while some did not have a service at all.
- Significant staff shortages causing variation in what specific services are available at each centre.
- At the time of the last comprehensive audit in 2006, approximately 47% of all metabolic patients were not being seen in a specialist centre.

## Ireland

Ireland has a single country-wide service—National Centre for Inherited Metabolic Disorders based in Dublin[28]—which provides an MDT approach to rare metabolic patient care.

### Key Statistics

- 8-10 outpatient clinics are run per week, including condition-specific clinics and clinics in other healthcare centres.

### Strengths

- MDT approach includes medical, nursing, dietetic, psychology, social work, laboratory staff and a play specialist.
- Lifelong treatment for patients with metabolic disorders, including support for their families.
- Linked to diagnostic laboratory, newborn screening and an inpatient high-dependency ward specifically for rare metabolic patients.
- Strong research links.

### Gaps

- Limited outreach clinics currently available, with plans to expand.
- No published audit data is available, so it is difficult to ascertain exact numbers of patients cared for and if there are any gaps in service provision.

## Brazil

Consultation with leading metabolic specialists in Brazil revealed that there is a well-developed network of services in the public system that supports clinicians and patients with rare metabolic conditions. There are information services, and networks for specific conditions have also been developed for lysosomal storage diseases, mucopolysaccharidoses and Niemann-Pick Type C.

The coordinating centre is located in Hospital de Clínicas de Porto Alegre, in the Southernmost state of Brazil Rio Grande do Sul, and was first established in 1982 becoming the coordinating centre in 2008. There are 4 associated centres in each region of Brazil, and 19 participant centres.

Current staff at the coordinating centre include:

- 13 rare metabolic clinicians
- 4 rare metabolic nurses
- 2 dietitians
- 2 psychologist/psychiatrists
- 1 social worker

Other:

- 12 lab staff (biochemists, molecular biologists, cytogeneticists, lab technicians)
- 4 administrative staff

These staff provide coordinated metabolic services across Brazil, servicing approximately 2,000 patients per year.

# Case Example: International Best Practice

## Excerpt from the Action Plan

"ERNs have achieved global recognition in facilitating the delivery of expert care, despite geographical boundaries and distances. Virtual ERNs are comprised of healthcare professionals and are spread throughout Europe. The ERNs' objective is to tackle complex or rare diseases and conditions that require highly specialised treatment and a concentration of knowledge and resources. ERNs facilitate the exchange of knowledge between healthcare professionals across borders, giving people access to the expert knowledge they need even if it's not available in their own country or region."[1]

## European Reference Networks

European Reference Networks were first approved by the European Commission in 2017 to allow a unique opportunity for clinicians and researchers to work across borders. There are currently 24 ERNs addressing common challenges in rare disease health care. These ERN's bring together the scarcity of patients and specific expertise from across Europe and the UK for the benefit of all people living with a rare disease. ERN's facilitate the sharing of knowledge, expertise, medical research, teaching and training resources, and coordination of care. They are grouped by therapeutic area and do not prevent patients from accessing care specific to their unique diagnosis. Patients with multi-systemic rare diseases are often cared for under several ERNs.[29]

## MetabERN

MetabERN is the ERN for hereditary rare metabolic disorders. This ERN includes approved CoE, healthcare providers and laboratories from across Europe and the UK. It connects specialised centers on inherited metabolic diseases to drive patient-centered care and improve quality of life for patients and their families. The MetabERN model facilitates care for over 60,000 patients.[30]

A continually growing and developing network, MetabERN represents 78 nationally certified healthcare providers from 23 European Member States, 41 patient organisations and it is endorsed by the Society for the Inborn Errors of Metabolism.[30]

## MetabERN's Mission:

'To identify and bring together the best expertise from across Europe to facilitate prevention, diagnosis, management, research and access to the best available care for patients affected by rare inherited metabolic diseases'[30]

## MetabERN's Aims:

- Pool knowledge and improve information exchange between network members;
- Improve diagnosis and care in disease areas where expertise is rare;
- Support all Member States to provide highly specialised care to patients affected by Inherited Metabolic Disorders (IMDs);
- Advance innovation in medical science and health technologies for IMDs;
- Provide cross-border medical training and research on IMDs; and
- Support all patient initiatives towards harmonising and improving all aspects of the care chain.[30]

For the best approach to managing this group of over 1,600 genetic disorders, MetabERN is structured under 7 disorder groups, which form the basis for MetabERN subnetworks.

## Application to the Australian Context

With Commonwealth support, this type of governance, structure and approach could be applied across the states and territories in Australia. Establishing networks similar to MetabERN would allow for the sharing of knowledge and expertise, while facilitating care-coordination so every Australian living with a rare metabolic condition has equitable access to quality metabolic care.

# MetabERN Structure and Governance

**METABERN BOARD**

(General assembly of representative from all HCPs, patient groups and other third party stakeholders involved in the activites of the MetabERN)

**Medical Executive Board (MEB)**
(Including coordinator, vice-coordinator and leadership of sub-networks)

**MetabERN Patient Board**
(Including ePAG representatives as well as all other patient epresentatives involved in the advisory boards of individual sub-groups)

**MetabERN Advisory Board (MAB)**
(Including ePAG patient and family associations representatives, policy-makers, foundations, ethics specialists, collaborating networks and partnering ERNs where an overlap of diseases requires coordination)

**External Experts Committee**
(Including external experts, lead and supervised by the SSIEM)

| Sub-Groups | Amino- and organic acid related disorders (AOA) | Carbohydrate, fatty acid oxidation, ketone bodies disorders (C-FAO) | Lysosomal storage disorders (LSD) | Peroxisomal disorders (PD) | Disorders of pyruvate metabolism, mitochondrial oxidative disorders, thiamine transport and metabolism (PM-MD) | Congenital disorders of glycosylation and intracellular trafficking CDG | Disorders of neuromodulators and other small molecules (NOMS) |
|---|---|---|---|---|---|---|---|
| | AOA Medical Executive Board | C-FAO Executive Board | LSD Executive Board | PD Executive Board | PM-MD Executive Board | CDG Executive Board | NOMS Executive Board |
| | AOA Advisory Board | C-FAO Advisory Board | LSD Advisory Board | PD Advisory Board | PM-MD Advisory Board | CDG Advisory Board | NOMS Advisory Board |
| | Phenylketonuria | Glycogen storage disorders | Mucopolysaccharidoses | Zellweger spectrum | TBD | n/a | Cerebral creatine deficiency |
| | Urea cycle disorders | Disorders of galactose metabolism | Sphingolipidoses | D-bifunctional protein | TBD | n/a | Disorders of minerals |
| | Maple syrup urine disease | Gluconeogenesis | Mucolipidoses | Refsum diseases | TBD | n/a | GABA Disorders |

**Thematic Committees**
(Including prevention and screening, new diseases diagnosis, guidelines and SOPs, quality of life, ethics and transparency, registries and outcome measures, patient empowerment, education and training)

*Image modified from* https://metab.ern-net.eu/about-us-3/#structure [31]

# CRITERIA TO ESTABLISH JURISDICTIONAL BASELINES FOR METABOLIC CARE

Our 5 key findings point to issues with the Australian rare metabolic workforce impacting current and future service provision. While these issues are present across Australia and many of the problems are shared, differences between states and territories mean there are different needs to address. To assess current gaps across state and territory services, it is necessary to establish a benchmark or set of criteria.

Below are a set of criteria, based on the Action Plan[1] and the key findings in this report, that we have applied to determine whether services across states and territories are able to meet current needs and adapt to future demands. We use these criteria in the next section to assess the current jurisdictional baselines across states and territories for rare metabolic services (the service level data presented in this section is accurate to the end of August 2021). Where these services do not exist, we provide an assessment of the current arrangements in those jurisdictions for the provision of specialist metabolic care.

