New South Wales

Special Commission of Inquiry into Healthcare Funding

Submission Number: 75

Name: Rare Voices Australia (RVA)

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31 October 2023

Mr Richard Beasley SC
Commissioner
The Special Commission of Inquiry into Healthcare Funding

Dear Mr Beasly,

Rare Voices Australia (RVA) commends the New South Wales government's intentions to review the funding of NSW health services and welcomes this opportunity to provide input into the Special Commission Inquiry into Healthcare Funding (Inquiry). The Terms of Reference are highly relevant in the context of rare disease healthcare.

Rare diseases, like many other chronic diseases, are often serious and progressive. People living with rare disease face common challenges, including the struggle for a timely and accurate diagnosis, limited access to the right rare disease expertise, limited treatment options and high degrees of symptom complexity necessitating access to a wide variety of specialties through coordinated multidisciplinary teams. Any efforts to improve healthcare funding in NSW should consider these challenges. These complexities require a carefully planned and nuanced approach to healthcare funding that is efficient, sustainable and responsive to change.

This funding review is timely regarding the Australian Government's recent commitment to equitable expansion of the Newborn Bloodspot Screening (NBS) program through national consistency and delivery of wrap around specialist care.[1] There is opportunity to leverage the Commonwealth investments to this program. The NSW Government has made a commitment to timely implementation of conditions recommended for NBS. Workforce readiness is crucial to this. Through this Inquiry, NSW can lead the way in Australia building and strengthening health workforce capacity through systematic and sustainable funding reforms.

About Rare Voices Australia

RVA is the national peak body for the estimated two million Australians living with a rare disease. RVA has a strong track record in systemic advocacy for broad rare disease policy reform across government departments, including health, disability and research. RVA's work is non-disease specific.

RVA led the collaborative development of the National Strategic Action Plan for Rare Diseases[2] (the Action Plan), the first nationally coordinated effort to address rare diseases in Australia. The Action Plan includes 3 interrelated pillars—Pillar 1: Awareness and education, Pillar 2: Care and Support, and Pillar 3: Research and Data.

Rare Disease and the Health Workforce in Australia – through the lens of rare metabolic disease care

The rare disease workforce in Australia is often under resourced and funding models are not-fit-for-purpose to optimally manage the complexity of rare conditions. There are limited treatments for rare disease. For many people living with a rare disease, clinical trials are the only way to access treatment. However, the health workforce in Australian is not resourced to embed research into clinical care. Until this happens patients cannot access best practice in rare disease care.

A key priority in the Action Plan, Priority 1.3, is the development of a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics. As a first step to achieving this, RVA engaged policy researchers and an expert project steering committee to conduct research and build evidence around the strengths and gaps in the healthcare system managing the care of people living with rare metabolic diseases. Rare metabolic diseases are a large group of over 1,600 complex multisystem rare diseases affecting approximately 12,700 Australians. These conditions are often diagnosed in asymptomatic newborns via newborn bloodspot screening.

This work led to publication of the Rare Metabolic Disease Workforce White Paper – Towards a strengthened Rare Disease Workforce in Australia (the White Paper),[3] which includes 5 key findings, highlighting critical workforce shortages and insufficient resources leading to suboptimal care for these patients even in Australia's most renowned specialist metabolic services. With only 4 specialist rare metabolic disease services in Australia resources are spread thin to accommodate for patients in other jurisdictions and funding for outreach services is limited and, in some cases, not available. There is also a lot of reliance on the goodwill of already burned-out clinicians.

Stakeholders consulted in development of the White Paper highlighted insufficient resources were likely attributed to a lack of appropriate fundings models for the rare metabolic disease workforce. According to many clinicians and members of the White Paper expert steering committee, activity-based funding models were not able to respond to the complex multidisciplinary care required for rare metabolic patients. One potential solution to this is block funding in line with the model used to fund metabolic services in the United Kingdom.

From our discussions with various stakeholders as the peak body, RVA suspects activity-based funding may not be being used to its full potential within these Australian services. Many clinicians and management staff are too time poor to learn ways to optimise the use of activity-based funding. Finding ways to modify or extend activity-based funding models and finding time to educate clinicians and staff on the optimal use of these existing models is highlighted in the Priority Actions under Goal 1: 'Sustainable Systems and Workforce', of the National Strategy for Australian Rare Metabolic Workforce (attached).

The White Paper includes details of the current services (at the time of writing) available for people living with a rare metabolic disease in NSW (see separate attachment). A Summary document outlining the current status (at the time of writing) of rare metabolic care in NSW is also attached separately. The Summary details a list of urgent key actions and next steps, including detailed recommendations for investment to improve rare metabolic care in NSW.

National Strategy for Australia's Rare Metabolic Disease Workforce

After the White Paper was launched in February 2022 RVA led a consultative process to develop a National Strategy for Australia's Rare Metabolic Disease Workforce (the Strategy). The goals, recommendations and priority actions in the Strategy were developed for broader applicability to the whole rare disease workforce. The goals for a recognised, connected, consistent, sustainable and innovative rare metabolic disease workforce directly align with the Terms of Reference in this consultation.

RVA looks forward to hearing results from the Special Commission and any systemic change that leads on from this work. RVA is open to further discussions and involvement in this important initiative by the NSW government towards a person-centred healthcare workforce resourced to meet current and future need and respond swiftly to innovation.

Kind regards,

Nicole Millis
Chief Executive Officer
Rare Voices Australia

References

1. Rare Voices Australia. RVA Education Webinar – Expanding Newborn Bloodspot Screening: progress Made and Next Steps. Accessed 30 October 2023. https://rarevoices.org.au/rva-education-webinar-expanding-newborn-bloodspot-screening-progress-made-and-next-steps/
2. Australian Government. Department of Health. National Strategic Action Plan for Rare Diseases. Canberra; 2020. 63 p.
3. Equity Economics and Rare Voices Australia. Rare Metabolic Disease Workforce White Paper: Towards a Strengthened Rare Disease Workforce for Australia. February 2022. 32 p. rarevoices.org.au/launched-rare-metabolic-disease-workforce-white-paper/https://www.health.gov.au/sites/default/files/documents/2020/03/national-strategic-action-plan-for-rare-diseases.pdf

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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
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- 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

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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.

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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

February 2023

Australian Government

Department of Health

National Strategic Action Plan for Rare Diseases

February 2020

We acknowledge Traditional Owners of Country throughout Australia and recognise the continuing connection to lands, waters and communities. We pay our respect to Aboriginal and Torres Strait Islander people, and to Elders both past and present.

Development of the National Strategic Action Plan for Rare Diseases was led by Rare Voices Australia with funding from the Australian Government Department of Health.

Title: The National Strategic Action Plan for Rare Diseases

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Foreword

Nicole Millis
Chief Executive Officer – Rare Voices Australia

Lack of awareness, the struggle for a timely and accurate diagnosis, limited care and support options, a lack of research, poor data collection and use…

The great complexity and unmet need in rare diseases can be overwhelming for the entire sector: for policy-makers, clinicians, practitioners, researchers, academics, industry and especially for the people who live with a rare disease. While individual diseases may be rare, globally, approximately eight per cent of the population live with a rare disease[1]. This equates to around two million Australians[2].

There is so much more to rare diseases than small numbers. Limited data is a common feature of rare diseases, often resulting in high uncertainty, which impacts every part of people's lives. People are faced with impossible choices based on incomplete knowledge and unclear pathways. When, eighteen years ago, my son was diagnosed with a rare disease, all I had were questions and very few answers. Why did this happen? How do I fix it? What do I do? Where do I go? Will he have a future and what will it look like?

To respond effectively to rare diseases, Australia needs to reduce uncertainty through policy. The National Strategic Action Plan for Rare Diseases (the Action Plan), with its three Pillars – Awareness and Education, Care and Support, and Research and Data – can be summarised into a Plan on a Page (page 6). The Action Plan has been written to provide a comprehensive policy framework. The scope of the Action Plan aligns with international rare diseases plans, and its flexibility allows the Action Plan to respond to changing policy contexts and opportunities. As rare diseases are often progressive, we know taking action is time critical. Importantly, the Action Plan clearly describes the implementation mechanisms that will drive progress to improve outcomes now and in the future.

Rare Voices Australia (RVA) would like to thank key stakeholders who contributed to the development of this Action Plan:

- people living with or caring for someone with a rare disease, who shared their lived experience of rare diseases and ideas for improvement;
- RVA Partner Organisations, who provided representative input into the development of this Action Plan and do such a wonderful job of supporting and advocating for their members;
- members of the Steering Committee, who participated in the initial Roundtable Consultation and provided valuable insights to help shape the Action Plan; in particular, its vision, critical enablers, principles and priorities; and
- RVA's Scientific & Medical Advisory Committee (SMAC) and Roundtable of Companies, as well as state and territory departments of health, each of whom contributed their expertise towards the development of this Action Plan.

