New South Wales

Special Commission of Inquiry into Healthcare Funding

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Name: Australian Society for Inborn Errors of Metabolism

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Australasian Society of
Inborn Errors of Metabolism
A special interest group of the
Human Genetics Society of Australasia

30th October 2023

Parliamentary Special Commission of Enquiry into Health Funding

Dear Mr Richard Beasley

Metabolic Genetic services in NSW

ASIEM is the peak body for metabolic genetic services – a specialty that looks after and manages rare metabolic conditions that have a genetic basis. The specialty is recognised as a sub-specialty of clinical genetics by RACP but has not been registered as a sub-specialty by AHPRA. Our specialty differs from clinical genetics in that there is active treatment and management of patients that are identified by molecular genetic or biochemical genetic testing. Whilst individually rare with incidences from 1 in 7,000 to 1 less than 1 in a million, collectively there are an estimated 4,000 individuals in NSW with a rare metabolic disorder.

The specialty of metabolic genetics is well recognised internationally including throughout Europe (European – metabolic reference network), Canada, New Zealand, (national metabolic service), UK (nationally commissioned), but is relatively under-developed in Australia. With the huge range of rare genetic disorders, there are a huge range of commercially sponsored clinical trials that could be brought to New South Wales and Australia, providing patients access to contemporary treatments. There is an opportunity with small infrastructure investments particularly in ultra-specialist dietetic services, nursing, genetic counselling and social work investment to optimise treatment. Generic rare disease services could provide with a broader suite of service including speech pathology, physiotherapy, psychology, neuro-psychology assessments. Often these allied health teams need the guidance of the specialist service (such as from a nurse practitioner) to target an appropriate assessment for the individual optimising person centered care. Two reports are attached to provide guidance on how these conditions have been assessed nationally – the RVA white paper and the RVA metabolic workforce strategy.

Since its inception in 1991, ASIEM has overseen the management of newborn bloodspot screening (NBS) in Australia. ASIEM incorporated the screening laboratory, confirmatory biochemical genetics laboratories and treating physicians in developing infrastructure for the newborn screening programme. Before 1998, approximately five conditions were screened in all newborns in New South Wales. The introduction of tandem mass spectrometry in 1998 increased the number of conditions to approximately 50 - all of these were rare metabolic genetic disorders. Some of these led to clinical symptomology in the first week of life, necessitating urgent intervention by the metabolic genetic team. There was no concomitant increase in funding for the clinical service managing these patients yet there was undoubtedly an improvement in survival and quality of life. NSW was world leading when it implemented this technology, but the clinical outcomes have not been systematically evaluated over the 25 years of this programme. In 2022, the federal government has led harmonisation and expansion of NBS but the state based clinical systems need development.

NSW Health has been leading the genomic revolution in Australia. This has now identified many metabolic genetic conditions that also would not have been identified previously. This has led to flow on effects of treatment by the metabolic genetic service. Previously, the diagnosis may not have been suspected or the patient may have died before testing could have commenced. Often the clinical team has no experience of the condition and have had to pivot to novel therapeutic modalities, by consultation with international leaders. Therefore, the paediatric service has had to intervene in situations that it didn't previously and provide bespoke treatment for conditions such as Wolman disease, CACT deficiency and HMG co A Lyase deficiency (all now in published literature). The adult metabolic service was first created in 2011 and has seen an exponential rise in demand due to novel diagnostics as

well as management of new long-term surviving children. This situation has been faced by services such as the adult cystic fibrosis service and adult congenital heart disease services and applies equally to metabolic genetic disorders.

Both the adult and the paediatric service operate as statewide services providing consultative interactions with referrals from all peripheral settings. Often the services, co-assist with management in other hospitals and health care settings across NSW. Periodically, both children and adults need to be retrieved to the adult hospital at Westmead, The Children's Hospital at Westmead (SCHN – Westmead) and Sydney Children's Hospital (SCHN-Randwick) where specialist tertiary level metabolic genetic services need to be delivered. The activity across these different services in New South Wales are not reflected in the medical records of Westmead Hospital or SCHN (Randwick and Westmead). Because the individual hospital does not see the activity, funding is not enhanced and indeed at times of stress in the system, staff are seconded to other services. This is further compromised by the fact that there is no AHPRA recognition of the specialty leading to activity being accredited to departments such as intensive care, emergency departments, clinical genetics, endocrinology, neurology or general paediatrics. These issues are reflected in “the rare metabolic workforce white paper,” published by Rare Voices Australia in 2022.

The issues faced by the metabolic genetic workforce in NSW will be compounded by further expansion of newborn screening services to metabolic genetic conditions proposed by the federal government. Diagnosis of these disorders in the newborn baby will affect the wider family (by obligatory cascade genetic screening) and will lead to rising demand for both paediatric and adult metabolic genetic services. It is difficult to predict what that demand will be.

Within the terms of reference to the enquiry,

- There are significant workforce shortages across the two metabolic genetic in NSW (paediatric and adult). Both operate as statewide services.
- The services evade accurate assessment of service delivery by current metrics.
- The therapeutic delivery of precision medicine to rare metabolic genetic disorders needs to be considered in a timely manner, by services equipped with a multi-disciplinary team to do so.
- Metabolic Genetic services have experience in rapidly assessing biochemical and genetic data to provide timely treatment to many rare disorders.
- Delivery of treatment could occur by a well-resourced hub and spoke model, in order to deliver person centred care.
- Services could adopt models from better resourced overseas services such as Japan, Canada, The Netherlands, Germany or UK and implement them in an Australian context.
- The patient populations of these services are expanding as genomics and NBS expands, and service delivery planning needs to occur imminently.