As was highlighted in Key Finding 1, not all patients with rare metabolic conditions are under the care of specialised metabolic services (Figure 13). Even in jurisdictions where these services do exist, there are other pockets of specialist care being provided to patients all across Australia, particularly in the specialty areas of clinical genetics, neurology, nephrology and endocrinology. Throughout this research we became aware of examples where this was occurring but could not comprehensively report or map these. However, it is important to recognise that other specialty services represent a vital part of current service arrangements. Without this additional help from other specialty services, existing metabolic services would be experiencing greater patient need.

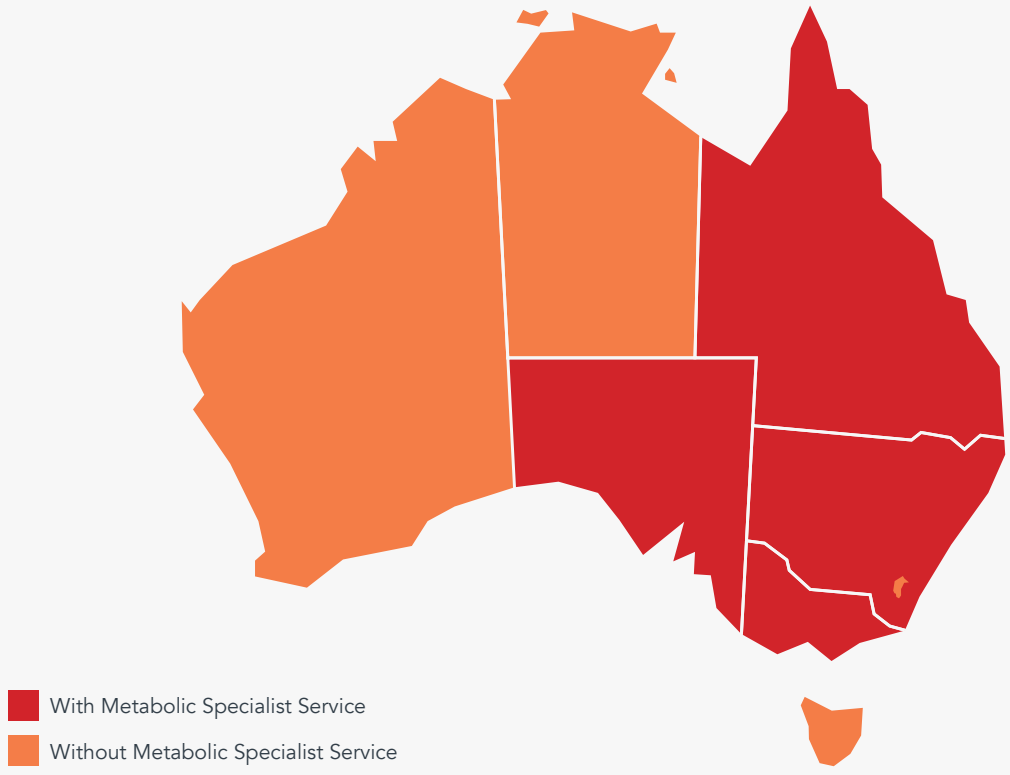
**Criteria applied to determine whether current services across states and territories are able to meet current needs and adapt to future demands:**

- **Criteria 1:** Timely and local access to multidisciplinary services that include specialist metabolic clinicians, diagnostics, metabolic dietitians, metabolic nurses, genetic counsellors, clinical trial staff, allied health, mental health, disability support and social workers.
- **Criteria 2:** Models of care that include formalised integrated whole-of-life care pathways, care coordination, 24-hour service and support to transition from paediatric to adult services.
- **Criteria 3:** Adequate levels of resourcing and infrastructure to support access to emerging therapies and technologies.
- **Criteria 4:** Well established training pathways and succession planning to ensure services are robust and sustainable.

This visual scale is used in the following section to show how each jurisdiction measures up to these 4 criteria. The box highlighted in red indicates current circumstances under which a criterion is or is not being met.

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | **Fully meets criteria** |
|---|---|---|---|---|

**FIGURE 13: STATES AND TERRITORIES WITH AND WITHOUT A METABOLIC SPECIALIST SERVICE**



# New South Wales

As the largest state in Australia, NSW has well established specialised metabolic clinical and diagnostic services.

Clinical specialist centres include the Adult Genetic Metabolic Disorders Services at Westmead Hospital; Genetic Disorders Service at The Children's Hospital, Westmead; and Genetic Disorders Service at The Children's Hospital, Randwick. Diagnostic services include the NSW Biochemical Genetics Service and NSW Newborn Screening Program. All these services also provide support to patients and diagnostic services to people living in the ACT. However, gaps remain in service coverage, and 35% of patients surveyed in NSW reported that their needs were not being fully met.

## How does NSW measure up to the criteria?

### Criteria 1: Timely and local access to multidisciplinary services that include specialist metabolic clinicians, diagnostics, metabolic dietitians, metabolic nurses, genetic counsellors, clinical trial staff, allied health, mental health, disability support and social workers.

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

The main specialist centres for rare metabolic conditions in NSW provide access to MDTs. However, this does not include integrated psychological services and there are gaps in access to allied health professionals across all services. For example, fewer than 50% of patients surveyed had seen a dietitian in the previous 12 months.

Importantly, the major centres at Westmead are providing outreach services to regional hospitals and diagnostic services operate state-wide, ensuring access to specialist services for people across NSW. However, because the specialist clinical services provided by Westmead result in savings to other health services, in terms of both reduced admissions and reduced length of stay for patients, there is ineffective cross subsidisation occurring.

### Criteria 2: Models of care that include formalised integrated whole-of-life care pathways, care coordination, 24-hour service and support to transition from paediatric to adult services.

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

NSW does not have one integrated whole-of-life centre; however, it does have pediatric and adult care linked by transition services. Nonetheless, fewer than one third of patients with rare metabolic conditions in NSW report having access to care coordination services, which are necessary to improve outcomes for patients with complex chronic conditions requiring multiple healthcare providers to effectively manage their care.

### Criteria 3: Adequate levels of resourcing and infrastructure to support access to emerging therapies and technologies.

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

Clinical trials are supported by services in NSW, providing patients with early access to new treatments. However, there is inadequate resourcing for services to take up all available opportunities or integrate these new therapies into care. As new therapies become available, these resource constraints will become more acute.

**Criteria 4: Well established training pathways and succession planning to ensure services are robust and sustainable.**

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

Services in NSW reported a lack of formal training programs for nursing and allied health staff, and the need for improved funding for trainees to allow future workforce development.

DIAGNOSTIC SERVICE

# NSW Biochemical Genetics Service | New South Wales

**SERVICE AREA**

All of NSW and provides services nationwide

## Arrangements

Works closely with the NSW Newborn Screening Program and linked to the Children's Hospital, Westmead (CHW). Also involved with diagnosis and follow-up of infants diagnosed via the Newborn Screening Program.

Currently 13 FTE staff.

## Infant Care

**Diagnosed:**

- 9 infants in 2020–21
- 10 infants in 2019–20
- 12 infants in 2018–19

**Tested:**

- 2,400 infants in 2020–21
- Approximately 4,000 infants in 2019–20 and 2018–19

## Paediatric Care

**Diagnosed:**

- 11 children in 2020–21
- 9 children in 2019–20
- 9 children in 2018–19

**Tested:**

- 7,200 children in 2020–21
- Approximately 9,000 children in 2019–20 and 2018–19

## Adult Care

**Diagnosed:**

- 5 adults in 2020–21
- 6 adults in 2019–20
- 5 adults 2018–19

**Tested:**

- 2,400 adults in 2020–21
- Approximately 3,000 adults in 2019–20 and 2018–19

## Services

Diagnostic services; genetic counselling and screening; research collaborations, including clinical trials; training of laboratorians; and physicians/ pathologists.