We were overwhelmed by the willingness and generosity of those involved at so many different levels. We thank everyone for working together towards the best possible health and wellbeing outcomes for Australians living with a rare disease. Such collaboration is essential and needs to be encouraged. For so long, progress in the rare disease sector has been largely fragmented and undertaken in isolation. The Action Plan shows us what we can do when we encourage collaboration.

Through national leadership and coordination, the Action Plan can drive and future-proof much needed reform. With effective policy, we can reduce the disempowering uncertainty of rare diseases. I know first-hand that effective policy can transform the lives of people living with a rare disease. I also know this Action Plan can realise its vision: the best possible health and wellbeing outcomes for Australians living with a rare disease.

Nicole Millis

rare voices
AUSTRALIA

Executive Summary

Developed by the rare disease sector, for the rare disease sector

The National Strategic Action Plan for Rare Diseases is the first nationally coordinated effort to address rare diseases in Australia. Due to the great complexity, significant unmet need and critical urgency associated with rare diseases, systemic reform is required. While there are many different rare diseases, they share countless commonalities. Informed by extensive stakeholder consultation, the Action Plan addresses this common ground. It represents the views of the rare disease sector and outlines a comprehensive, collaborative and evidence-based approach to achieving the best possible health and wellbeing outcomes for Australians living with a rare disease. It is built on three principles: *person-centred*, *equity of access* and *sustainable systems and workforce*.

The Action Plan aligns with, and expands on, the *Call for a National Rare Disease Framework: 6 Strategic Priorities,* which was published by RVA in June 2017. Additionally, it aligns with the *National Strategic Framework for Chronic Conditions*, the *National Aboriginal and Torres Strait Islander Health Plan 2013–2023* and the *WHO Global Action Plan for the Prevention and Control of Noncommunicable Diseases*. Crucially, it also aligns with the *Asia-Pacific Economic Cooperation (APEC) Action Plan on Rare Diseases*. Covering a wide scope, the Action Plan is comparable to other international rare disease plans and strategies, including those in Europe, the United Kingdom (UK), Canada and the United States (US).

The Action Plan is comprised of three core Pillars, with each Pillar outlining priorities, actions and implementation areas. The Pillars are:

1. Awareness and Education
2. Care and Support
3. Research and Data

The Pillars are easily recognisable to people living with a rare disease. For the purposes of this Action Plan, each Pillar is presented separately. However, in reality, all Pillars are interrelated. As such, the strongest policy responses address priorities across multiple Pillars. Effective policy reform in one area will create change and momentum in other areas. The wide scope of the Action Plan allows it to respond to changing policy contexts and leverage from current and future opportunities.

The importance of national leadership and coordination in rare diseases cannot be underestimated. Through the stakeholder consultations, the sector highlighted the following critical enablers of effective rare diseases policy:

- multi-stakeholder involvement and engagement;
- collaborative governance and leadership;
- state, national and international partnerships; and
- comprehensive, high quality collection, and effective use of rare diseases data.

Time is also critical in rare diseases. Consequently, the Action Plan highlights efficient and sustainable mechanisms that will drive implementation. The Action Plan will help to achieve the sector's vision of the best possible health and wellbeing outcomes for Australians living with a rare disease.

Plan on a Page

VISION

The best possible health and wellbeing outcomes for Australians living with a rare disease.

CRITICAL ENABLERS

Multi-stakeholder involvement and engagement

Collaborative governance and leadership

State, national and international partnerships

High quality, comprehensive collection, and effective use, of rare disease data

PILLAR 1: AWARENESS AND EDUCATION

Priority 1.1: Increase every Australian's awareness of rare diseases including, where applicable, relevant prevention measures.

Priority 1.2: Ensure Australians living with a rare disease have access to information and education that enables them to be active participants in their rare disease journey.

Priority 1.3: Develop a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics.

PILLAR 2: CARE AND SUPPORT

Priority 2.1: Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family-centred.

Priority 2.2: Ensure diagnosis of a rare disease is timely and accurate.

Priority 2.3: Facilitate increased reproductive confidence.

Priority 2.4: Enable all Australians to have equitable access to the best available health technology.

Priority 2.5: Integrate mental health, and social and emotional wellbeing, into rare disease care and support.

PILLAR 3: RESEARCH AND DATA

Priority 3.1: Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning.

Priority 3.2: Develop a national research strategy for rare diseases to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps.

Priority 3.3: Ensure research into rare diseases is collaborative and person-centred.

Priority 3.4: Translate research and innovation into clinical care; clinical care informs research and innovation.

THESE PRINCIPLES ARE THE FOUNDATION FOR THIS ACTION PLAN

Person-centred

Equity of access

Sustainable systems and workforce

Contents

About the Action Plan

RVA was commissioned by the Australian Government to develop the Action Plan on behalf of the rare disease sector.

The Action Plan provides guidance and direction around key goals and priorities for Australians living with a rare disease. It sets out actions and activities, as determined by the sector, which could be introduced to improve the health and wellbeing of Australians living with a rare disease.

The actions identified in the Action Plan are for consideration by a wide range of stakeholders, including governments at all levels, non-government organisations, the public and private health sectors, industry, researchers and academics, rare disease organisations and the wider community. The implementation of any of the actions outlined is a decision for each stakeholder, based upon their area of responsibility, governance remit, existing activities and future planning and directions in relation to rare diseases.

Introduction

What is a rare disease?

The most widely accepted definition is that a rare disease is one that affects less than five in 10,000 people [3,4]. While estimates of the number of rare diseases may vary between countries and studies, due to differing definitions and challenges with data collection, it is prominently cited that there are more than 7,000 different rare diseases[5]. The increasing precision of genomic technologies means that new diseases are being discovered regularly[6]. While individual diseases may be rare, the total number of Australians living with a rare disease is not. Approximately eight per cent of Australians live with a rare disease[7]. Extrapolated to an Australian population of over 25 million people[8], this equates to around two million Australians.

Approximately 80 per cent of rare diseases are of genetic origin[9]. Types of non-genetic rare diseases include cancers, infections and autoimmune disorders[10]. In some cases, health professionals may assess that a person's phenotype[a] strongly suggests the presence of a genetic condition, however, a diagnosis that explains all symptoms is unable to be made[11,12]. This may be because the condition has not yet been discovered (and thus a diagnosis is not yet possible) or because health professionals have not yet made the correct diagnosis. For brevity, the term 'undiagnosed rare diseases' is used throughout this Action Plan to describe these conditions.

Rare diseases, like many other chronic diseases, are often serious and progressive. They typically display a high level of symptom complexity and thus are a significant cause of ongoing health and psycho-social challenges. There is no cure for many rare diseases, and so improving quality of life and extending life expectancy of people living with a rare disease relies on appropriate treatment and care[13].

While there is large variation among rare diseases, people living with a rare disease face common challenges. They include the struggle for a timely and accurate diagnosis, limited care and support options and a lack of research into rare diseases, despite the recognised gaps in knowledge[14]. People living with a rare disease and their families also experience financial impacts, either due to out-of-pocket costs associated with care and support, or due to loss of income associated with taking on a carer role[15].

Rare diseases are often difficult for health professionals to diagnose, not only due to their rarity but also because of their high level of symptom complexity. Health professionals are not typically taught sufficiently about rare diseases as part of their standard training[16]. Significant diagnostic delay and misdiagnosis is common, and is often referred to as the 'diagnostic odyssey'. Yet, a timely diagnosis is critical for better patient outcomes, the provision of the best possible care and treatment options, access to services and support, increased reproductive confidence and the ability to participate in clinical trials.

Rare diseases pose many challenges for all: from the person living with a rare disease, to their family and carers, rare disease organisations, the wider community, health professionals, researchers, the pharmaceutical industry and governments. It can be difficult for health professionals to gain deep, specialised knowledge and experience when seeing low patient numbers in comparison to more common diseases. Researchers face an uphill battle in securing funding and in coordinating statistically robust studies. Pharmaceutical industry interest in rare disease research and development can be low due to the relatively low demand[17].

[a] Phenotype is a term that refers to 'the observable physical properties of an organism… [including its] appearance, development and behavior.'

Rare diseases: the international approach

'No country can claim to have achieved universal healthcare if it has not adequately and equitably met the needs of those with rare diseases.'