Best wishes
Yours sincerely

Conjoint A/Prof Kaustuv Bhattacharya UNSW
Senior Staff Physician – Genetic Metabolic Disorders Service SCHN – (Westmead and Randwick)
President - ASIEM

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EQUITY ECONOMICS

National Strategy for Australia's Rare Metabolic Disease Workforce

Rare Voices Australia and
Equity Economics and Development Partners

February 2023

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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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.

www.equityeconomics.com.au

Project Steering Committee

RVA wishes to acknowledge the following individuals who contributed their time and expert advice to the development of the *Rare Metabolic Disease Workforce White Paper: Towards a Strengthened Rare Disease Workforce for Australia* (White Paper) and this National Strategy for Australia's Rare Metabolic Disease Workforce (Strategy).

- Professor Gareth Baynam, Medical Director, Rare Care Centre, Perth Children's Hospital
- Dr Kaustuv Bhattacharya, Chair of the Australasian Society for Inborn Errors of Metabolism (ASIEM); Metabolic Paediatrician, Genetic Metabolic Disorders Service Children's Hospital Westmead, Sydney
- Dr Drago Bratkovic, Head of the Metabolic Unit at the Women's and Children's Hospital, Adelaide
- Dr Sarah Donoghue, Metabolic Physician, Royal Children's Hospital Melbourne; Victorian Clinical Genetic Services, Murdoch Children's Research Institute, Melbourne
- Dr Maureen Evans (PhD), Director & Senior Dietitian, Department of Metabolic Medicine, Royal Children's Hospital Melbourne
- Ms Anita Inwood, Metabolic Nurse Practitioner, Service Director, Queensland Lifespan Metabolic Medicine Service, Queensland Children's Hospital and Mater Adults Hospital; Associate Lecturer, School of Nursing, Midwifery and Social Work
- Dr Heidi Peters, Department Metabolic Medicine, Royal Children's Hospital Melbourne; Department Paediatrics, University of Melbourne
- Dr Michel Tchan, Department of Genetic Medicine, Westmead Hospital, Sydney; Sydney Medical School, University of Sydney
- Associate Professor Veronica Wiley, Former Principal Scientist at New South Wales Newborn Screening, Children's Hospital, Westmead, Sydney

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Authors

Dr Falak Helwani

Falak is the Research and Evaluation Manager at RVA. She is a former molecular cell biologist with lived experience in rare disease. Falak has a PhD in cell biology from the Institute for Molecular Bioscience at the University of Queensland and has co-authored several peer-reviewed papers. She also has 6 years' postdoctoral experience in stem cell research at the Mater Medical Research Institute in Queensland.

Dr Angela Jackson

Angela is an economist with expertise across health, disability, gender and fiscal policy. She was Deputy Chief of Staff to Australia's Finance Minister, the Hon Lindsay Tanner MP, during the Global Financial Crisis. Angela is currently a member of the Victorian National Heart Foundation Advisory Board, part time Commissioner at the Commonwealth Grants Commission and Deputy National Chair of the Women in Economics Network.

Lauren Geatches

Lauren is Senior Manager of Social Policy at Equity Economics and is an experienced health and social policy analyst with nearing a decade of experience working for the Australian Government Department of Health and consulting for private and not-for-profit organisations. Lauren has previously led key reforms across the rural health workforce and primary care, and now works to address social and health inequities through applied research, most recently in the rare diseases space. Lauren holds a Bachelor of Social Science, majoring in Sociology and Anthropology, from the University of Newcastle, and an Excellence in Sociology Award from the Australian Sociological Association.

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EXECUTIVE SUMMARY

A national rare disease workforce strategy that responds to current and future demands, including the impact of genomics, is a key priority of the Australian Government's National Strategic Action Plan for Rare Diseases (the Action Plan).[1] Presented herein, is a workforce strategy for a specific, but significant, subset of the rare disease workforce — the rare metabolic disease workforce. This Strategy responds to workforce challenges evidenced by the Rare Metabolic Disease Workforce White Paper: Towards a Strengthened Rare Disease Workforce for Australia (White Paper)[2] — a pilot study that informs broader workforce issues in caring for Australians living with a rare disease.

An estimated 12,700 Australians live with a rare metabolic disease. Rare metabolic diseases include over 1,800 known rare disorders, which result from defects in the breaking down and conversion of carbohydrates, proteins and fats.[3] They are a highly heterogenous group of complex, multisystemic conditions and knowledge about individual metabolic diseases is limited by low patient numbers. The variability and complexity of rare metabolic diseases, together with the rarity of each condition, places overwhelming medical and social burden on families.[4] This complexity means patients require care from a variety of specialists.

The White Paper gathered expertise and experience from patients, carers, families healthcare professionals and industry.[2] Despite growing demand due to improvements in diagnosis and more rare metabolic patients surviving into adulthood, specialist metabolic services in Australia are under-resourced, and staff are struggling to keep up with current demand — this is negatively impacting patients.

The White Paper highlighted the breadth and scope of rare metabolic services 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 the best efforts of those involved. There was a resounding frustration across all the voices that contributed to this research and a palpable desire for change.

Since the launch of the White Paper, RVA has engaged with several rare disease patient group leaders, members of government, state health departments and professional medical bodies in one-on-one meetings and forums to discuss the best way forward.

STAKEHOLDER MEETINGS TO FEBRUARY 2023

Human Genetics Society of Australasia (HGSA)
Australasian Society for Inborn Errors of Metabolism (ASIEM)
Key rare metabolic disease patient advocate group leaders
New South Wales (NSW) State Government
Western Australian (WA) State Government and Health Department
Queensland (QLD) State Government
Victorian (VIC) State Government and Health Department
South Australian (SA) State Government and Health Department
Tasmanian (TAS) State Government

In response to the White Paper findings, and these subsequent consultations with the sector, the following report outlines a national strategy for improving the health and wellbeing outcomes for Australians living with a rare metabolic disease. It provides a framework that prioritises addressing gaps and draws on key strengths of the rare metabolic disease workforce in Australia. As all stakeholders work to implement and adopt this Strategy, it is important to remember that rare metabolic care varies across states and territories, and responses will need to be customised based on specific needs in each jurisdiction.