## Strengths

- Has access to specialised staff and high-end technologies for diagnosis and monitoring.
- Opportunities for research collaboration with nearby centres.
- Close relationships with newborn screening program, molecular genetics and cytogenetics laboratories, as well as metabolic and clinical genetics services.

## Challenges

- The new supervisory regulation introduced by National Pathology Accreditation Advisory Council (NPAAC).
- A lack of funded training opportunities for scientists in biochemical genetics, or other genetics laboratorian training programs.
- Seeing an impending critical shortage of scientists in the diagnostic field, particularly for scientists with specific esoteric skill sets.

DIAGNOSTIC SERVICE

# NSW Newborn Screening Programme | New South Wales

**SERVICE AREA**

All of NSW and supporting ACT

## Arrangements

Linked to CHW and the Sydney Children's Hospital's Network (SCHN).

Has offered services to all infants born in the Australian Capital Territory (ACT) since 1983.

## Infant Care

**Diagnosed:**

- 153 infants in 2020–21
- 163 infants in 2019–20
- 157 infants in 2018–19

Testing all infants in NSW and the ACT each year.

## Adult Care

**Testing:**

- Approximately 480 adults each year

Made zero diagnoses.

## Paediatric Care

**Testing:**

- 202 children in 2020–21
- 210 children in 2019–20
- 201 children in 2018–19

## Strengths

- Highly competent and dedicated team.
- Ability to develop new techniques.

## Services

- Diagnostic services.
- Genetic counselling and screening.

## Challenges

- Staff shortages due to requiring a pathologist with scope of practice in biochemical genetics, according to new regulation by NPAAC.
- Staff shortages in hospital scientists and clinical nurse specialists.
- Providing ongoing training to staff and scientists.
- Needing to replace ageing equipment to provide the best possible service. Shortage of space to meet current and future needs. This challenge is exacerbated by the COVID-19 pandemic social-distancing requirements.
- A lack of recognition of the multidisciplinary aspects of the service.

CLINICAL SERVICE

# Genetic Metabolic Disorders Service

**The Children's Hospital, Westmead, Sydney | New South Wales**

**SERVICE AREA**

All of NSW. Supports Australian Capital Territory and Western Australia.

## Staff Profile

- 3 FTE Metabolic clinicians
- 2 FTE Metabolic nurses
- 2 FTE Metabolic dietitians
- 1 FTE Metabolic social worker

## Infant Care

**Diagnosed:**

- 30 infants in 2020–21
- 22 infants in 2019–20
- 9 infants in 2018–19

**Provided services to:**

- 39 infants in 2020–21
- 43 infants in 2019–20
- 25 infants in 2018–19

## Paediatric Care

**Diagnosed:**

- 35 children in 2020–21
- 26 children in 2019–20
- 24 children in 2018–19

**Provided services to:**

- 1,012 children in 2020–21
- 1,035 children in 2019–20
- 998 children in 2018–19

## Telehealth

- 378 services in 2020–21
- 156 services in 2019–20

## Vacancies

- 1 FTE Metabolic clinician
- 1 FTE Metabolic dietitian

## Outpatient Clinics

- 321 held in 2020–21, 598 patients
- 261 held in 2019–20, 930 patients
- 255 held in 2018–19, 808 patients

## Services

Diagnostic services; clinical care; care coordination; specialist dietitian services; genetic counselling; paediatrics; formal/informal transition programs for paediatric to adult care; research collaborations, including clinical trials; outpatient clinics; outreach services to regional hospitals; 24-hour on-call services linked to emergency departments; telehealth services; clinical support to state-based biochemical genetics and newborn screening laboratories; NSW and ACT education and training; and social work support.

## Strengths

- Multidisciplinary team.
- Access to specialist laboratory services.
- Collaboration with genetics services; nutrition education assistant; national metabolic network; international networks; and services across SCHN.
- Accredited metabolic medical workforce training site.
- Established supportive relationship with state-based adult metabolic service.
- Established comprehensive model of care providing surveillance to prevent admissions.
- Transition services increasing in comprehensiveness.
- Shared model of care for delivery of enzyme replacement therapy across NSW and ACT.
- Collaborative work with sub-specialty services across CHW.
- Integration of clinical service with laboratory services (biochemical genetics and laboratory services) including governance processes for supporting laboratory follow-up.
- Established intake process to govern and close the loop on consultations and referrals with peer review.

CLINICAL SERVICE

## Genetic Metabolic Disorders Service

**The Children's Hospital, Westmead, Sydney | New South Wales**

**SERVICE AREA**

All of NSW. Supports Australian Capital Territory and Western Australia.

### Challenges

- Service is beyond capacity with current staffing.
- Clinical care to large geographical region with regional and remote communities.
- Limited capacity to provide social and nursing care.
- Limited staff and resources to conduct clinical trial research.
- Lack of resources to meet clinical requirements for novel therapeutics.
- Lack of formal training program for nursing and allied health staff.
- No integrated psychologist services.
- Inadequate counselling support for increasing novel diagnostics.
- Physical space and facilities limitations.
- Unable to provide further comprehensive education, training and research.
- Lack of formalised models of care.
- Large proportion of activity not counted toward funding.
- Limited patient database.
- Telehealth services not established and unfunded.
- Lack of capacity to provide education for non-healthcare services (schools, childcare centres).

CLINICAL SERVICE

# Genetic Metabolic Disorders Service

**Sydney Children's Hospital, Randwick, Sydney | New South Wales**

**SERVICE AREA**

All of NSW. Also provides services to Australian Capital Territory.

## Staff Profile

- 1 FTE Metabolic clinician
- 1 FTE Nurse practitioner
- 1 FTE Social worker

## Infant Care

Diagnosed approximately 10 infants per year from 2018–21.

## Telehealth

**Provided:**

- Over 100 services in 2020–21
- Over 50 services in 2019–20
- No services provided in 2018–19

## Paediatric Care

Diagnosed approximately 20 children per year from 2018–21.

Provided services to over 100 children per year.

## Challenges

Additional resources needed, particularly staffing.

## Outpatient Clinics

**Operated:**

- 45 clinics from 2018–21
- Approximately 150 patients attended these outpatient clinics each year

## Services

Diagnostic; clinical care; care coordination; specialist dietitian services; genetic counselling; paediatrics; formal/informal transition services for paediatric to adult care; research collaboration, including clinical trials; outpatient clinics; 24-hour on-call service linked to emergency departments; and telehealth services.

## Strengths

- Networked service with CHW.
- Skilled clinicians.
- Access to diagnostic services on-site.
- Strong collaborations with other centres.
- Access to clinical and molecular geneticists.

CLINICAL SERVICE

# Adult Genetic Metabolic Disorders Service

**Westmead Hospital, Sydney | New South Wales**

**SERVICE AREA**

All of NSW. Also provides services to Australian Capital Territory.

## Staff Profile

- 2 FTE Metabolic clinicians
- 2 FTE Metabolic nurses
- 1 FTE Metabolic dietitian
- 1 FTE Other

## Telehealth

**Provided:**

- 150 services in 2020–21
- 182 services in 2019–20

## Outpatient Clinics

**Operated:**

- 80 adult clinics in 2018–21

**Provided outpatient services to:**

- 293 patients in clinics in 2020–21
- 671 patients in clinics in 2019–20
- 595 patients in clinics in 2018–19

## Adult Care

**Testing:**

- 65 adults in 2020–21
- 68 adults in 2019–20
- 58 adults in 2018–19

**Provided services to:**

- 277 adults in 2020–21
- 452 adults in 2019–20
- 354 adults in 2018–19

## Strengths

- Coordinated care with the co-located paediatric metabolic service (at CHW), including laboratory services.
- Integration of clinical research and trials with standard care.