– Helen Clark, United Nations Development Programme (2009–2017)[18]

Internationally, momentum is building to address the needs of those living with a rare disease. Canada, the US, France and the UK are among the countries already tackling rare diseases through policy and legislation. Of importance to Australia, the APEC Action Plan on Rare Diseases envisions that, by 2025, 'APEC member economies will aim to improve the economic and social inclusion of all those affected by rare diseases by addressing barriers to healthcare and social welfare services'[19].

Common to international rare diseases strategies and plans are objectives relating to: prevention; early detection and screening; accurate and timely diagnosis; access to care and clear care pathways; supporting people living with a rare disease beyond the health domain; increasing development of, and access to, therapies (including orphan drugs); and the crucial role of research and innovation. Significant international initiatives include the International Rare Diseases Consortium (IRDiRC), which promotes international collaboration and advances research into rare diseases worldwide[20]; EURORDIS – European Organisation for Rare Diseases[21]; Rare Disease Day – an international day to raise awareness about rare diseases and their impact on patients' lives[22]; and Orphanet – the portal for rare diseases and orphan drugs[23].

Rare diseases: the Australian context

The 'Awakening Australia to Rare Diseases' international symposium of over 200 delegates was held in Fremantle, Western Australia in 2011[24]. In response, a national peak body was established for rare diseases in Australia: RVA. Since its establishment in 2012, RVA has advocated for a national plan for rare diseases and more effective policy for Australians living with a rare disease.

In 2014, RVA undertook a roadshow to progress a national plan for rare diseases. Roundtable discussions with key stakeholders in Queensland, Victoria, South Australia, New South Wales and Western Australia were held. Key findings on the principles and objectives to progress a national plan were presented at the National Rare Disease Summit in 2015. The collaborative outcome of the Summit was a *Communiqué to progress a National Rare Disease Plan* that listed key principles and objectives. The *Communiqué* was subsequently endorsed by more than 170 organisations and individuals in the rare disease community and was key to RVA's advocacy for a coordinated national response to rare diseases[25]. In June 2017, the themes of the *Communiqué* were further developed into the key advocacy and policy document, *Call for a National Rare Disease Framework: 6 Strategic Priorities*[26]. This made the case for a nationally coordinated approach to effective rare diseases policy and was presented to the Minister for Health, the Hon Greg Hunt MP. This was critical in creating momentum in rare diseases policy reform, particularly around reforms to the Life Saving Drugs Program (LSDP)[27]; Medical Research Future Fund (MRFF) grant opportunities targeting rare diseases (*Rare Cancers and Rare Diseases and Unmet Needs* competitive grant program)[28]; and fee exemptions in relation to the Therapeutic Goods Administration (TGA) reforms to Orphan Drug Designation[29].

The Australian Government has also made significant investments into genomics, a health technology with great potential for rare diseases. This was done through Australia's first *National Health Genomics Policy Framework 2018–2021*[30] and associated Implementation Plan[31]. At a state and territory level, notable examples include the Western Australian Rare Diseases Strategic Framework 2015–2018[32] and the Western Australian Undiagnosed Diseases Program[33].

Despite these measures, in Australia, the response to the spectrum of rare diseases (genetic, undiagnosed, cancers, infections and autoimmune disorders) is varied and inconsistent. Areas of significant unmet need persist, and the burden of rare diseases remains unacceptably high. There is room for improvement in terms of measuring and tracking rare diseases. Hospitals can only code 517 of the almost 7,000 rare diseases coded by Orphanet[34]. Quality national data on congenital anomalies, a large class of mainly rare diseases, is unavailable, despite recognition that they are a significant public health problem in Australia[35]. On average, Australians are waiting anywhere between two to four years longer for access to Government-funded treatment for rare diseases in contrast to comparable countries[36]. There are also cases where a medicine that will help a rare disease is only subsidised under the Pharmaceutical Benefits Scheme (PBS) for a common disease, meaning rare diseases patients have to pay more for the exact same medicine. Commonwealth, state and territory approaches to rare diseases remain fragmented.

In November 2018, the Federal Minister for Health, the Hon Greg Hunt MP, announced, with bipartisan support, funding for RVA to collaboratively develop and deliver the Action Plan, the first nationally coordinated effort to address rare diseases in Australia[37]. Collaborative work towards the development of the Action Plan began immediately at the 2018 National Rare Disease Summit, which brought all key stakeholders in the rare disease sector together, including rare disease organisation leaders, clinicians, researchers, government and industry.

Priority Populations

Action and policy for rare diseases must recognise all Australians and focus on access and equity for priority populations. As such, relevant implementation recommendations in the Action Plan identify the need to undertake targeted activities to improve health and wellbeing outcomes for priority populations.

Australians living with a rare disease

The overarching priority population is, of course, the approximately two million Australians living with a rare disease, and their families and carers. The term 'people living with a rare disease' is used deliberately to acknowledge the whole person. The prominent role of families and carers is widely recognised in the rare disease community and where possible, is acknowledged in terminology throughout.

Several other priority populations emerged through consultations with the rare disease community and subject matter experts: Australians living with an undiagnosed rare disease; Australians with an increased chance of developing a rare disease or of having a child with a rare disease; Aboriginal and Torres Strait Islander people; people living in regional, rural and remote areas; people from culturally and linguistically diverse (CALD) backgrounds; and people experiencing socio-economic disadvantage.

Australians living with an undiagnosed rare disease

Both diagnostic delay and misdiagnosis are common features of rare diseases, and can negatively impact the level of care and support received by individuals[38]. Without a diagnosis, Australians living with an undiagnosed rare disease cannot be provided with an accurate prognosis and have no access to evidence-based treatment. Moreover, a lack of diagnosis is reported as a roadblock to obtaining adequate funding from the National Disability and Insurance Scheme (NDIS)[39].

Australians with an increased chance of developing a rare disease or of having a child with a rare disease

This population can be considered in three categories:

- **hereditary (genetic):** this category comprises Australians who have known or unknown genetic variant/s that may cause them to develop a rare disease in the future, and/or their children to develop a rare disease. It includes those with a family history of a rare disease. This is especially significant as approximately 80 per cent of rare diseases are of genetic origin[40].
- **non-hereditary, related to pregnancy:** in certain instances, pregnant women may have an increased chance of being vulnerable to diseases such as rubella (German measles) and chickenpox (varicella). While these diseases are not considered rare, passing them onto their unborn child may result in rare congenital disorders in the child (such as congenital rubella syndrome, or congenital varicella syndrome)[41]. Evidence also indicates women who have, or may be at risk of developing, certain chronic conditions, such as diabetes, epilepsy or thyroid disorders (for example, through family history of these disorders), may have an increased chance of having babies with rare congenital anomalies[42,43].

- **non-hereditary, unrelated to pregnancy:** some Australians are at an increased risk of developing or contracting rare non-hereditary diseases. Examples of these populations include: people who may be exposed to certain factors in their environment (such as rural populations or farm animal handlers to Q fever, or those living in areas where *Mycobacterium ulcerans* is present); and those with low immunity and thus increased vulnerability (such as newborns prior to receiving scheduled vaccinations, such as for whooping cough, or the immunocompromised).

Aboriginal and Torres Strait Islander people

While Aboriginal and Torres Strait Islander people are not necessarily at greater risk of rare diseases, several factors increase the potential impact of rare diseases on Aboriginal and Torres Strait Islander people. The lack of research into rare diseases means our knowledge on which rare diseases are most prevalent within Aboriginal and Torres Strait Islander people is incomplete. Given the genetic basis of most rare diseases, research exploring this is vital.

The following factors further contribute to the unique challenges faced by Aboriginal and Torres Strait Islander people:

- while the areas of genetics and genomics are expanding rapidly, inequity exists in the inclusion of Aboriginal and Torres Strait Islander people genetics, genomics, and clinical phenotype diagnostic support tools[44];
- many Aboriginal and Torres Strait Islander people have Indigenous language/s as their first language/s. As a result, diagnostic and clinical concepts, including around emerging health technologies, can be challenging for non-Indigenous Australian practitioners to adequately convey[45];
- more work is required to understand the health information needs of Aboriginal and Torres Strait Islander people with rare diseases (and their families and kinship groups) to ensure available information and services are both culturally safe and appropriate[46]; and
- proportionately, more Aboriginal and Torres Strait Islander people live in regional, rural and remote areas, which can pose significant challenges to their ability to access services[47].