GOALS, RECOMMENDATIONS AND PRIORITY ACTIONS

Below are goals, recommendations and priority actions for rare metabolic disease workforce reform, which directly respond to key findings in the White Paper.

GOAL 1: SUSTAINABLE SYSTEMS AND WORKFORCE

Why is this important?

One of the most alarming revelations of the White Paper was the high level of unmet need among Australians living with a rare metabolic disease.[2] There were many examples of how far services and individual healthcare practitioners are presently stretched. All stakeholders agreed there is insufficient workforce capacity to meet current and future needs. Even the largest, most successful specialist metabolic services in Australia are finding it impossible to manage current patient loads. And demand continues to grow.

There is an urgent need to address the critical workforce shortages in existing specialist metabolic services across diagnostic and clinical settings, disability, allied health and mental health. Immediate action and additional resources are required to address these shortages, together with longer-term reforms that support the supply and distribution of a sustainable pipeline of suitably trained and experienced specialised metabolic healthcare practitioners. Such action aligns with the vision of the National Medical Workforce Strategy 2021–2023, to ensure a sustainable medical workforce that delivers high-quality care.[7]

Even Australia's best metabolic service model, remains unsustainably funded. Staff to patient ratios in Australian specialist metabolic services are much lower than those in the Netherlands, United Kingdom, Canada and Ireland. These jurisdictions have prospectively invested in the workforce to meet the challenge of novel genomic therapeutics for rare genetic metabolic diseases. In line with this international direction, annual service planning for future forecasting in Australian metabolic services is vital.

As part of this service planning, metabolic services should include regular reviews of the hours of work and rotations of clinicians that consider and mitigate the risks outlined in the Australian Medical Association's National Code of Practice - Hours of Work, Shiftwork and Rostering for Hospital Doctors.[17]

Current workforce shortages are driven not only by the lack of succession planning to address the impending retirement of key personnel, but also by the lack of funded positions. It is difficult to determine the overall adequacy of current Australian rare metabolic workforce staff to patient ratios in each jurisdiction, because there is no evidence to support a benchmark for effective ratios. Finding the right balance is not straightforward given the number and complexity of rare metabolic conditions and the variability in care requirements. However, this knowledge together with an understanding of the minimum core roles required to establish a metabolic service (see Priority Action 3.3.1), would support strategic and timely improvements in workforce shortages.

Since over 99% of parents consent to Newborn Bloodspot Screening at birth, national benchmarks for the minimum full-time equivalent across the core roles in a metabolic service could be based on the number of registered births in Australia, the prevalence of rare metabolic diseases and international best practice.

To future-proof the healthcare needs of these patients, their families and carers, modeling would also need to appropriately consider the complexities of rapid advances in technology and the availability of new treatments resulting in increased survival into adulthood.

Building an effective metabolic workforce should not depend solely on consultant metabolic doctors. An efficient use of specialist metabolic nursing, dietetics and junior medical staff would enable more patients to be seen in a metabolic service. Formal addition of administrative support would further enable all specialist metabolic clinicians to work at the top of their scope of practice.

Rare metabolic disease patient advocate leaders consulted for the White Paper cited the mental health burden on their communities as a major concern.[2] They also highlighted gaps in access to, and integration of, mental health and disability support services for people living with rare metabolic diseases.[2] The psychological and psychosocial impact of rare metabolic conditions is well documented, and a large proportion of rare metabolic patients require access to allied health and disability support services.[3] Building these supports into specialist metabolic service delivery is necessary for a holistic approach to care. It is vital to plan for inevitable increases in demand across all facets of specialist metabolic care, from access to diagnosis to longitudinal multidisciplinary care with formal links to mental health, allied health and disability supports.

White Paper Key Finding 4 pointed to fundamental flaws in the logic underpinning current funding of roles in specialist metabolic services within tertiary hospitals across Australia.[2] These flaws centred on the failure of activity-based funding models to capture the true activity associated with delivering ongoing, complex and coordinated multidisciplinary care to people living with rare metabolic conditions. Current overly simplistic methods for calculating the required workforce capacity or service load do not consider the savings from tasks that lead to preventable hospitalisations. Nor do these methods account for the distribution of benefits and savings beyond Local Health District borders and, in some cases, across state and territory borders. Any strategy for a sustainable rare metabolic workforce must recognise these benefits, address the complexities of caring for people living with a rare metabolic disease, and shift towards more fit-for-purpose funding models.

RECOMMENDATIONS AND PRIORITY ACTIONS FOR SUSTAINABLE SYSTEMS AND WORKFORCE

Recommendation 1.1

Urgently respond to critical funding shortages to increase the capacity and sustainability of existing services for paediatric and adult patients.

Priority Action 1.1.1

Ensure there is ongoing sustainable funding for existing services.

Recommendation 1.2

Urgently address critical workforce shortages.

Priority Action 1.2.1

Address gaps in critical areas of care including:

- Metabolic clinicians (adult and paediatric), dietitians, nurse specialists/coordinators, nurse practitioners, neuropsychologists, social workers and psychologists, clinical trial coordinators and nurses, and research managers;
- Mental health support, including adequate psychosocial services to meet clinical trial site requirements;
- Disability support to facilitate linkages to the National Disability Insurance Scheme (NDIS) and My Aged Care;
- Allied health, including genetic counselling.

Priority Action 1.2.2

Formalise and resource existing interjurisdictional arrangements, and progress work to build new connections to ensure all Australians have access to specialist metabolic care in their local jurisdiction.

Recommendation 1.3

Adopt a revised funding framework for both clinical and diagnostic specialist metabolic services that adequately responds to and captures:

- All service activities, including but limited to care management/coordination and data collection;
- Unserviced patients; and
- Underserviced patients.

Recommendation 1.4

Through a multifaceted approach, modify or extend the current activity-based funding models to better respond to the complexity and resource intensity of tasks associated with rare metabolic care.