## Services

Clinical care; care coordination; specialist dietitian services; genetic counselling; formal/informal transition programs for paediatric to adult care; research collaborations, including clinical trials; outpatient clinics; 24-hour on-call service linked to emergency departments; telehealth services; and metabolic advice for pregnancies.

## Challenges

- Inadequate funding for trainees to allow future workforce development.
- Inadequate funding of allied health and nursing support to the service to meet current patient need.
- Lack of formalised education for clinicians in adult presentations of inborn errors of metabolism.

## Australian Capital Territory

There are an estimated 215 patients in the ACT with rare metabolic conditions. There is no specialist metabolic service operating in the ACT; however, patients can access diagnostic services through NSW's Newborn Screening Program and clinical services through Westmead Hospital.

### How does ACT measure up to the criteria?

**Criteria 1: Timely and local access to multidisciplinary services that include specialist metabolic clinicians, diagnostics, metabolic dietitians, metabolic nurses, genetic counsellors, clinical trial staff, allied health, mental health, disability support and social workers.**

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | **Partially meets criteria through formal arrangements with other jurisdiction/s** | Partially meets criteria through service within own jurisdiction | Fully meets criteria |
|---|---|---|---|---|

Patients in the ACT benefit from services provided through Westmead Hospital; however, the patient survey results indicated that 50% of adult patients residing in the ACT are not engaged with specialist services. There are also some outreach clinics in the ACT run by Westmead Hospital, which are funded by the ACT government.

**Criteria 2: Models of care that include formalised integrated whole-of-life care pathways, care coordination, 24-hour service and support to transition from paediatric to adult services.**

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | **Partially meets criteria through formal arrangements with other jurisdiction/s** | Partially meets criteria through service within own jurisdiction | Fully meets criteria |
|---|---|---|---|---|

Patients in the ACT do not have access to an integrated whole-of-life centre; however, they are linked to transition services through Westmead Hospital's paediatric and adult care.

### Criteria 3: Adequate levels of resourcing and infrastructure to support access to emerging therapies and technologies.

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | **Partially meets criteria through formal arrangements with other jurisdiction/s** | Partially meets criteria through service within own jurisdiction | Fully meets criteria |
|---|---|---|---|---|

Patients in the ACT have access to clinical trials supported by services in NSW, providing patients with early access to new treatments. However, distance may make it harder to participate and there is inadequate resourcing for services to take up all available opportunities or to integrate these new therapies into care. As new therapies become available, these resource constraints will become more acute, and the lack of local services are even more limiting for patients in the ACT.

### Criteria 4: Well established training pathways and succession planning to ensure services are robust and sustainable.

| **Does not meet criteria** | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | Fully meets criteria |
|---|---|---|---|---|

# Australian Capital Territory

**SERVICE AREA**

Australian Capital Territory – Supported by New South Wales

## Staff Profile

**No dedicated specialised metabolic service.**

## Infant Care

NSW's Newborn Screening Program covers the ACT, with approximately 7,000 babies screened per year.

## Paediatric Care

Paediatric metabolic patients are typically referred to the CHW.

## Arrangements

There is no longer an overarching formalised arrangement between ACT Health and NSW Health (historically there was some decades ago), but ACT provides some funding to NSW Hospitals for services. These arrangements depend on the condition.

## Access

Some ACT families routinely travel to Sydney for specialised care.

## Adult Care

Adults requiring specialised metabolic care are typically engaged with Westmead Hospital, Sydney.

Westmead runs some clinics in the ACT, funded by the ACT.

## Services

ACT endocrinologists and neurologists working out of The Canberra Hospital (and privately) manage rare metabolic patients with conditions that are endocrine or neurological in nature.

## Gaps

4 out of 8 adult patients surveyed in this research and living in the ACT were not engaged with specialist metabolic services at all.

## Victoria

There are an estimated 3,340 people living with rare metabolic conditions in VIC. VIC is serviced by well-established specialised metabolic clinical and diagnostic services that also service patients across TAS.

Clinical specialist centres include the Metabolic Diseases Unit at Royal Melbourne Hospital (RMH) and the Department of Metabolic Medicines at the Royal Children's Hospital (RCH).

Diagnostic services include Victorian Clinical Genetics Services (VCGS), which is a subsidiary of the Murdoch Children's Research Institute, RCH.

However, there remain gaps in service coverage and 41% of patients surveyed in VIC reported that their needs were not being fully met. It should be noted that the assessment below is done without a detailed response from the service at the RMH.

### How does VIC measure up to the criteria?

**Criteria 1: Timely and local access to multidisciplinary services that include specialist metabolic clinicians, diagnostics, metabolic dietitians, metabolic nurses, genetic counsellors, clinical trial staff, allied health, mental health, disability support and social workers.**

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

The main specialist centres for rare metabolic conditions in VIC provide access to MDTs; however, limitations are reported across all roles at the RCH due to lack of staffing. Importantly, only 25% of Victorian patients surveyed who access care in specialist centres also had access to specialist dietitian services. This is compared to 42% in NSW. Issues also exist around the extent of outreach services, with no funding to cover such services for those states serviced by the RCH (including TAS), and limited access for regional VIC.

### Criteria 2: Models of care that include formalised integrated whole-of-life care pathways, care coordination, 24-hour service and support to transition from paediatric to adult services.

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

VIC does not have one integrated whole-of-life centre; however, it does have paediatric and adult care linked by well-developed transition services. Around 40% of patients receiving specialist metabolic care receive care coordination services, which are necessary to improve outcomes for patients with complex chronic conditions requiring multiple healthcare providers to effectively manage their care.

### Criteria 3: Adequate levels of resourcing and infrastructure to support access to emerging therapies and technologies.

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

Clinical trials are supported by services in VIC, providing patients with early access to new treatments. The strong linkages between services and co-located research facilities represent a particular strength in this state. However, there is inadequate resourcing for services to take up all available opportunities or integrate these new therapies into care. In particular, issues were raised with the sustainability of current data collection processes and limitations with addressing the expansion in known rare metabolic conditions.

### Criteria 4: Well established training pathways and succession planning to ensure services are robust and sustainable.

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | Fully meets criteria |
|---|---|---|---|---|

The lack of ongoing funding for metabolic trainees was highlighted, alongside issues with the lack of clear training and accreditation pathways for allied health specialists, including genetic counsellors.

DIAGNOSTIC SERVICE

## Victorian Clinical Genetics Services (VCGS) | Victoria

**SERVICE AREA**

VIC. Also provides services to all other states and territories.

### Arrangements

A subsidiary of the Murdoch Children's Research Institute, Royal Children's Hospital.

### Infant Care

**Diagnosed:**

- 23 infants in 2020–21
- 25 infants in 2019–20
- 29 infants in 2018–19

### Adult Care

**Diagnosed:**

- 14 adults in 2020–21
- 13 adults in 2019–20
- 13 adults in 2018–19

### Paediatric Care

**Diagnosed:**

- 18 children in 2020–21
- 17 children in 2019–20
- 13 children in 2018–19

### Services

- Diagnostic services
- Genetic counselling and screening

### Strengths

Close interactions with other laboratories and clinical specialist services.

### Challenges

- Facing future staff shortages: senior scientific staff with experience in genetics are ultra-rare due to waning career prospects
- Additional staff and equipment required for planned introduction of new NBS testing.

CLINICAL SERVICE

# Department of Metabolic Medicine

**The Royal Children's Hospital, Melbourne | Victoria**

**SERVICE AREA**

All of VIC, and supporting Tasmania.