People living in regional, rural and remote areas

Where people live can have a significant impact on their ability to access services. The lack of rare disease expertise nationally is accentuated by Australia's vast size. Most rare disease expert centres are located in capital cities thus travel creates a financial barrier for regional, rural and remote Australians living with a rare disease and their families. Australians living in rural and remote areas experience poorer health and welfare outcomes, including higher rates of chronic disease, disease burden and mortality (death)[48] compared to those living in metropolitan areas. This inequity is likely exacerbated when people living in regional, rural and remote areas are also living with a rare disease as they have lower access to health services, and are more likely to present to hospital with conditions that could have been treated by a primary health care practitioner[49]. For Australians living with a rare disease, including an undiagnosed rare disease, living in regional, rural and remote areas may have implications in terms of:

- receiving a timely and accurate diagnosis;
- continuity of care and access to appropriate treatments; and
- having higher exposure to modifiable risk factors, such as smoking, risky alcohol consumption, not getting enough exercise, and being overweight or obese[50]. These risk factors are associated with adverse pregnancy outcomes, and their modification has been identified by EUROCAT, European Surveillance of Congenital Anomalies, as being important in the primary prevention of rare congenital anomalies[51].

People from CALD backgrounds

Both language barriers and cultural differences can have negative impacts on the way people from CALD backgrounds access and experience health care and support services[52]. Information is often not available in all languages, and cultural norms around health and support, including mental health, can vary significantly[53]. Furthermore, some rare diseases are more prevalent in people from certain CALD backgrounds, such as thalassaemia in Australians of Mediterranean origin.[54]

People experiencing socio-economic disadvantage

The experience of socio-economic disadvantage can have significant, cumulative effects on a person's health and wellbeing, including their ability to access and maintain engagement with services[55]. Many people living with a rare disease, including families and carers, report psychological, emotional and financial impacts[56]. Combining these two effects highlights the need for additional consideration to be made for this group.

Partnerships

The effective prevention and management of chronic conditions, including rare diseases, is strongly influenced by the contributions made by a wide range of Partners. These Partners include:

- individuals, carers and families;
- communities;
- all levels of government;
- non-government organisations;
- the public and private health sectors, including all health care providers and private health insurers;
- industry; and
- researchers and academics.

All Partners have shared responsibility for health and wellbeing outcomes according to their role and capacity within the health care and social systems. Greater cooperation between Partners will lead to more successful individual and system outcomes. Actions included in this Action Plan are intended to guide Partner investment in the prevention and management of rare diseases and should be implemented collaboratively to achieve the best health and wellbeing outcomes.

Pillar 1: Awareness and Education

Why is this important?

Increased awareness and education at the individual and community level is vital. It is common for people to have never heard of the rare disease with which they, or their child, are diagnosed. It can be difficult to find a practitioner who is educated about the disease. Yet, people living with a rare disease, and their families and carers, are reliant on services for both care and support. Lack of awareness of rare diseases often contributes to people feeling isolated and misunderstood, as well as to delays in diagnosis and treatment, potentially missing opportunities for early intervention and improved outcomes[57].

While awareness is important, with thousands of different rare diseases, it is impossible for any individual (including health professionals) to be aware of them all. Awareness activities must be supported by systematic identification, classification, and a prioritised response to rare diseases and undiagnosed rare diseases[58]. This would mean people do not need to know everything about all rare diseases, but rather, would know how to find relevant information as it is needed. Rare diseases data collection and use is explored further in Pillar 3.

Rare disease organisations currently play a key role in raising disease awareness and providing critical person-centred information. These organisations are vital to the rare disease sector and often fill gaps in the system, not just in terms of awareness and education, but also care and support and, increasingly, in the research sphere[59]. However, stakeholder consultations undertaken during the development of the Action Plan identified that these organisations are under-resourced, are largely volunteer-based and often have a limited ability to raise funds, posing a risk to their long-term sustainability.

Education about rare diseases needs to empower people living with a rare disease to become active participants in their rare disease journey. Education needs to respond to the fact that people living with a rare disease are constantly learning and at the same time, teaching others about their disease[60].

There is also an urgent need for focused education of the workforce that supports people living with a rare disease to increase its capacity to meet care and support requirements. While this may apply to the care and support workforce broadly, there are certain segments of the workforce, such as those involved in mental health care, with known and urgent awareness and education needs[61].

What we hope to achieve:

- Increase every Australian's awareness of rare diseases including, where applicable, relevant prevention measures.
- Ensure Australians living with a rare disease have access to information and education that enables them to be active participants in their rare disease journey.
- Develop a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics.

Priority 1.1

Increase every Australian's awareness of rare diseases including, where applicable, relevant prevention measures.

Action 1.1.1

Develop and conduct national awareness and education activities for rare diseases.

Implementation

1.1.1.1. Analyse existing resources and distribution channels and build on these to coordinate national media and communications material that promotes and distributes the latest information relating to rare diseases. Responsibility for deliverables will be identified as part of this process.

1.1.1.2. Ensure national media and communications material highlights challenges common across rare diseases. Market research will determine key messaging and how material is delivered. For example, it may be grouped for all rare diseases or tailored for individual rare diseases.

1.1.1.3. Address urgent funding gaps for rare disease organisations to enable them to sustain and expand upon current awareness and education activities. These may include hosting information sessions, workshops, conferences or sending e-newsletters.

1.1.1.4. Collaborate with targeted stakeholders to maximise the reach and appropriateness of materials to Aboriginal and Torres Strait Islander people, those with CALD backgrounds, and other priority populations.

Action 1.1.2

Raise awareness of, and deliver, relevant prevention measures for non-hereditary rare diseases, such as cancers, infections and autoimmune disorders.

Implementation

1.1.2.1. Governments, health care services and stakeholder organisations raise awareness of, and educate about, relevant prevention measures for non-hereditary rare diseases, including through national and localised media and communications material.

1.1.2.2. Governments and health care services deliver relevant prevention measures to reduce the incidence of non-hereditary rare diseases. An example is activity targeted at geographic locations with low whooping cough vaccination rates.

Action 1.1.3

Develop, deliver and promote targeted awareness and education activity to support people in their preparation for conception and pregnancy.

Implementation

1.1.3.1. Governments and health care services raise awareness of rare diseases and educate people preparing for conception and pregnancy. This is incorporated into existing pre-conception and perinatal care. This could include education and awareness pertaining to both preventive measures and rare diseases testing and screening opportunities, such as:

- Pre-conception (carrier) testing and screening;
- Pre-implantation genetic diagnosis (testing) and screening;
- Ante-natal testing and screening; and
- Newborn testing and screening.

1.1.3.2. Governments and health care services support people in their preparation for conception and pregnancy through evidence-based, high-quality pre-conception and ante-natal care, including vaccinations. An example is educating women of child-bearing age about the benefits of vaccination against Rubella (German measles) prior to conception to prevent the development of congenital abnormalities.

1.1.3.3. Develop and promote non-directive education materials for use by individuals and families following access to the range of genetic testing or screening opportunities for rare diseases. These materials are to be developed in partnership with health care professionals and the public to identify the ethical, legal, social and other issues families may wish to take into consideration.

Priority 1.2

Ensure Australians living with a rare disease have access to information and education that enables them to be active participants in their rare disease journey.

Action 1.2.1

Raise awareness among people living with a rare disease, and their families and carers, about the care and support services available to them.

Implementation

1.2.1.1. Develop and maintain an accessible multi-purpose digital repository to detail available care and support services and to provide general rare diseases information. The repository can be used to identify gaps and opportunities for improvement. Promote the repository to rare disease organisations, for distribution to people living with a rare disease, and their families and carers.

1.2.1.2. Build on existing activities of rare disease organisations to raise awareness of care and support services available to people living with a rare disease, and their families and carers. Identify gaps and opportunities for improvement.

Action 1.2.2

Improve consultation and communication between policy-makers and the rare disease community.

Implementation

1.2.2.1. Rare disease organisations strengthen their connections with policy-makers. This would build on current coordination by existing national and state-based collaborations.

1.2.2.2. Further articulate the consumer voice through the facilitation of an advisory group to improve consultation and communication on a range of issues including disability, health, housing, education and employment.

Priority 1.3

Develop a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics.

Action 1.3.1

Develop a national rare disease workforce strategy.

Implementation

1.3.1.1. Identify existing gaps in the workforce that support people living with a rare disease and outline a path towards a sustainable workforce, taking into account the impact of genomics.

1.3.1.2. Ensure collaboration and consultation occurs between implementation Partners, including education providers, professional bodies, and other key stakeholders.

1.3.1.3. Ensure the strategy includes measures to empower practitioners to provide culturally safe and appropriate care and support to Aboriginal and Torres Strait Islander people, those with CALD backgrounds, and other priority populations.

Action 1.3.2

Equip and encourage frontline health professionals to consider, investigate and refer for a potential rare disease diagnosis.