Priority Action 1.4.1

Improve utilisation of activity-based funding models. E.g education to staff to optimise the use of activity-based funding codes.

Priority Action 1.4.2

Ensure the funding model has the flexibility to adequately recognise and support multidisciplinary teams in metabolic services. E.g. dedicated funding codes for multidisciplinary team activities, metabolic medicine outpatient clinics and specialist metabolic dietitians, as well as higher activity plans revised through annual service planning.

Recommendation 1.5

Ensure adequate, responsive and sustainable staff to patient ratios across all roles in a metabolic service.

Priority Action 1.5.1

Develop a flexible model for staff to patient ratios across all roles in a metabolic service (see minimum core roles under Goal 3) that considers:

- The increasing number of known rare metabolic diseases;
- The expansion of metabolic conditions screened at birth, considering advances in genomic newborn screening;
- Improved outcomes as a result of novel medicines and new technologies that lead to increased survival into adulthood and the increasing need for long-term complex care;
- Increasing recognition of adult-onset metabolic diseases;
- The cumulative public health burden of metabolic diseases – long-term holistic needs of families and carers.

Recommendation 1.6

Enable funding models to expand services to respond to future needs. Existing service funding growth should be indexed to projected growth in demand and changing requirements of the patient population.

Priority Action 1.6.1

Regularly review the number of funded positions based on evidence-based growth in patient numbers and life expectancy — taking into account the impact of genomics — to ensure future needs are met.

GOAL 2: CONNECTED AND COORDINATED CARE

Why is this important?

Care coordination is critical for supporting the complex needs of people living with a rare metabolic disease to access multidisciplinary care.[2]

Only 40% of specialist metabolic services surveyed for the White Paper have a lead position for care coordination, and most are not separately funded.

In the White Paper, many fundamental components of delivering multidisciplinary, coordinated care—such as cohesive teamwork, strong collaboration and close linkages or relationships with other services—were reported as existing strengths by over 80% of specialised metabolic clinical and diagnostic services.[2] Nonetheless, only 40% of specialist metabolic services surveyed for the White Paper have a lead position for care coordination, and most are not separately funded.[3] There were several instances of nurses taking on coordinator roles without recognition or targeted funding.

Currently, most efforts to connect and coordinate the care of people living with a rare metabolic disease across disciplines and services rely on clinicians' awareness of the right services for referrals, as well as good communication, and even goodwill. This cobbled together, patchwork of informal care arrangements is leading to significant variations and inconsistencies in the types and mix of care received by patients living with the same rare metabolic conditions across Australia.

The development of diagnostic and clinical pathways into and through care would tangibly improve patient care and patient experience of care. This includes the wider use of individualised care plans. Individualised care plans are one mechanism to facilitate connected and coordinated care. They have long been used for the coordinated management of more common complex or chronic health conditions. The benefits of individualised care plans are well established, and their limited use for rare metabolic conditions was consistently raised by patients and clinicians consulted on the White Paper. Leveraging this established way of working could address the challenges faced by patients in understanding and navigating their care—particularly those without local access to a specialist metabolic service. While creating and maintaining these plans requires significant time and resources for clinical services, they are likely to be offset by more efficient and effective communication between multidisciplinary care teams, patients, families and carers.

In addition to the need for disease management criteria highlighted in Priority Action 4.2.2, defined and nuanced models of care (MoC) offer a mechanism to embed consistency and clarity of roles. This is acknowledged more broadly in the National Medical Workforce Strategy 2021—2031, which highlights the need to adapt to and better support new MoC that address workforce shortages in flexible and innovative ways.[7] This policy framework, further emphasises the need to develop a medical workforce with sufficient breadth, reach and adaptability to initiate and respond with agility to opportunities to develop and implement more effective MoC, particularly as new technologies become available.[7] Consistent MoC that include a holistic approach to multidisciplinary models and team-based care, together with robust paediatric to adult transition services, would offer a nationally consistent overarching framework for connected and coordinated management of rare metabolic conditions.

RECOMMENDATIONS AND PRIORITY ACTIONS FOR CONNECTED AND COORDINATED CARE

Recommendation 2.1

Develop, strengthen and formalise MoC for rare metabolic diseases that incorporate multidisciplinary teams and care coordination.

Priority Action 2.1.1

Under the leadership of the Australasian Society for Inborn Errors of Metabolism (ASIEM) and with adequate resourcing, involve metabolic workforce expertise and patient experience to develop nationally consistent principles for diagnostic, clinical care and referral pathways or MoC for rare metabolic conditions.

Priority Action 2.1.2

Ensure national principles and MoC include measures to empower practitioners to provide culturally safe and appropriate care and referrals for Aboriginal and Torres Strait Islander people, those with culturally and linguistically diverse (CALD) backgrounds, and other priority populations outlined in the Action Plan.[1]

Priority Action 2.1.3

Adequately resource specialist metabolic services to employ nurse coordinators to arrange all elements of an individual's care with input from all relevant healthcare providers, including specialist physicians, generalists and allied health professionals, including genetic counsellors.

Recommendation 2.2

Develop and adopt nationally or jurisdictionally consistent and formalised diagnostic, clinical care and transition pathways for each rare metabolic disease group, based on available services (e.g. using 'HealthPathways' or similar).

Priority Action 2.2.1

Identify existing strengths and gaps in diagnostic, clinical care and transition pathways for each disease group, and prioritise work to leverage strengths, address gaps and progress consistent care pathways.

Priority Action 2.2.2

Develop and foster necessary professional relationships across multidisciplinary teams to support effective referral processes.

Recommendation 2.3

Strengthen care coordination using individualised care plans as standard practice for rare metabolic patients, similar to existing individualised chronic care plans.

Priority Action 2.3.1

Develop and integrate individualised care plans as part of core care coordination activities across all specialist metabolic services.

GOAL 3: CONSISTENT CARE INFORMED BY SPECIALIST METABOLIC EXPERTISE

Why is this important?