## Staff Profile

- 2.5 FTE Metabolic clinicians
- 1.6 FTE Metabolic nurses
- 2.2 FTE Metabolic dietitians
- 0.6 FTE Metabolic social worker

## Infant Care

**Diagnosed:**

- 32 infants in 2021 (YTD*)
- 28 infants in 2020
- 27 infants in 2019
- 32 infants in 2018

**Provided services to:**

- 136 infants in 2021 (YTD*)
- 140 infants in 2020
- 140 infants in 2019
- 136 infants in 2018

## Outpatient Clinics

- 1,860 in 2021 (YTD*)
- 1,890 in 2020
- 1,890 in 2019

## Vacancies

- 1 FTE Social worker

## Paediatric Care

**Diagnosed:**

- 13 children in 2021(YTD*)
- 24 children in 2020
- 21 children in 2019
- 18 children in 2018

**Provided services to:**

- 967 children in 2021 (YTD*)
- 883 children in 2020
- 865 children in 2019
- 789 children in 2018

## Services

Diagnostic; clinical care; care coordination; specialist dietitian services; genetic counselling; paediatrics; formal or informal transition programs for paediatric to adult care; research collaborations, including clinical trials; outpatient clinics; 24-hour on-call service linked to emergency departments; telehealth services; consultative advice to other Victorian/ Tasmanian health providers; newborn screening; and consultative services to NBS, metabolic laboratories and genetic services.

## Strengths

- Multidisciplinary; excellent access to diagnostic and screening laboratories.
- Royal Children's Hospital is a tertiary and quaternary hospital providing full range of surgical/ medical specialties.
- Shared campus with research, teaching and clinical services.

## Challenges

- Workforce limitations across all roles.
- Limitations in access to expensive diagnostic testing and expensive therapies and medications.
- Provision of services across 2 states with no funding for outreach services.
- Very limited access to clinicians experienced in metabolic disorders are available in regional services to support care.
- Limitation in sustainability of data collection and databases/ data registries with current workforce.
- Limitations in administrative support.
- Limitations in expanding conditions detected by newborn screening.

**Year to Date (YTD) – January to August 2021*

CLINICAL SERVICE

# The Metabolic Diseases Unit

**Royal Melbourne Hospital | Victoria**

**SERVICE AREA**

All of VIC. Also provides services to Tasmania.

## Services

Diagnostic; clinical care; care coordination; specialist dietitian services; genetic counselling; paediatric to adult care transition programs/assistance; research collaborations, including clinical care; outpatient clinics; outreach services to regional hospitals; 24-hour on-call service to emergency departments; and telehealth services.

## Strengths

- Close relationships with the RCH, VCGS, neuropsychiatry and geneticists.
- Diagnostic services in close proximity (within local area/ city).

## Challenges

- Lack of ongoing funding for metabolic trainees.
- COVID-19 pandemic impacted diagnostic testing turnaround times.

## Tasmania

With no dedicated specialist metabolic service, the estimated 270 people in TAS with rare metabolic conditions rely on diagnostic services from SA and specialist clinical services from VIC. The lack of funding for outreach programs means patients must travel to VIC for care or rely on telehealth services.

### How does TAS measure up to the criteria?

**Criteria 1: Timely and local access to multidisciplinary services that include specialist metabolic clinicians, diagnostics, metabolic dietitians, metabolic nurses, genetic counsellors, clinical trial staff, allied health, mental health, disability support and social workers.**

| Does not meet criteria | **Partially meets criteria through informal arrangements with other jurisdiction/s** | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | Fully meets criteria |
|---|---|---|---|---|

The lack of local metabolic disease specialists or associated services means patients in TAS are travelling to VIC for their care. While this provides access, there is a lack of formalised outreach services and locally based specialists, which puts a strain on patients, families, carers and specialists in other centres.

**Criteria 2: Models of care that include formalised integrated whole-of-life care pathways, care coordination, 24-hour service and support to transition from paediatric to adult services.**

| Does not meet criteria | **Partially meets criteria through informal arrangements with other jurisdiction/s** | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | Fully meets criteria |
|---|---|---|---|---|

In VIC, where most Tasmanian patients are managed, there is no integrated whole-of-life centre; however, paediatric and adult care are linked by well-developed transition services. Tasmanian patients also have access to VIC's 24-hour service.

### Criteria 3: Adequate levels of resourcing and infrastructure to support access to emerging therapies and technologies.


While there is potential for access to emerging therapies and technologies through VIC services, the lack of local services limits potential access for Tasmanian patients.

### Criteria 4: Well established training pathways and succession planning to ensure services are robust and sustainable.

# Tasmania

**SERVICE AREA**

TAS (Supported by Victoria)

## Staff Profile

**No dedicated specialised metabolic service.**

## Infant Care

Analysis of Tasmanian newborn samples is undertaken by an SA service. This creates a disconnect with VIC services that provide specialised metabolic care to patients in TAS.

## Paediatric Care

A small number of Tasmanian-based paediatricians are involved in the care of Tasmanian children with rare metabolic conditions.

Specialised metabolic care for Tasmanian infants and children is provided by the RCH, Melbourne.

## Arrangements

There are agreements in place between the Tasmanian and Victorian Governments, but this needs to be reviewed as they are not fit-for-purpose.

## Access

There is no funding in place for outreach clinics in TAS, so patients are flown to VIC on an individual basis for consultations.

## Adult Care

The Metabolic Diseases Unit at RMH provides services to Tasmanian adults with rare metabolic conditions.

Some adult patients may instead receive specialist services from other 'crossover' specialities (i.e. haematologists, nephrologists, neurologists, endocrinologists) based in Tasmanian hospitals.

## Services

The Tasmanian Clinical Genetics Service (TCGS) is a statewide service providing assessment, diagnosis, genetic testing, management advice, counselling and support for paediatric and adult individuals who have, or are at risk of having, a metabolic condition.

TCGS is a state-wide service based at the Royal Hobart Hospital with Genetics Clinics also held regularly at Launceston General Hospital and North West Regional Hospital, Mersey Campus, Latrobe.

Some inpatient care for metabolic patients occurs at Hobart or Launceston Hospitals. However, most patients with acute presentations are admitted to Victorian hospitals.

## Gaps

Communication gap between newborn screening results in SA and VIC, reliant on individual paediatricians communicating results.

Regular outreach clinics from Victorian services or incentivising Tasmanian physicians to undertake specialised metabolic training pathways would help address the current gaps in care.

## Queensland

There are an estimated 2,590 patients in QLD with rare metabolic conditions. The main specialist service for the state is the only whole of lifespan clinic in the country, which is co-located at the Queensland Children's and Mater Hospitals. This specialist service provides family focused, intergenerational and sibling care that is critical given the inherited nature of many rare metabolic conditions.

### How does QLD measure up to the criteria?

**Criteria 1: Timely and local access to multidisciplinary services that include specialist metabolic clinicians, diagnostics, metabolic dietitians, metabolic nurses, genetic counsellors, clinical trial staff, allied health, mental health, disability support and social workers.**

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

Patients in QLD have access to specialist services that provide multidisciplinary care through the Lifespan Metabolic Medicine Service, which acts as a one-stop-shop. However, clinical research and genetic counselling is not integrated within the service. There is also unmet need for speech pathologists, occupational therapists and physiotherapists within the service.

**Criteria 2: Models of care that include formalised integrated whole-of-life care pathways, care coordination, 24-hour service and support to transition from paediatric to adult services.**

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | **Fully meets criteria** |
|---|---|---|---|---|

The Lifespan Metabolic Medicine Service provides integrated whole-of-life care pathways, care coordination and a 24-hour on-call service linked to emergency departments. Patients seen regularly (once weekly) have regular team reviews to ensure optimal care planning.

### Criteria 3: Adequate levels of resourcing and infrastructure to support access to emerging therapies and technologies.

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

The Lifespan Metabolic Medicine Service can facilitate both adult and pediatric clinical trials; however, levels of staffing are not adequate to meet current or future needs. In particular, the lack of an integrated clinical research manager on staff is highlighted as a key challenge to building research and clinical trials into clinical care.