Implementation

1.3.2.1. Develop and promote guidelines for Australia in line with the International Joint Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients[62]. These guidelines will provide support for clinicians in identifying possible rare disease in people who present with complex symptoms. They will also articulate the key role that health professionals play in meeting the support needs of individuals and families through the diagnostic journey.

1.3.2.2. Through relevant professional peak bodies, promote the use of the accessible multi-purpose digital repository by health professionals. This will support health professionals to consider a rare disease diagnosis when people present with complex and unexplained symptoms.

1.3.2.3. Develop awareness and education that empowers frontline health professionals to provide culturally safe and appropriate care and support to Aboriginal and Torres Strait Islander people, those with CALD backgrounds, and other priority populations.

Pillar 2: Care and Support

Why is this important?

Early diagnosis enables the best clinical care, treatment options, access to services, peer support, increased reproductive confidence and access to participation in clinical trials. Yet diagnostic delay and misdiagnosis is common in rare diseases. Thirty per cent of Australian adults living with a rare disease are impacted by a diagnostic delay of more than five years, while almost half have received at least one misdiagnosis.[63] This has physical, psychological, emotional and financial costs for the person and family living with a rare disease[64].

There is a real need for rare disease care and support to be less fragmented and more integrated[65]. This care and support needs to be both person and family-centred. Multiple stakeholder consultation processes, including the consultations undertaken as part of the development of the Action Plan, have confirmed that living with a rare disease does not only affect a person's health; it impacts every facet of their life, including education, employment and mental and physical health[66,67,68]. For example, it is currently difficult for people living with a rare disease to navigate their way through the health and disability systems as there is a lack of clear referral pathways[69,70]. Effective care and support must respond to this (Appendix 3 provides further detail on rare disease care and support.)

Stakeholder consultations undertaken as part of the development of the Action Plan confirmed that valuable care and support is provided by rare disease organisations in Australia. This includes peer support, the provision of information, access to resources, and individual and systemic advocacy. Many rare disease organisations are run by people living with their own rare disease challenges, which can affect their ability to offer services (including in awareness, education, care, support and research) and impacts their sustainability[71].

There are limited treatment options for rare diseases[72], and even when a treatment does exist, financial support may not be available in Australia and thus accessibility may be limited[73]. Reimbursement of health technologies for rare diseases, using models designed for more common diseases, is challenging as smaller patient numbers impact cost effectiveness[74], and there is often less clinical evidence available due to the challenges of conducting large-scale clinical trials[75]. This highlights the importance of alternative approaches to both identifying treatment options and funding health technologies for rare diseases[76].

There are many examples of an approved medicine (for a more common condition) demonstrating benefits for rare diseases[77]. However, due to small numbers, it is not always commercially viable for companies to seek reimbursement for a rare diseases indication[78]. Without government reimbursement, many rare diseases medicines are unaffordable for people living with a rare disease and their families. As many rare diseases are progressive, time is often critical, making timely and equitable reimbursement essential for people living with a rare disease to benefit from new and transformative health technologies[79].

What we hope to achieve:

- Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family-centred (see Appendix 3).
- Ensure diagnosis of a rare disease is timely and accurate.
- Facilitate increased reproductive confidence.
- Enable all Australians to have equitable access to the best available health technology.
- Integrate mental health, and social and emotional wellbeing, into rare disease care and support.

Priority 2.1

Provide rare disease care and support that is integrated and appropriate for all Australians living with a rare disease, while being both person and family-centred.

Action 2.1.1

Provide rare disease care and support that is integrated, incorporating clear pathways throughout health, disability and other systems.

Implementation

2.1.1.1. Establish standards for care and support that are integrated and incorporate clear pathways throughout all systems. Ensure these are informed by clinical and consumer rare disease experts and that such consultation informs policy development.

2.1.1.2. To reduce fragmented care, ensure policy meets people's full range of needs, including health, disability and education. Support this work with a cross-jurisdictional, cross-sectoral working party.

2.1.1.3. Increase the utilisation of digital health, including virtual clinics and telehealth (telemedicine) services. Leverage existing infrastructure, such as My Health Record, to support care and improve integration.

2.1.1.4. Ensure care and support is responsive to the specific needs of rural and remote communities and health services, Aboriginal and Torres Strait Islander people, those with CALD backgrounds, and other priority populations.

Action 2.1.2

Build a broad range of care and support services that are responsive to the changing needs of people living with a rare disease and their families.

Implementation

2.1.2.1. Develop an accessible multi-purpose digital repository, incorporating elements targeted at the workforce that supports people living with a rare disease. With access to adequate information, health care and social support professionals will be equipped to support people living with rare disease and their families to navigate health, disability and other systems.

2.1.2.2. Strengthen the National Disability Insurance Agency's response to the nature of disability caused by rare disease that can manifest as chronic, intermittent and often progressive. Initial implementation should prioritise:

- fast tracking access to the NDIS; and
- ensuring NDIS participants can access an appropriate range of respite to meet the needs of families.

2.1.2.3. Through regular stakeholder consultations, determine strategies to improve access to rare disease care and support services for Aboriginal and Torres Strait Islander people, those with CALD backgrounds, those living in rural and remote areas, and other priority populations.

Action 2.1.3

Ensure services support people living with a rare disease through significant life-stage transitions.

Implementation

2.1.3.1. Enhance existing transition services to ensure people living with a rare disease experience seamless transitions between services as they move through life stages. Common transitions include:

- reaching the age cut-off point for paediatric services (e.g. transitioning from child to adult hospitals);
- relocating; and
- when needs change significantly (such as at end-of-life).

2.1.3.2. Increase awareness among health and other professionals, and rare disease organisations of the multi-faceted role of palliative care, including peri-natal palliative care. Palliative care can achieve a range of objectives for people and families living with a rare disease, including improving quality of life and providing end-of-life care. This will assist to improve:

- uptake of palliative care by people living with a rare disease or by families who may benefit from peri-natal palliative care; and
- timeliness of referrals by health professionals.

2.1.3.3. Promote the specific needs of people living with a life-limiting rare disease to the palliative care sector.

2.1.3.4. Ensure transition services are culturally safe and appropriate, in recognition of unique life-stage challenges faced by Aboriginal and Torres Strait Islander people.

Action 2.1.4

Develop the capacity of rare disease organisations to represent and advocate for people living with a rare disease and their families.

Implementation

2.1.4.1. Rare disease organisations represent and advocate for people living with a rare disease and their families through a range of activities including:

- written submissions;
- consumer hearings;
- communicating with their community;
- representing their community to stakeholders, such as government and industry; and
- advocating for reimbursement of health technologies after independent health technology assessment (HTA) has demonstrated effectiveness (see Appendix 4 for more on HTA).

Further support the current activities of rare disease organisations through additional resourcing as well as further national collaboration.

2.1.4.2. Ensure consultation with targeted stakeholders to strengthen the capacity of rare disease organisations to appropriately represent and advocate for Aboriginal and Torres Strait Islander people living with a rare disease and their families.

Action 2.1.5

Embed the voice of people living with a rare disease and their families and carers throughout structures and systems that impact rare diseases.

Implementation

2.1.5.1. Capture and promote the voice of people living with a rare disease and their families and carers by:

- involving people living with a rare disease at every level of decision-making;
- ensuring ongoing engagement to capture broader input from people living with a rare disease through surveys, focus groups, newsletters and representation on boards; and
- calling for key structures and systems to routinely and effectively capture broader input from consumers, as done currently in some research settings and HTA (consumer hearings).

2.1.5.2. Enhance culturally safe and appropriate approaches for Aboriginal and Torres Strait Islander people, including aligning with existing initiatives to develop and implement ways to integrate Indigenous Australian languages to equitably enhance care and support.

Priority 2.2

Ensure diagnosis of a rare disease is timely and accurate.

Action 2.2.1

Ensure all Australians have equitable access to a range of diagnostic tools and tests, providing the best chance of early and accurate diagnosis.

Implementation

2.2.1.1. Further the development of, and investment into, the range of existing specialist diagnostic responses, such as genomics technology, including for Aboriginal and Torres Strait Islander people; interdisciplinary undiagnosed disease programs; clinical phenotype diagnostic support tools; centres of expertise; genetic counsellors and peer support groups.

2.2.1.2. Ensure all existing screening and testing programs are sustainable and evolve in line with innovation over time.

2.2.1.3. Enhance culturally safe and appropriate approaches for Aboriginal and Torres Strait Islander people, including aligning with existing initiatives to develop and implement ways to integrate Indigenous Australian languages to equitably enhance diagnosis.

Action 2.2.2

Develop policy that supports the implementation of diagnostic tools and tests.