Often, people living with a rare metabolic disease access a variety of specialties outside metabolic medicine. But, for best practice care and outcomes, their care must be informed by a metabolic physician. This is consistent with international guidelines[5,8] and must be addressed to align with the following core theme in the vision of the Australian Government's National Medical Workforce Strategy 2021–2031, 'Use data and evidence – Draw on integrated data sets and common methodologies to support significant workforce decisions'.[7] However, the geographic spread of Australia's population, coupled with the paucity of individuals living with one of many rare metabolic conditions makes accessing the right metabolic expertise a challenge.

Variations in the composition and maturity of specialised metabolic services across Australia are resulting in inconsistent care and health outcomes for people living with a rare metabolic condition.[2] These variations affect clinical management, as well as access to timely diagnostic testing and treatments. Many are disadvantaged by living in jurisdictions without adequate specialist metabolic care and, in some cases, if that care is available, it is not readily accessible. In states or territories without a dedicated metabolic service (including, WA, TAS, Northern Territory (NT), Australian Capital Territory (ACT)),[2] or even where regional referral pathways to tertiary hospitals are not sufficient, there is an increased risk that patients are not connected with appropriate metabolic expertise. These challenges can and must be overcome.

The White Paper highlighted extensive variation in the core roles across existing specialist rare metabolic services in Australia[2]. These findings, supported by international best practice[5], identified several core roles and services essential to supporting people living with a rare metabolic condition. These roles and services include:

- Specialist metabolic physicians, including adult metabolic specialists
- Metabolic nurses and nurse coordinators
- Nurse practitioners
- Metabolic dietitians
- Genetic counsellors
- Research and clinical trial staff
- Administrative staff, including data management staff
- Transition clinicians
- Neuropsychology services
- Allied health services, including psychologists, occupational therapists, physiotherapists, speech pathologists and social work services
- Specialised diagnostic services
- Scientists/laboratorians

No specialist rare metabolic service in Australia has all these roles represented. Based on current (at the time of writing the White Paper) full-time equivalent allocations, no existing specialist metabolic services have sufficient capacity across these core roles to offer best-practice care and meet current and future levels of service demand.

Some jurisdictions (ACT, NT, TAS), do not have the population to support a state-based specialist metabolic service. For WA, which has a population comparable to that in South Australia, the challenge lies in the limited number of senior metabolic physicians available, and the number of metabolic clinicians with the right expertise to lead a service.

Working separately, it is unlikely that all Australian specialist metabolic services will be able to provide equal expertise across the over 1,800 known rare metabolic conditions.[3] A minimum cohort of service providers in each jurisdiction will be important to enable comprehensive clinical care across each disorder group. A networked hub and spoke model would further support recognition of leadership in particular services or jurisdictions for single conditions and facilitate sharing of rare metabolic disease knowledge and expertise.

Globally, countries are strengthening rare disease services and workforce by moving to a model that is centred on connecting and maximising expertise.[9] A structure that develops existing specialised services or pockets of expertise into recognised 'Centres of Excellence' linked in a formalised network offers considerable advantages over the current fragmented and inconsistent approach to rare disease care in Australia. An expansion of outreach and networked models of care is also one of 50 potential medical workforce solutions put forward in consultations for the development of the National Medical Workforce Strategy 2021–2031.[7] This policy highlighted the need for specialties to operate to their full scope outside metropolitan centres through innovative service provision approaches, network models and building relationships between hospitals.[7]

The United Kingdom (UK) and Europe drive high-quality care and maximise the reach of existing expertise by embedding key mechanisms for national knowledge sharing through the European Reference Network (ERN).[9] The metabolic ERN—MetabERN—is a world-leading example of this approach.[8] Similarly for Australia, connecting specialist services and infrastructure through an Australian Reference Network (ARN) would improve access and care for all Australians living with a rare metabolic disease. The ERN model is a tried and tested mechanism for overcoming the geographical spread of knowledge and expertise and eliminating distance barriers for those living with a rare metabolic disease in regional and remote areas.

RECOMMENDATIONS AND PRIORITY ACTIONS FOR CONSISTENT CARE INFORMED BY SPECIALIST METABOLIC EXPERTISE

Recommendation 3.1

Reduce urgent gaps by resourcing fit-for-purpose reciprocal arrangements in all states and territories for the diagnosis and management of rare metabolic diseases.

Priority Action 3.1.1

Provide funding for outreach clinics, so people living with a rare metabolic disease have access to routine care informed by specialist metabolic expertise in their own jurisdictions.

Priority Action 3.1.2

Leverage telemedicine in multidisciplinary team meetings to increase knowledge sharing and build a network of generalists, including general paediatricians, with a special interest in metabolic medicine. This approach would ensure local access to metabolic expertise and emergency care in rural and remote regions.

Priority Action 3.1.3

Develop interjurisdictional care arrangements that accurately reflect best-practice MoC, which include specific arrangements for acute care, subacute care and general management.

Priority Action 3.1.4

Ensure interjurisdictional care arrangements include measures that empower frontline health professionals and metabolic specialists to provide culturally safe and appropriate care and support to Aboriginal and Torres Strait Islander people, those with CALD backgrounds, and other priority populations outlined in the Action Plan.[1]

Recommendation 3.2

Patient care should always be informed by specialist metabolic expertise to reduce inconsistencies.

Priority Action 3.2.1

Where a patient's clinical care is led by a different specialty, develop referral pathways and clinical arrangements to ensure their care is consistently informed by specialist metabolic expertise.

Recommendation 3.3

Demonstrate progress towards implementing the full range of metabolic workforce expertise in each state and territory.