### Criteria 4: Well established training pathways and succession planning to ensure services are robust and sustainable.

| **Does not meet criteria** | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | Fully meets criteria |
|---|---|---|---|---|

DIAGNOSTIC SERVICE

# Pathology Queensland and Mater Pathology | Queensland

**SERVICE AREA**

All of QLD. Also provides services nationally to all other states and territories.

## Arrangements

Linked to QLD Lifespan Metabolic Medicine Service, with close working relationships across the clinical and diagnostic services.

Formalised arrangement in place for newborn screening of infants born in the northern part of NT.

## Infant Care

**Diagnosed:**

- 41 infants in 2020–21
- 49 infants in 2019–20
- 37 infants in 2018–19

Testing over 60,000 infants per year.

## Adult Care

**Diagnosed:**

Approximately 10 per year

**Tested:**

- 376 adults in 2020–21
- 350 adults in 2019–20
- 330 adults in 2018–19

## Paediatric Care

**Diagnosed:**

- 15 children in 2020–21
- 13 children in 2019–20
- 9 children in 2018–19

Testing approximately 540 children per year.

## Strengths

- Excellent team of dedicated scientists who are keen to provide an excellent service.
- There is a joint clinical/laboratory case presentation each week to ensure coordination and high-quality care.

## Services

Diagnostic services

## Challenges

- Staffing shortages—if a single lab member takes sick leave or holidays, the service faces significant pressure.
- A national shortage of biochemical genetic pathologists—a role that is now required to supervise all metabolic testing under new NPAAC guidelines.
- Succession planning due to retirement or imminent retirement of a lot of senior laboratory scientists.
- COVID-19 pandemic presented a new challenge—lab work cannot be done remotely, so a confirmed exposure site or case within the service threatens lab closure.
- A clear path for adding tests to the national newborn screening panel.

CLINICAL SERVICE

# Queensland Lifespan Metabolic Medicine Service

**Queensland Children's Hospital, Brisbane | Queensland**

**SERVICE AREA**

All of QLD. Also provides services to New South Wales and the Northern Territory.

## Staff Profile

- 2.9 FTE Metabolic clinicians
- 2 FTE Metabolic nurses
- 0.8 FTE Nurse practitioners
- 1.6 FTE Metabolic dietitians
- 1 FTE Social worker

## Infant Care

**Diagnosed:**

- 30 infants in 2020–21
- 25 infants in 2019–20
- 23 infants in 2018–19

Provided services to approximately 430 infants between 2018–2021.

## Telehealth

- Up until the COVID-19 pandemic, the clinic was doing about 10% telehealth.
- Since the beginning of the COVID-19 pandemic, more than 50% of services have become telehealth (during lockdowns it became 100% telehealth).

## Outpatient Clinics

- 3 adult clinics per week
- 9 paediatric clinics per week

## Paediatric Care

**Diagnosed:**

- 15 children in 2020–21
- 13 children in 2019–20
- 9 children in 2018–19

Provided services to approximately 1,400 children between 2018–2021.

## Adult Care

**Diagnosed:**

- 20 adults in 2020–21
- 18 adults in 2019–20
- 17 adults in 2018–19

Provided services to approximately 1,730 adults between 2018–2021.

## Services

Diagnostic services; clinical care; care coordination; specialise dietitian services; genetic counselling; paediatrics; formal transition programs for paediatric to adult care; outpatient clinics; outreach services to regional hospitals; clinical trials and research, 24-hour on-call service linked to emergency departments; and telehealth services

## Strengths

- Services delivered across the lifespan (family-focused with intergenerational and sibling care).
- All patients seen each week are subject to a team review, to ensure optimal care planning and support best clinical outcomes.
- Partnership with the diagnostic service, with systems (such as IT) linked to support regular communication between scientific and clinical staff.
- Diagnostic service staff attend weekly education meetings with clinical team.
- The service is engaged with research and participates in clinical trials.

## Challenges

- Current staffing inadequate to meet current and future demand.
- Insufficient budget for testing.
- The need to integrate a clinical research manager and a genetic counsellor in the service.
- Unmet need for integrated speech pathologists, occupational therapists and physiotherapists.

## South Australia

There are an estimated 885 patients in SA with rare metabolic conditions. The Metabolic Clinic operates across the Women's and Children's Hospital and the Royal Adelaide Hospital, where 480 active patients were registered in 2020.

The Metabolic Clinic also provides services to patients in the NT, where a lack of specialist care has led to tragic neonatal deaths.

### How does SA measure up to the criteria?

**Criteria 1: Timely and local access to multidisciplinary services that include specialist metabolic clinicians, diagnostics, metabolic dietitians, metabolic nurses, genetic counsellors, clinical trial staff, allied health, mental health, disability support and social workers.**

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

Patients in SA have access to a specialist service that provides multidisciplinary care, but the lack of resources, particularly for adult services, limits the amount of care available. In particular, there is not currently capacity for nursing support for all rare metabolic conditions.

**Criteria 2: Models of care that include formalised integrated whole-of-life care pathways, care coordination, 24-hour service and support to transition from paediatric to adult services.**

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

SA services provide a strong care coordination model, which provides patients with the support they require to navigate their healthcare journey. However, resourcing constraints undermine transition and other services, which require greater investment to meet the needs of a growing cohort of patients.

### Criteria 3: Adequate levels of resourcing and infrastructure to support access to emerging therapies and technologies.

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

While the SA service is involved in clinical trials, there is inadequate funding to manage the cost or workload of implementing new and emerging therapies.

### Criteria 4: Well established training pathways and succession planning to ensure services are robust and sustainable.

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | **Partially meets criteria through service within own jurisdiction** | Fully meets criteria |
|---|---|---|---|---|

There is capacity for some training, but not full time, and the SA service is heavily reliant on one or two key physicians, representing a risk to continuity.

DIAGNOSTIC SERVICE

## SA Pathology | South Australia

**SERVICE AREA**

SA, and supporting Tasmania and the Northern Territory.

### Infant Care

**Diagnosed:**

- Approximately 10 infants in each period of 2018–19, 2019–20 and 2020–21

Provides approximately 26,000 testing services each year.

### Paediatric Care

**Diagnosed:**

Approximately 12 children in each period of 2018–19, 2019–20 and 2020–21.

### Services

- Diagnostic services
- Biochemical genetics services
- Infant and paediatric services

### Challenges

Service is not adequately staffed or resourced.

CLINICAL SERVICE

# Metabolic Clinic

**Women's and Children's Hospital, Adelaide | South Australia**
**Royal Adelaide Hospital, Adelaide | South Australia**

**SERVICE AREA**

All of SA. Also supports the Northern Territory.

## Staff Profile

*(FTE for paediatric care only. No adult care FTE data acquired)*

- 1.4 FTE Metabolic clinicians
- 0.1 FTE Trainee medical officer
- 0.7 FTE Metabolic nurse consultant
- 0.5 FTE Metabolic dietitians

## Telehealth

Teleheath services were greatly increased in 2020–21 compared with previous years.

## Outpatient Clinics

Operated 208 clinics in each period of 2018–19, 2019–20 and 2020–21.

## Strengths

Multidisciplinary approach and care coordination model.

## Infant and Paediatric Care

**Active Patients:**

- 390 in 2018
- 400 in 2019
- 480 in 2020

**Diagnosed:**

- 10 children in 2020–21
- 8-9 children in 2019–20
- 8-9 children in 2018–19
- 8-10 infants in 2020–21
- 8 infants in 2019–20
- 7-8 infants in 2018–19

## Adult Care

- Diagnosed 2–3 adults in 2020–21
- Provided services to 40–50 adults 2020–21

## Services

Diagnostic services; clinical care; care coordination; specialist dietitian services; genetic counselling; paediatrics; formal/informal programs for transitioning from paediatric to adult care; research collaborations, including clinical trials; outpatient clinics; 24-hour on-call services linked to emergency departments; and telehealth services.