Implementation

2.2.2.1. Support national leadership and coordination of a range of screening and diagnostic tools and tests jointly funded by Commonwealth and state/territory governments, to enable more consistent service and equitable access.

2.2.2.2. Address urgent funding gaps associated with the effective implementation and sustained success of the Newborn Bloodspot Screening (NBS) National Policy Framework.

2.2.2.3. Ensure equity of access to genetic testing and counselling, and referral to genetic peer support groups. Genetic testing plays a critical role in diagnosing rare diseases, while genetic counselling and peer support groups provide support to the individuals undergoing testing.

2.2.2.4. Develop non-directive education materials for use by individuals and families following access to genetic testing. These materials are to be developed in partnership with health care professionals and the public to identify the ethical, legal, social and other issues that individuals and families may wish to take into consideration in their decision-making.

2.2.2.5. Develop comprehensive, best-practice support standards and materials that address the diverse possibilities that may arise from individuals and families undertaking genetic testing.

Action 2.2.3

People with an undiagnosed rare disease are identified and have priority access to the most appropriate specialised and expert diagnostic response.

Implementation

2.2.3.1. Flag in health information systems when someone presents with an undiagnosed rare disease.

2.2.3.2. Develop guidelines for Australia in line with the International Joint Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients[80]. This will provide support for clinicians in:

- identifying possible rare disease in people who present with complex symptoms;
- outlining best practice and timely diagnostic pathways. Eg. including best practice around reducing wait times for genetic counselling appointments; and
- ongoing management of the person with a suspected undiagnosed rare disease, in order to aid diagnosis.

2.3.3.3. Increase the capacity and reach of the existing state-based best-practice undiagnosed disease program models to achieve national coverage.

Action 2.2.4

Support people with a suspected but undiagnosed rare disease on their diagnostic journey.

Implementation

2.2.4.1. Undertake a survey on existing support for people with an undiagnosed rare disease delivered by rare disease organisations, to identify gaps and opportunities for improvement.

2.2.4.2. Address gaps and opportunities for improvement, identified through the survey, by funding awareness raising and support activities conducted by SWAN Australia and other relevant organisations.

2.2.4.3. Rare disease organisations collaborate to develop supporting material that describes best-practice approaches to delivering this support, taking into account the great diversity of rare disease journeys.

2.2.4.4. Ensure consultation with targeted stakeholders to maximise appropriateness of this support for Aboriginal and Torres Strait Islander people, those with CALD backgrounds, and other priority populations.

Priority 2.3

Facilitate increased reproductive confidence.

Action 2.3.1

Ensure individuals and families known to have an increased chance of being carriers of genetic variants for rare diseases have equitable access to peri-conception genetic testing and counselling, which can provide them with information about becoming pregnant and pregnancy.

Implementation

2.3.1.1. Further to the National Health Genomics Policy Framework and Pregnancy Care Guidelines, develop consistent and comprehensive clinical guidelines for all relevant health professionals to support individuals and families to access peri-conception genetic testing, counselling and peer support groups.

2.3.1.2. Ensure equity of access for all Australians with an increased chance of being carriers of genetic variants for rare diseases to peri-conception genetic testing and counselling. Promote connection to genetic peer support groups for further support.

2.3.1.3. Develop non-directive education materials for use by individuals and families surrounding access to peri-conception genetic testing. These materials are to be developed in partnership with health care professionals and the public to identify the ethical, legal, social and other issues that individuals and families may wish to take into consideration in their decision-making.

2.3.1.4. Continue to support individuals and families through the range of possible outcomes following peri-conception genetic testing.

Action 2.3.2

Women who have, or are at risk of developing, certain chronic conditions, such as diabetes, epilepsy or thyroid disorders, have an increased chance of having babies with rare congenital anomalies. Provide these women with access to evidence-based, high-quality pre-conception and peri-natal care.

Implementation

2.3.2.1. Governments and health care services provide women planning a pregnancy with increased and equitable access to a pre-conception consultation program to investigate whether they have, or are at risk of developing, certain chronic conditions, such as diabetes, epilepsy or thyroid disorders, that may affect their pregnancy.

2.3.2.2. Governments and health care services support these women to access evidence-based, high-quality pre-conception or pregnancy care and support when they are planning a pregnancy or already pregnant. This includes equitable access to the range of rare diseases testing and screening opportunities such as:

- ante-natal testing and screening; and
- newborn testing and screening.

2.3.2.3. Address urgent funding gaps in the NBS National Policy Framework for effective implementation and sustained success, including:

- equipment costs;
- ongoing secretariat support of the NBS Program Management Committee;
- funding for the national decision-making process;
- implementation of new screening tests recommended through the national decision-making process;
- funding for monitoring and evaluation of programs; and
- national communication and education resources.

Priority 2.4

Enable all Australians to have equitable access to the best available health technology.

Action 2.4.1

Develop policy that supports people living with a rare disease to have timely and equitable access to new and emerging health technologies.

Implementation

2.4.1.1. Broaden the description and understanding of the principles underpinning Australian HTA processes to acknowledge the challenges associated with assessing health technologies for rare diseases.

2.4.1.2. Align with and build on the existing National Health Genomics Policy Framework for the systematic, equitable and timely delivery of genomic services, such as genetic testing (diagnostics) and gene therapies (treatments) and genetic counselling to Australians with, suspected of having, or with an increased chance of a rare disease.

2.4.1.3. Develop comprehensive, best-practice support standards and materials to ensure timely delivery of high-quality care by genetic services.

2.4.1.4. Build rare disease expertise within the Office of Health Technology Assessment (OHTA) that is responsible for analysing potential rare disease impacts.

2.4.1.5. Ongoing review of health technology policy in line with advancements in health technology. For example, mitochondrial donation involves removing the nuclear DNA from a woman's egg containing faulty mitochondria and inserting it into a healthy donor egg, which has had its nuclear DNA removed. This prevents mitochondrial DNA defects from being inherited by a genetically related offspring. Mitochondrial donation is not yet legal in Australia.

Action 2.4.2

Ensure funding and reimbursement pathways are fit-for-purpose and sustainable for current and new health technologies for rare diseases.

Implementation

2.4.2.1. Build on the current processes within the OHTA to ensure all rare diseases submissions are flagged as complex and may require additional scoping and engagement to address potential challenges and uncertainties.

2.4.2.2. Raise awareness among industry and rare disease organisations as to the availability of the HTA Access Point.

2.4.2.3. Ensure rare disease expertise exists, or can be accessed, on all reimbursement pathways and HTA advisory bodies.

Action 2.4.3

Ensure people living with a rare disease have equitable access to medicines with demonstrated clinical benefit for a rare disease, including those that are already funded for another condition.

Implementation

2.4.3.1. Ensure the HTA Consumer Evidence and Engagement Unit provides education and support to people living with a rare disease and their families and carers, and/or rare disease organisations to support them to take a more active role in HTA processes.

2.4.3.2. Rare disease organisations work with the HTA Consumer Evidence and Engagement Unit to submit an application for public reimbursement of a technology eligible for assessment by the OHTA.

2.4.3.3. The TGA and OHTA continue to work together to develop clear processes and pathways for sponsors considering submitting applications for the repurposing of medicines already approved for use in treatment of other conditions.

Priority 2.5

Integrate mental health, and social and emotional wellbeing, into rare disease care and support.

Action 2.5.1

Ensure people living with a rare disease, including their families and carers, receive the community, clinical and digital mental health supports and services they need.

Implementation

2.5.1.1. Enable people living with a rare disease, including their families and carers (with appropriate consent) to access information and resources (including digital) customised for rare diseases as part of Chronic Disease Management Plans and Mental Health Care Plans.

2.5.1.2. Ensure Aboriginal and Torres Strait Islander people living with a rare disease have access to customised resources (including digital), in recognition of the greater challenges to achieving the best possible social and emotional wellbeing support outcomes for Aboriginal and Torres Strait Islander people.

Action 2.5.2

Implement care and support systems to address the mental health and wellbeing of Australians impacted by a rare disease.

Implementation

2.5.2.1. Empower rare disease care and support providers to deliver the best possible mental health and social and emotional wellbeing support outcomes through a range of initiatives, including:

- access to evidence that aids providers in their understanding of and ability to respond to mental health and social and emotional wellbeing support needs, such as a rare disease mental health checklist;
- awareness around the existing range of free or low cost digital mental health services that provide support, such as Head to Health;
- education about how to access and utilise these services; and
- cultural competency education that empowers providers to effectively support Aboriginal and Torres Strait Islander people.