Priority Action 3.3.1

Ensure all patients living with a rare metabolic disease have access to the full range of expertise integrated within a specialist metabolic service. Ideally, the following core roles should be embedded within specialist metabolic services or readily accessible through timely formal referrals:

- Specialist metabolic physicians, including adult metabolic specialists
- Metabolic nurses and nurse coordinators
- Nurse practitioners
- Metabolic dietitians
- Genetic counsellors
- Research and clinical trial staff
- Administrative staff, including data management staff
- Transition clinicians
- Neuropsychology services
- Allied health services, including psychologists, occupational therapists, physiotherapists, speech pathologists and social work services
- Specialised diagnostic services
- Scientists/laboratorians

Recommendation 3.4

Progress the establishment of a network of specialised 'whole-of-life' metabolic service hubs incorporating research and innovation, in line with international direction for rare metabolic care (e.g. MetabERN).

Priority Action 3.4.1

In collaboration with ASIEM, further develop relationships and communication channels within the existing rare metabolic network.

Priority Action 3.4.2

Increase the capacity and formalisation of this network through Commonwealth, state and territory government funding.

Priority Action 3.4.3

Develop a national governance framework and criteria for a network of metabolic service hubs customised from international best practice models (e.g. MetabERN Structure and Governance).

GOAL 4: RECOGNITION OF THE METABOLIC SPECIALTY AND BEST PRACTICE CRITERIA

Why is this important?

Metabolic medicine is a subspecialty that brings together a broad range of clinical expertise centred around a comprehensive and precise knowledge of the biochemical processes of metabolism. People living with a rare metabolic disease, their families and carers require access to this specialist metabolic knowledge for accurate diagnosis, appropriate management and timely delivery of new therapies, which are vital for improving quality of life and extending life expectancy.[5]

The White Paper findings highlighted that without specialist metabolic oversight rare metabolic disease patients may face serious and avoidable consequences. These include delays in diagnosis and misdiagnosis, poor management of the underlying causes of disease and even death.[2]

Metabolic medicine is a relatively new subspecialty area, pressured by rapidly growing demand thanks to advances in diagnosis and management. Nonetheless, the metabolic specialty in Australia remains unrecognised and under-resourced, which limits opportunities for knowledge sharing, specialist professional education and pathways to innovative new treatments.[2]

Currently, the Royal Australasian College of Physicians (RACP) states, 'Metabolic medicine and cancer genetics represent subspecialties of clinical genetics and are not recognised by regulators in Australia or Aotearoa New Zealand as specialties in their own right'.[6]

Formal recognition of metabolic medicine as a specialty, by the RACP, is an essential first step to advocate for the development and resourcing of multidisciplinary and highly specialised rare metabolic services. This would enable a pipeline of suitably trained and experienced metabolic physicians, dietitians, genetic counsellors and nurses minimally required for best practice in a specialist metabolic service. This step further aligns with Priority 2 of the Australian Government's National Medical Workforce Strategy 2021–2031 to 'Rebalance Supply and Distribution' of the workforce by addressing undersupplied specialties across the country.[7]

Specialised metabolic dietitians are paramount in the care of most people living with a rare metabolic disease. But limited access to specialist metabolic dietitians, due to major skills and expertise shortages, is one example of several issues affecting the specialist metabolic workforce and patients in Australia.[2] Compounding these shortages is the lack of formal recognition of dietitians specialising in the treatment of rare metabolic diseases and no metabolic specialist metabolic registration for the nursing profession. Similarly, there is ad hoc limited funding for training and no clear training pathways for metabolic biochemical pathologists, who are critical to the provision of specialised biochemical diagnostics.

Findings in the White Paper also emphasised difficulties around classifying and defining rare metabolic diseases, and the lack of clear and consistent best-practice criteria for specialist metabolic services.[2] The roles of the specialist metabolic workforce are also unclear. Clearer classification of rare metabolic diseases, and criteria for specialist metabolic services, together with the inclusion of specific guidance on metabolic workforce roles and referral pathways, are needed.

This will reduce the current confusion, overlap and inconsistencies that create gaps, both within and across jurisdictions. Specialist metabolic services in some jurisdictions may have internal criteria already in use, but there is significant benefit to striving for consistency across Australia for what constitutes cost-effective and high value care.

Establishing a nationally consistent framework defining the role and composition of a rare metabolic service would provide all clinical specialties, diagnostic services (including specialised biochemical genetic pathologists), nursing, dietetics and allied healthcare practitioners (including genetic counsellors, neuropsychologists and social workers), with greater clarity around how to structure multidisciplinary care in a specialist metabolic service. This clarity will support development of business cases for building or growing a specialist metabolic service. It would also support coordination of care with generalist roles (general practitioners, paediatricians) to ensure timely referral of patients into specialist metabolic services for better outcomes. Where specialist metabolic services don't exist locally or within a patient's state or territory, clear disease management criteria would become a roadmap for coordinating generalists to connect patients with care in major centres or other jurisdictions.

RECOMMENDATIONS AND PRIORITY ACTIONS FOR CONSISTENT CARE INFORMED BY SPECIALIST METABOLIC EXPERTISE

Recommendation 4.1

Recognise the importance of specialist metabolic care for the estimated 12, 700 Australians living with a rare metabolic disease.

Priority Action 4.1.1

RACP to review the current classification of metabolic medicine as a subspecialty and consider formal recognition as a specialty.

Recommendation 4.2

Develop and/or recognise a best-practice framework for rare metabolic services for all Australians, that includes guidance on the roles of the specialised metabolic workforce and connections with other specialties.

Priority Action 4.2.1

Prioritise the development of nationally consistent criteria for rare metabolic services, led by ASIEM and endorsed by the HGSA. This should align with existing international best practice e.g. the ERN model.

Priority Action 4.2.2

Develop/identify and formally adopt disease-specific management criteria that recognise and are informed by clinical expertise.

Priority Action 4.2.3

Ensure disease specific management criteria recognise and respond to the needs of Aboriginal and Torres Strait Islander peoples, those with CALD backgrounds, and other priority populations outlined in the Action Plan.[1]

Recommendation 4.3

Develop and embed formal training pathways for the rare metabolic workforce that include adult and paediatric metabolic clinicians, dietitians, nurses, allied health and mental health professionals, and biochemical genetic pathologists.