## Challenges

- Providing enough medical and nursing FTE to meet current and future demand—SA does not have capacity for nursing support for all rare metabolic conditions, only some.
- Monitoring the neurological outcomes of patients with neuropsychological testing.
- Managing cost and workload of implementing new therapies.
- Requirement for significantly greater resourcing for adult metabolic services as more paediatric patients survive and transition.
- Access to diagnostic services within the local area.

## Northern Territory

There are an estimated 120 patients in the NT with rare metabolic conditions, with no specialist services. People in the NT receive care from either SA or QLD; however, delays in access have had tragic outcomes, especially for newborn babies.

### How does NT measure up to the criteria?

**Criteria 1: Timely and local access to multidisciplinary services that include specialist metabolic clinicians, diagnostics, metabolic dietitians, metabolic nurses, genetic counsellors, clinical trial staff, allied health, mental health, disability support and social workers.**

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | Fully meets criteria |
|---|---|---|---|---|

A combination of remoteness and the lack of a dedicated service in the NT means people with rare metabolic conditions are not benefiting from local access to multidisciplinary services. There is also a lack of formal agreements in place to ensure access to clinical and diagnostic services from SA. Further impeding streamlined access to services, some newborn screening for babies in the NT is done in QLD.

A lack of funding for outreach clinics means a strong reliance on telehealth service with SA clinicians.

**Criteria 2: Models of care that include formalised integrated whole-of-life care pathways, care coordination, 24-hour service and support to transition from paediatric to adult services.**

| Does not meet criteria | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | Fully meets criteria |
|---|---|---|---|---|

Patients in the NT are seen in SA, QLD and NSW, which provide varying levels of integrated whole-of-life care, care coordination and a 24-hour on-call service linked to emergency departments. However, the lack of formal arrangements creates issues for pathways and is likely resulting in many Northern Territorians not receiving adequate care.

### Criteria 3: Adequate levels of resourcing and infrastructure to support access to emerging therapies and technologies.

| | | | | |
|---|---|---|---|---|
| **Does not meet criteria** | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | Fully meets criteria |

There is no local resourcing or infrastructure in the NT, which is a barrier to accessing emerging therapies. The extent to which there is access for patients in the NT to emerging therapies is related to services in other jurisdictions. People living with a rare metabolic condition in the NT are reliant on the knowledge of their clinicians around upcoming new treatments and communications between NT health services and those in other jurisdictions where new therapies are being trialled or accessed.

### Criteria 4: Well established training pathways and succession planning to ensure services are robust and sustainable.

| | | | | |
|---|---|---|---|---|
| **Does not meet criteria** | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | Fully meets criteria |

# Northern Territory

**SERVICE AREA**

NT, supported by South Australia and Queensland.

## Staff Profile

**No dedicated specialised metabolic service.**

## Infant and Paediatric Care

The Metabolic Clinic located in SA's Women's and Children's Hospital supports care for some infants and children from the NT.

## Adult Care

The Metabolic Clinic located in SA's Women's and Children's Hospital (but providing adult clinics from the Royal Adelaide Hospital) supports care for some adults from NT.

## Arrangements

SA provides specialised metabolic services to approximately 20 families in the NT. There is no formal arrangement in place between the NT and SA.

The informal arrangement originated from a link between Adelaide and Alice Springs, which extended out to cover Darwin. The support from SA has grown overtime but is completely unfunded.

QLD provides newborn screening services to the top-half of NT.

## Access

There is no funding in place for outreach clinics to NT, so there is a strong reliance on telehealth with SA clinicians.

## Services

Some inpatient care for metabolic patients occurs at Darwin or Alice Springs Hospitals. However, most patients with acute presentations are admitted to South Australian hospitals.

## Gaps

The lack of a formalised arrangement with SA means there are patients who receive either no specialised metabolic care or those who seek out care from another jurisdiction.

Of note, one adult from the NT surveyed in this research was engaged with the Department of Genetic Medicine, Westmead Hospital Sydney.

Regular formalised outreach clinics from SA clinical services would help address the current gaps in care.

## Western Australia

There are an estimated 1,330 patients in WA with rare metabolic conditions. There is no metabolic specialist service operating in WA; however, there are services offered for specific conditions, including the Fabry Clinic run by a nephrologist out of Royal Perth Hospital.

WA patients with other rare metabolic conditions currently rely on care from specialists outside of metabolic medicine including nephrologists, endocrinologists or neurologists, but there is a lack of formalised support arrangements with these other physicians. The only specialist dietitian in WA recently moved on, leaving patients in the state without this critical allied health support.

### How does WA measure up to the criteria?

**Criteria 1: Timely and local access to multidisciplinary services that include specialist metabolic clinicians, diagnostics, metabolic dietitians, metabolic nurses, genetic counsellors, clinical trial staff, allied health, mental health, disability support and social workers.**


While patients with Fabry disease are benefiting from multidisciplinary care, patients with other metabolic disorders are not receiving multidisciplinary care that includes metabolic clinicians, dietitians and nurses.

**Criteria 2: Models of care that include formalised integrated whole-of-life care pathways, care coordination, 24-hour service and support to transition from paediatric to adult services.**


While the Fabry Clinic provides these services to 70 patients in the state, people with other conditions are not currently benefiting from these whole-of-life pathways, or care coordination services.

### Criteria 3: Adequate levels of resourcing and infrastructure to support access to emerging therapies and technologies.

| | | | | |
|---|---|---|---|---|
| **Does not meet criteria** | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | Fully meets criteria |

The lack of a metabolic service in the state is hampering access to clinical trials and will impact the ability of patients to access emerging therapies in the years ahead. There are also no formal arrangements with other states, which puts serious limitations on knowledge of and access to new treatments and novel technologies through clinical trials that might be available in other judications.

### Criteria 4: Well established training pathways and succession planning to ensure services are robust and sustainable.

| | | | | |
|---|---|---|---|---|
| **Does not meet criteria** | Partially meets criteria through *informal* arrangements with other jurisdiction/s | Partially meets criteria through *formal* arrangements with other jurisdiction/s | Partially meets criteria through service within own jurisdiction | Fully meets criteria |

# Western Australia

**SERVICE AREA**

WA (Supported by other jurisdictions)

## Staff Profile

Some coverage of specialised metabolic services.

## Infant and Paediatric Care

The Metabolic Medicine department at Perth Children's Hospital provides diagnostic services and care to children with genetic metabolic disorders and manages follow-up care for newborn babies diagnosed through the WA Newborn Screening Program.

## Adult Care

Adult patients are typically receiving services from other WA specialists, such as clinical geneticists, neurologists, endocrinologists and nephrologists.

Some adults in WA are still engaged with paediatric care.

## Access

Access to services in WA is patchy and varies significantly depending on the rare condition.

## Arrangements

Previous attempts to establish a dedicated metabolic service in WA were hampered by sustainability issues associated with only one metabolic clinician residing in the state.

WA has previously reached out to South Australia's Metabolic Clinic to provide two clinics per year for management of complex patients, but this was not implemented.

## Gaps

WA has a lack of dedicated state-based specialised metabolic services, and any formalised supporting arrangements from other jurisdictions.

Current coverage is also at risk with the upcoming retirement of senior specialist roles. The only WA-based dietitian with specialist experience treating rare metabolic conditions has also recently moved on.

WA clinicians seek advice across jurisdictions. NSW, Victoria, SA and other jurisdictions have attempted to assist WA clinicians in many ways

## Services

WA has limited dedicated specialist metabolic services, with some coverage provided by other WA specialists such as clinical geneticists, neurologists, endocrinologists and nephrologists. These specialists may dedicate a portion of their time and services to metabolic patients (i.e. 0.2 or 0.3 FTE).