2.5.2.2. Provide mental health care that recognises the unique challenges associated with rare diseases via existing systems, such as Chronic Disease Management Plans and Mental Health Care Plans.

Action 2.5.3

Develop the capacity of rare disease organisations to provide wellbeing and mental health support.

Implementation

2.5.3.1. Better resource existing social and emotional wellbeing support provided by rare disease organisations including:

- peer support;
- family days;
- community engagement; and
- information sessions and workshops.

2.5.3.2. Provide education and training to rare disease organisations to increase their awareness of mental health issues and guide people to seek further support.

2.5.3.3. Undertake targeted stakeholder consultations to ensure appropriate social and emotional wellbeing support, including appropriate referral to GPs, for Aboriginal and Torres Strait Islander people, those with CALD backgrounds, those living in rural and remote areas, and other priority populations.

Pillar 3: Research and Data

Why is this important?

In Australia, data for most rare diseases is not captured in either health information systems or registries[81] and there is no coordinated strategy to collect, measure, build and translate data that does exist. Multiple research papers and stakeholder consultation processes in Australia have identified the need for a national, coordinated, and systematic approach to the collection and use of rare diseases data, including registries[82,83,84,85]. Such an approach will enable monitoring and the accumulation of knowledge about rare diseases to inform clinical practice, research and health service planning.[86]

For key decision-makers at all levels, greater knowledge of rare diseases can facilitate more responsive and appropriate services for people living with a rare disease and their families and carers. This Action Plan responds to the significant potential for positive change in this space.

For many rare diseases, there are a number of barriers to effective research and no active research programs. One of the biggest challenges is that rare diseases have small patient numbers and are often very complex.[87] Depending on the specific rare disease, research priorities can be different. For example, while funding for translational research may be important for many rare diseases, some rare diseases are not yet in the position to prioritise translational research. For some rare diseases, the unmet research needs are basic discovery research or investment into data collection and natural history studies.

Investment into all types of research related to rare disease is needed. This research includes:

- rare disease coding;
- data collection and registries;
- fundamental discovery research (also known as basic research);
- qualitative research;
- pre-clinical testing; and
- clinical trials.

According to the International Rare Diseases Research Consortium Goals for 2017–2027, people living with a rare disease need research into:

- diagnostics (including genomics);
- development and testing of new health technologies;
- precision or personalised medicine; and
- care and support[88].

For many people living with a rare disease, participation in a clinical trial may be the only way to access treatment[89]. A 2016 Australian study found that almost 90 per cent of respondents living with a rare disease were interested in joining a patient registry, in recognition of the key role that registries play in linking people living with a rare disease with clinical trials for new health technologies (drug treatments and therapies)[90]. The translation of rare diseases research into clinical settings, while currently hampered[91], is vital. This two-way relationship benefits from active participation by patients, their families and carers, and patient advocacy groups to ensure the best outcomes for people living with a rare disease[92].

There is an understanding in the Australian rare disease community that, while research may not lead to better outcomes for people currently living with a rare disease, participating in research may drive change for future generations. This is supported by outcomes of the Rare Barometer survey undertaken in February 2018 by EURORDIS, Rare Diseases Europe[93].

Research into rare diseases must address existing gaps and the coordination of research projects must be prioritised[94]. Improved policy settings, and national and international collaborations, will help to drive strong research and innovation for all rare diseases[95]. Research into rare diseases needs to inform evidence-based policy across all systems, extending beyond health to incorporate disability, social/welfare, mental health, education, employment and housing.

What we hope to achieve:

- Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning.
- Develop a national research strategy for rare diseases to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps.
- Ensure research into rare diseases is collaborative and person-centred.
- Translate research and innovation into clinical care; clinical care informs research and innovation.

Priority 3.1

Enable coordinated and collaborative data collection to facilitate the monitoring and cumulative knowledge of rare diseases, informing care management, research and health system planning.

Action 3.1.1

Health information systems identify and measure rare diseases and undiagnosed rare diseases.

Implementation

3.1.1.1. The Australian Institute of Health and Welfare (AIHW) re-establishes the Australian National Congenital Anomalies Register (NCAR), including rare disease coding (Orphacodes). This will accelerate, extend and nationalise rare disease coding already underway in the Western Australian Register of Developmental Anomalies (WARDA), and contribute to International Classification of Diseases 11th Revision (ICD-11) preparedness.

3.1.1.2. Develop a nationally-recognised definition of undiagnosed rare diseases in consultation with relevant experts. Using this definition, provide for an undiagnosed rare disease code in an individual's health record that is compatible with Orphacodes, ICD-11 and other relevant classifications. This code could:

- raise a flag or alert to health professionals when they access the individual's health record (similar to drug allergy alerts), thus prioritising a diagnostic response and;
- support data collection for undiagnosed rare diseases, and hence strategic decision-making, such as service planning.

3.1.1.3. Provide for rare disease codes in patient records that are compatible with Orphacodes, ICD-11 and other relevant classifications. This code could:

- raise a flag or alert to health professionals when they access the individual's health records, thus leading to appropriate care that takes into account the rare disease diagnosis and;
- support data collection for rare diseases, and hence strategic decision-making.

3.1.1.4. Ensure rare disease and undiagnosed rare disease codes link with a person's Aboriginal and Torres Strait Islander status to allow for culturally appropriate care, and to build evidence of rare disease epidemiology among Aboriginal and Torres Strait Islander people.

Action 3.1.2

Undertake broad epidemiological surveillance of rare diseases to support decision-makers to access the information they need to improve the health and wellbeing of Australians living with a rare disease.

Implementation

3.1.2.1. Building on existing newborn screening and congenital anomalies data collections, further develop Australia's monitoring of rare diseases and undiagnosed rare diseases. Examples may include and extend beyond:

- newborn and paediatric age ranges; and
- the rare diseases currently captured in newborn screening and congenital anomalies data collections.

3.1.2.2. Establish a dedicated Rare Disease Office within the AIHW that publishes periodic national reports on the epidemiology of rare diseases and undiagnosed rare diseases in Australia, including among Aboriginal and Torres Strait Islander people.

Action 3.1.3

Improve rare disease data collection and use, including best-practice safe storage, data sharing, custodianship, analysis, reporting and privacy requirements.

Implementation

3.1.3.1. Establish a dedicated Rare Disease Office within the AIHW. Included in its remit will be systematic improvements in rare disease data integration and interoperability.

3.1.3.2. Publish appropriate data collected through post-market surveillance mechanisms, including under the LSDP to enable better data use and the accumulation of rare diseases knowledge.

Action 3.1.4

Develop a national approach to person-centred rare disease registries to support national standards, best practice and minimum data sets.

Implementation

3.1.4.1. Develop a summary report of all existing Australian and relevant international rare disease registries, collecting information on:

- governance standards;
- management practices;
- data sets, including patient numbers, estimated incidence, prevalence and coverage; and
- classification systems used (for interoperability with other registries and health information systems).

This information will support national coordination of rare disease registries and the establishment of minimum data sets, provide a better understanding of who is currently being counted and aid identification of best practice.

3.1.4.2. Further develop and resource the existing RVA-led National Alliance of Rare Disease Registries to encourage collaboration, shared knowledge, standardisation, alignment with research initiatives and best practice.

Priority 3.2

Develop a national research strategy for rare diseases to foster, support and drive all types of research for rare diseases, contributing to agreed priorities and systematically addressing gaps.

Action 3.2.1

Develop a national research strategy for rare diseases, to keep pace with genomic advancements, precision medicine and innovation.

Implementation

3.2.1.1. Undertake a national stakeholder consultation process to set agreed priorities for a national research strategy for rare diseases, including:

- surveys;
- public forums;
- targeted themed roundtables; and
- opportunities for public submissions.

3.2.1.2. Develop a national research strategy for rare diseases, building in regular reviews.

Action 3.2.2

Proactively address evidence gaps in areas that are important to people living with a rare disease.

Implementation

3.2.2.1. Ensure lived experience drives research by encouraging collaboration between researchers and people living with a rare disease through workshops, conferences and consumer reference groups.

3.2.2.2. Research funding bodies identify and report on research related to rare diseases.

3.2.2.3. In response to limited evidence and in line with open publishing (also known as open access initiatives), share outputs from research, including publications and data.

3.2.2.4. Prioritise and encourage fundamental discovery research for rare diseases through research funding. This is in recognition of its central importance to the development and testing of much-needed innovation in health technology for rare diseases.

Action 3.2.3

Support collaborative research into rare diseases in Australia and internationally.

Implementation

3.2.3.1. Encourage and facilitate greater research collaboration nationally, internationally and with industry. Examples of how this may be achieved include:

- financial incentives for research teams that can demonstrate collaboration with national, international and industry partners; and
- the development of customised research grants for rare diseases that require a degree of collaboration with national, international and industry partners.