Priority Action 4.3.1

Provide a specific pathway to metabolic fellowship for medical graduates and biochemical genetic pathologists under a new RACP specialty of Metabolic Medicine.

Priority Action 4.3.2

Fund more training positions for biochemical genetic pathologists to meet the requirements/ensure compliance with National Pathology Accreditation Advisory Council (NPAAC) guidelines.

Priority Action 4.3.3

Fund more positions for metabolic dietitians and encourage graduate dietitians to undertake provisional training in a specialist metabolic service in a tertiary hospital setting, with direction from an experienced metabolic dietitian.

Priority Action 4.3.4

Guided by ASIEM, develop learning modules and a competency framework for metabolic dietetics similar to those already in place for paediatric nutrition. These can be customised for the Australian context from existing learning modules already available in the UK and the United States of America.

Priority Action 4.3.5

Fund more positions for metabolic nurses in specialist metabolic services, including nurse coordinators, research nurses and nurse practitioners. Encourage nurse practitioners to consider a career in rare metabolic care.

Priority Action 4.3.6

Encourage tertiary institutions to include a strengthened focus on the treatment of rare metabolic conditions in Approved Programs of Study for the endorsement of metabolic nurse practitioners, with guidance from the Nursing and Midwifery Board of Australia.

Priority Action 4.3.7

Develop and embed awareness and education that empowers all metabolic specialists to provide culturally safe and appropriate care and support to Aboriginal and Torres Strait Islander people, those with CALD backgrounds, and other priority populations outlined in the Action Plan.[1]

GOAL 5: CARE RESPONSIVE TO INNOVATION

Why is this important?

Access to research and innovation and the ability to offer appropriate clinical management for newly approved medicines is vital for delivering best practice in rare disease care.[1] Priority 3.4 in the Action Plan, 'Translate research and innovation into clinical care; clinical care informs research and innovation'[1] must be adopted as part of any rare disease workforce strategy. In keeping with the guiding principle of the National Metabolic Workforce Strategy 2021–2031 — 'be brave and aspirational' — a national rare metabolic workforce strategy must embed research and innovation into care. Without this, Australia cannot be responsive to new technologies or keep up with international directions.

Challenges associated with embedding research and innovation into specialist metabolic services—including insufficient staffing and infrastructure—were consistently reported in consultations for the White Paper[2], and further highlighted in The New Frontier - Delivering better health for all Australians.[10]

There are exciting and rapid developments on the horizon for new therapies and technologies in metabolic medicine, particularly with a move towards more personalised approaches like cell and gene therapies. However, Australians living with a rare metabolic disease 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.

The uptake of any new therapy or technology for metabolic conditions approved for clinical trials or granted subsidisation under Australian reimbursement pathways such as, the Life Saving Drugs Program (LSDP)[11], the Pharmaceutical Benefits Scheme (PBS)[12] and the Medicare Benefits Schedule (MBS)[13] relies on a suitably skilled metabolic workforce. However, resources within existing specialist metabolic services are already stretched beyond capacity, so clinical trials and new therapies for rare metabolic diseases cannot be prioritised without compromising current caseloads. This means Australians living with a rare metabolic disease are routinely missing out on new therapies.

Moving forward, Australia should look to international leaders, such as Great Ormond Street Hospital, which is a centre of excellence in metabolic medicine with a dedicated metabolic research facility and high capacity for clinical trials.

RECOMMENDATIONS AND PRIORITY ACTIONS FOR CARE RESPONSIVE TO INNOVATION

Recommendation 5.1

Establish dedicated staff and infrastructure within specialist metabolic services for ongoing participation in research, clinical trials and clinical management of newly approved health technologies.

Priority Action 5.1.1

Identify, develop, maintain and embed workforce skills and capabilities to better respond to emerging technologies.

Recommendation 5.2

Build capacity for Australia-wide coordination of specialist metabolic services to deliver innovative therapies so all Australians living with a rare metabolic disease have timely access to clinical trials and newly approved health technologies.

Priority Action 5.2.1

Provide nationally coordinated infrastructure to develop and formalise metabolic centres of excellence that embed research as part of best practice care.

Priority Action 5.2.2

Provide nationally coordinated infrastructure to develop and formalise rare metabolic disease data collection and registries to build knowledge, identify gaps and facilitate opportunities for clinical trials and the approval of new drugs.

Priority Action 5.2.3

Commonwealth to adopt the Australian Commission on Quality and Safety in Health Care's National One Stop Shop and Clinical Trials Front Door platform for health-related human research.[14]

Priority Action 5.2.4

In response to the global health data standard World Health Organization mandate[15] and Action 3.1 in the Action Plan, implement ORPHACodes[16] in reporting rare metabolic diseases. Contribute to the mapping and interoperability of existing health data sets to unlock knowledge, and ensure rare diseases are better represented in future implementations of classifications, such as International Classification of Diseases 11th revision - Australian Modification (ICD-11-AM).

NEXT STEPS

Thirty years ago, people living with a rare metabolic disease often had no diagnosis or explanation for their symptoms, and treatments were limited or did not exist. Many children either died or lived with severe disability into adulthood. Owing to advancements in genetic testing and medical treatments, together with the accumulation of clinical knowledge over time, the over 12,700 Australians estimated to be living with a rare metabolic disease now have the prospect of living fuller and longer lives.

This Strategy provides an evidence-based, expert-backed framework of goals, recommendations and priority actions that address current high levels of unmet need through a nationally consistent and sustainable workforce resourced to respond swiftly to innovation and deliver best practice care for better health and wellbeing outcomes.

A person-centred approach to implementing this Strategy is the responsibility of all stakeholders. It requires cooperation and commitment from governments, hospital administrators, healthcare providers, specialist physician groups and policymakers across states and territories, and at a national level. Implementation would be best achieved in alignment with the framework established for the broader National Medical Workforce Strategy 2021–31.[7]

The Strategy's goals for a recognised, connected, consistent, sustainable and innovative rare metabolic disease workforce, should be further leveraged to respond to broader rare disease workforce challenges in Australia.