WA has a well-developed Fabry service run out of Royal Perth Hospital by a nephrologist.

The Fabry Clinic services about 70 Fabry patients. There is a quarterly afternoon multidisciplinary clinic for complex patients—which includes another nephrologist, a cardiologist, neurologist, geneticist, pain specialist and psychiatrist. The clinic offers a home-support nurse, dedicated home and hospital infusion nurses and another nursing role that manages registry data entry.

PathWest, WA's genetics and laboratory service, is involved in biochemical genetics testing, including tests for rare metabolic conditions.

Genetic Services Western Australia–a state-wide clinical genetic service–does not provide metabolic services.

# SUMMARY

This White Paper has highlighted the breadth and scope of rare metabolic services being provided around Australia through the combined efforts of committed clinicians, hospital staff, patient groups, families, allied health providers and many others. An emerging area of medicine, Australian specialist metabolic services are defined by the people committed to ensuring that patients with rare metabolic conditions receive the best care possible. However, critical shortages in professional expertise and resources are hampering even the very best efforts of those involved. There was resounding frustration across all the voices that contributed to this research and a palatable desire for change.

Through wide consultation and input from rare metabolic patients and service providers, including hundreds of individuals, peak organisations and metabolic specialist centres, we were able to comprehensively capture strengths and gaps across the rare metabolic workforce in Australia.

We found that the role and need for a specialised rare metabolic workforce is not well understood by medical peak bodies and administrators, making it difficult to address the serious issues facing the workforce, patients and their families. Therefore, to facilitate the reforms and investments needed across the workforce, greater recognition of the specialty of metabolic medicine is paramount.

Patients and healthcare professionals flagged the need for more formalised care pathways, better models of care and individualised care plans in line with those already in use for complex chronic health conditions. Indeed, many of the gaps in the metabolic workforce can be filled by leveraging and modifying resources, infrastructure and care pathways that already exist across other more developed medical services. However, some aspects of care for patients with metabolic conditions require a nuanced approach informed by specialist metabolic expertise. Currently, this is not always possible all throughout Australia due to inadequate funding and care models. Metabolic services and metabolic healthcare staff are being forced to come up with creative ways to get funding to meet the basic needs of patient care. This is far from ideal; it is leaving healthcare staff burnt out and many patients receiving suboptimal care at times.

Across some services, there are critical workforce shortages that cannot easily be rectified due to poor alignment between 'activity-based' hospital funding models and the complex, multidisciplinary care that rare metabolic patients require. Rare metabolic diseases are highly complex chronic conditions that require new models of care, staffing and investment to ensure Australians can access quality and timely care.

There is also significant variation in the composition and maturity of metabolic services both *within* and *across* states and territories, and across rare metabolic conditions. This means the standard of care received by patients is dependent on their diagnosis and postcode, highlighting serious inequities that must be urgently addressed.

Rare metabolic patients are deteriorating without opportunities to benefit from new treatments and technologies for their condition. According to our research, Australian specialist metabolic services are almost universally not sufficiently resourced to contribute to innovation. This is a major barrier to the development and trialling of emerging therapies and technologies for rare metabolic conditions in Australia. As highlighted in the Action Plan, "for many people living with a rare disease, participation in a clinical trial may be the only way to access treatment."[1]

Overall, the findings show that Australia's rare metabolic health care does not compare well to international best practice. However international models, like the ERN and MetabERN, do provide a potential way forward for vital systemic change. A move towards this network type approach with established criteria for rare disease CoE also aligns with the Action Plan's foundation principles: person-centred; and sustainable systems and workforce; as well the critical enabler: state, national and international partnerships.[32]
The Action Plan specifically outlines the strengths of CoE and ERNs as international exemplars that should be customised for the Australian context.[1] The White Paper findings certainly support MetabERN as a model that could be potentially customised for Australia.

# NEXT STEPS FOR A STRENGTHENED METABOLIC WORKFORCE IN AUSTRALIA

This White Paper highlights the current state of play across the rare metabolic workforce in Australia. Australia has an urgent need to invest in this critical workforce and improve outcomes and access to health care for the estimated 12,700 Australians living with a rare metabolic condition.

The key findings presented herein, support the need for further strategic work towards a more sustainable metabolic workforce to provide the best quality care and outcomes for patients living with rare metabolic conditions in Australia today and into the future.

Following this White Paper, RVA will:

- Communicate these findings to all key stakeholders;
- Develop draft strategic actions in response to the key findings;
- Engage and collaborate with Commonwealth, state and territory governments, hospital administrators and ASIEM to review draft strategic actions and develop effective and sustainable solutions; and
- Finalise strategic actions to progress much needed change in each jurisdiction that is aligned with international best practice.

The White Paper findings should be further leveraged to respond to broader rare disease workforce issues in Australia. Continued work in this area should be prioritised by all jurisdictions to respond to the Australian Government's Action Plan in which workforce is a foundation principle and priority. The establishment of an Australian Reference Network (ARN) customised from the ERN model is one way to address the geographical spread and inherent scarcity of rare disease patients, and specialist expertise that already exist in Australia. Bringing together the best knowledge, expertise and resources from across Australia can ensure that all Australian rare disease patients have access to the best available care.

The research for this White Paper has also strengthened relationships with international metabolic leaders who have encouraged Australia to contribute to the ERN model. This type of international collaboration aligns with the Action Plan, which highlights the importance of state, national and international partnerships. This work can, and needs to, start now.

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# APPENDIX

## List of abbreviations

**ACT:** Australian Capital Territory

**APEC:** Asia Pacific Economic Corporation

**ARN:** Australian Reference Network

**ASIEM:** Australasian Society for Inborn Errors of Metabolism

**CHW:** Children's Hospital Westmead

**CoE:** Centre of Excellence

**COVID-19:** Coronavirus (SARS-CoV-2) disease

**ePAG:** European Patient Advocacy Board

**ERN:** European Reference Network

**FTE:** Full Time Equivalent

**GP:** General Practitioner

**GSAC:** Genetic Services Advisory Committee

**HGSA:** Human Genetic Society of Australasia

**HHS:** Hospital and Health Services

**IEM:** Inborn Errors of Metabolism

**IMD:** Inherited Metabolic Disorders

**IT:** Information Technology

**LSDP:** Life Saving Drugs Program

**MBS:** Medicare Benefits Schedule

**MDT:** Multidisciplinary Teams

**MetabERN:** European Reference Network for Hereditary Metabolic Disorders

**MTHFR:** Methylenetetrahydro-folate reductase

**NBS:** Newborn Bloodspot Screening

**NDIS:** National Disability Insurance Scheme

**NPAAC:** The National Pathology Accreditation Advisory Council

**NSW:** New South Wales

**NT:** Northern Territory

**QCH:** Queensland Children's Hospital

**QLD:** Queensland

**RACP:** Royal Australasian College of Physicians

**RCH:** Royal Children's Hospital

**RMH:** Royal Melbourne Hospital

**RVA:** Rare Voices Australia

**SA:** South Australia

**SCHN:** Sydney Children's Hospital's Network

**SOPs:** Standard Operating Procedures

**SSIEM:** Society for the Study of Inborn Errors of Metabolism

**TCGS:** Tasmanian Clinical Genetics Service

**The Action Plan:** The National Strategic Action Plan for Rare Diseases

**The White Paper:** Rare Metabolic Diseases White Paper: Towards a Strengthened Rare Disease Workforce for Australia

**UK:** United Kingdom

**VCGS:** Victorian Clinical Genetics Services

**VIC:** Victoria

**WA:** Western Australia

**YTD:** Year-to-Date

RARE METABOLIC DISEASE WORKFORCE WHITE PAPER

# Towards a Strengthened Rare Disease Workforce for Australia

February 2022