The rare disease workforce must be equipped and nuanced to cover the over 7,000 rare diseases impacting an estimated 2 million Australians.[1] 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 ARN customised from the ERN model is recommended to address the geographical spread and inherent scarcity of rare disease patients and specialist expertise that already exists in Australia. Bringing together the best knowledge, expertise and resources from across the country will ensure all Australians living with a rare disease have access to the best available care.

REFERENCES

1. Commonwealth of Australia. Department of Health. National Strategic Action Plan for Rare Diseases. Canberra; 2020. 63 p. Available from: https://www.health.gov.au/sites/default/files/documents/2020/03/national-strategic-action-plan-for-rare-diseases.pdf
2. Equity Economics and Rare Voices Australia (2022), Rare Metabolic Disease Workforce White Paper: Towards a Strengthened Rare Disease Workforce for Australia, February 2022. Available from: https://rarevoices.org.au/launched-rare-metabolic-disease-workforce-white-paper/
3. Inborn Errors of Metabolism Knowledgebase [Internet]. 2018 [updated 2022 28 June]. Available from: http://www.iembase.org/index.asp
4. Anderson M, Elliott, EJ, Zurynski YA. Australian families living with rare disease: Experiences of diagnosis, health services use and needs for psychosocial support. Orphanet J. Rare Dis. [Internet]. 2013;8:22. Available from: https://doi.org/10.1186/1750-1172-8-22
5. Burton H, Sanderson S, Shortland G, Lee P. Needs assessment and review of services for people with inherited metabolic disease in the United Kingdom. J. of Inherit. Metab. Dis. [Internet]. 2006;29:667–76. Available from: https://doi.org/10.1007/s10545-006-0374-0
6. RACP Specialists Together. 2022 Handbook [Internet]. n.d. Program Overview, Clinical Genetics. Available from: https://www.racp.edu.au/trainees/advanced-training/advanced-training-programs/clinical-genetics
7. Commonwealth of Australia. Department of Health. National Medical Workforce Strategy 2021–2031. Canberra; 98 p. 2021. Available from: https://www.health.gov.au/initiatives-and-programs/national-medical-workforce-strategy-2021-2031#:~:text=The%20National%20Medical%20Workforce%20Strategy%202021%E2%80%932031%20identifies%20achievable%2C%20practical,emerging%20health%20needs%20of%20Australians
8. European Reference Network. MetabERN, European Reference Network for Hereditary Metabolic Disorders. MetabERN Vision [Internet]. n.d. Available from: https://metab.ern-net.eu/about-us-3/#vision
9. European Commission, Public Health. European Reference Networks [Internet]. n.d. Available from: https://ec.europa.eu/health/european-reference-networks/overview_en
10. Parliament of the Commonwealth of Australia. The New Frontier—Delivering better health for all Australians. Canberra; 2021. 406 p. Available from: https://www.aph.gov.au/Parliamentary_Business/Committees/House/Health_Aged_Care_and_Sport/Newdrugs/Report
11. Commonwealth of Australia. Department of Health and Aged Care. Life Saving Drugs Program. [updated 2022 July]. Available from: https://www.health.gov.au/initiatives-and-programs/life-saving-drugs-program
12. Commonwealth of Australia. Department of Health and Aged Care. Pharmaceutical Benefits Scheme [updated 2022 September]. Available from: https://www.pbs.gov.au/pbs/home
13. Commonwealth of Australia. Department of Health and Aged Care. Medicare Benefits Schedule [updated 2022 January]. Available from: http://www.mbsonline.gov.au/internet/mbsonline/publishing.nsf/Content/Home
14. Australian Commission on Safety and Quality in Health Care. The National One Stop Shop – a national platform for health-related human research. [updated 2022 June]. Available from: https://www.safetyandquality.gov.au/our-work/health-and-human-research/national-one-stop-shop-national-platform-health-related-human-research

15. World Health Organization. International Statistical Classification of Diseases and Related Health Problems. [updated 2022 September]. Available from: https://www.who.int/standards/classifications/classification-of-diseases
16. RD Code. What are ORPHAcodes? Available from: http://www.rd-code.eu/introduction/
17. Australian Medical Association. AMA National Code of Practice – Hours of Work, Shiftwork and Rostering for Hospital Doctors. Australian Capital Territory; 2016. 35 p. Available from: https://ama.com.au/sites/default/files/documents/FINAL_NCP_Hours_of_work_2016.pdf

APPENDIX

List of Abbreviations

ARN	Australian Reference Network
ASIEM	Australasian Society for Inborn Errors of Metabolism
CALD	Culturally and Linguistically Diverse
Equity Economics	Equity Economics and Development Partners
ERN	European Reference Network
HGSA	Human Genetic Society of Australasia
ICD-11	International Classification of Diseases 11th revision
LSDP	Life Saving Drugs Program
MBS	Medicare Benefits Schedule
MetabERN	Metabolic European Reference Network
MoC	Model of Care
NDIS	National Disability Insurance Scheme
NPAAC	The National Pathology Accreditation Advisory Council
NSW	New South Wales
ORPHAcodes	Orphanet nomenclature of rare diseases
PBS	Pharmaceutical Benefits Scheme
QLD	Queensland
RACP	Royal Australasian College of Physicians
RVA	Rare Voices Australia
SA	South Australia
TAS	Tasmania
The Action Plan	The National Strategic Action Plan for Rare Diseases
The Strategy	National Strategy for Australia's Rare Metabolic Diseases Workforce
The White Paper	The Rare Metabolic Workforce White Paper: Towards a Strengthened Rare Disease Workforce for Australia
UK	United Kingdom
VIC	Victoria
WA	Western Australia

National Strategy for Australia's Rare Metabolic Disease Workforce

Rare Voices Australia and Equity Economics and Development Partners

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