Action 3.2.4

Building on existing initiatives, continue to foster an environment conducive to clinical trials for rare diseases taking place in Australia.

Implementation

3.2.4.1. Develop recommendations to encourage and enable more clinical trials for rare diseases to take place in Australia.

3.2.4.2. Increase the economies of scale of research into rare diseases by, for example, operating multi-trial sites that share common resources.

3.2.4.3. Encourage the adoption of unique and appropriate trial designs that overcome rare disease research challenges.

Action 3.2.5

Investigate and promote options that enable Australians living with a rare disease to participate in clinical trials and other research activity, both in Australia and internationally (without needing to leave Australia).

Implementation

3.2.5.1. Identifying and maximising utilisation of available resources and assets to the extent possible, link people living with a rare disease to research activity, such as data collection, registries, natural history studies, qualitative research and clinical trials based in Australia and internationally.

3.2.5.2. Investigate and promote options for a Trials Enabling Program (TEP)[96] for trials for rare diseases in Australia, leveraging a partnership approach that involves philanthropy and industry in the absence of relevant clinical trials in Australia.

Priority 3.3

Ensure research into rare diseases is collaborative and person-centred.

Action 3.3.1

Provide people living with a rare disease or an undiagnosed rare disease with the opportunity and support to participate in research.

Implementation

3.3.1.1. Health professionals inform and connect people living with a rare disease to research as part of their ongoing care.

3.3.1.2. Develop opportunities for individuals to share their lived experience to contribute to research. Rare disease organisations can promote this by increasing their liaison with researchers and clinicians, and by disseminating information.

3.3.1.3. Promote culturally safe and appropriate approaches for Aboriginal and Torres Strait Islander people.

Action 3.3.2

Enable researchers, funders and policy-makers to access the voice of people living with a rare disease in driving and delivering research into rare diseases.

Implementation

3.3.2.1. Develop and support consumer reference groups to promote additional pathways for researchers, research funders, policy-makers and other decision-makers to be informed about the rare disease community's needs and priorities. Wherever possible, leverage and build on the expertise and resources of existing groups and mechanisms.

3.3.2.2. The rare disease sector promotes the importance of a person-centred approach to research, and the mechanisms to achieve this, including co-design.

Priority 3.4

Translate research and innovation into clinical care; clinical care informs research and innovation.

Action 3.4.1

Support partnerships between researchers and clinicians in research into rare diseases.

Implementation

3.4.1.1. Research funding bodies prioritise research proposals and applications for rare diseases that can demonstrate support from, and close working relationships with, clinicians.

3.4.1.2. Support and foster interdisciplinary research teams to encourage more person-centred research, and a dual focus on research and clinical care where appropriate.

Action 3.4.2

Identify, leverage and enhance existing capability and infrastructure to ensure appropriate and experienced resourcing is available within clinical teams that deliver rare disease care.

Implementation

3.4.2.1. In partnership with industry, philanthropy and trial sites, identify and enhance existing capability and infrastructure within clinical centres to ensure appropriate capability is available to support the operation of clinical trials for rare diseases.

3.4.2.2. Support clinical teams to collect and input data, contributing to research and evidence-building.

Achieving Progress

This Action Plan and its Priorities, Actions and Implementation activity has been informed by stakeholder consultation with the rare disease sector, from which a number of key themes emerged. The Action Plan has been developed 'by the rare disease sector, for the rare disease sector'. The themes detailed below will become its measures of progress over time.

Theme #1
The need for national leadership, coordination and consistency

This theme emerged most frequently from the consultation process, with participants calling for:

- **A national plan for Australia that is in line with global standards**
  Australia remains in danger of falling further behind many countries already tackling rare diseases through policy and legislation. This Action Plan is an opportunity for Australia to adopt a national plan for rare diseases that aligns with global standards. The value of a nationally coordinated plan cannot be underestimated.
- **Annual implementation plan**
  Existing plans for rare diseases, including those in Europe and the UK, are accompanied by an implementation plan. In Australia, this plan should be developed collaboratively by the sector and could be led by the existing national peak body for rare diseases. Progress in implementation should be regularly monitored and reviewed.
- **Ongoing stewardship and policy sustainability**
  Ongoing stewardship of the Action Plan is critical to ensure policy change is long-lasting and sustainable. In the US, UK and many European countries, the sustainability of rare diseases policy is enshrined in legislation[97].

Theme #2
The need to prioritise the systematic building of knowledge, evidence and expertise

There is urgent need for the expansion of rare disease expertise and further development of evidence-based rare disease care. Systems must actively respond to existing evidence gaps. Processes that will build knowledge and evidence both quickly and sustainably must be prioritised. Clearer pathways through health and other systems are a necessity.

Throughout the consultation process, stakeholders consistently raised the need for centres of excellence for rare diseases. Currently, rare disease clinics and research institutes with a focus on rare diseases are significantly under-resourced, and often work in isolation. To achieve real progress, existing strengths must be built upon to formalise a network of centres of excellence that is appropriate and accessible for all Australians.

Theme #3
The need for a person-centred approach and ongoing collaboration

To be successful, this Action Plan must progress meaningful involvement of people living with a rare disease across all areas. This includes ongoing collaboration and co-design with the many rare disease organisations that represent Australians living with a rare disease. These organisations enable connection and support, lead advocacy and awareness, and encourage active consumer participation. This Action Plan presents an important opportunity to embed the rare disease consumer voice in the design, implementation and evaluation of services for Australians at all levels. This has the potential to lead to better outcomes for people living with a rare disease as well as their families and carers.

Theme #4
The need to measure rare diseases

Limited data is a common feature in rare diseases. This is heightened by poor quality, disjointed collection methods and the ineffective use of data for rare diseases. Such limitations are evident across a range of areas, from health system classification to research. Research, monitoring and ongoing evaluation are critical in rare diseases because, ultimately, if we are not counting rare diseases, people living with rare diseases do not count.

Theme #5
The need for sustainable systems and workforce

Sustainable systems and workforce are critical to the long-term success of this Action Plan. Throughout stakeholder consultations, we heard many reports of staff shortages and a lack of funding. As such, there is a real need to build on, and invest in, the existing strengths of the workforce for rare diseases. National leadership is required to coordinate stakeholders to develop and implement a workforce strategy for rare diseases.

The essential role of rare disease organisations must also be recognised and sustained. Rare disease organisations play a key role in raising disease awareness and providing much-needed person-centred information. These organisations often fill gaps in the system, not only in terms of awareness and education, but also in peer support and, increasingly, in the research and data sphere. However, these organisations are significantly under-resourced and are largely volunteer-based, posing a risk to their long-term sustainability.

Theme #6
The need for stakeholder collaboration

The success of this Action Plan is underpinned by stakeholder involvement, collaboration and engagement. It is essential that all key stakeholders in the rare disease community, including people living with a rare disease, clinicians, researchers, governments and industry work together to progress this Action Plan.

Theme #7
State, national and international partnerships as well as cross-sector collaboration

Given the small populations and complexity involved in rare diseases, strong ongoing partnerships are invaluable. Global collaboration and the sharing of knowledge and expertise are often required to ensure the best outcomes for people living with a rare disease. Due to the nature of Australia's health and social systems, state and national partnerships are vital, as is the need for the ongoing facilitation of these partnerships.

The complex nature of rare diseases requires the integration of numerous public domains that extend beyond health to disability, social/welfare, education, employment, housing and many other areas. At a national level, policy leadership is required to enable the effective and efficient delivery of integrated whole-of-life care that supports and responds to people's needs. Similarly, national policy leadership is also required to seamlessly address health and social system challenges.

Theme #8
The need to progress early implementation wherever possible

Rare diseases are often progressive and shorten life expectancy, and the burden of rare diseases remains unacceptably high[98]. As such, implementation activities must build on successful initiatives already underway to address the need for urgency and to continue to build capacity, collaboration and coordination.

Implementation Mechanisms

International Exemplars

A number of international exemplars of quality implementation mechanisms already exist. They include:

The Genetic and Rare Diseases (GARD) Information Center

GARD is a program of the National Center for Advancing Translational Sciences (NCATS) and is funded by two parts of the US's National Institutes of Health (NIH). The GARD Information Centre demonstrates international best practice in providing information and resources for[99]:

- people living with a rare disease and their family;
- health care providers;
- social workers;
- teachers who work with people living with a rare disease;
- scientists undertaking relevant research;
- community leaders who guide people towards quality resources about their rare disease;
- advocacy groups; and
- the general public.

Rare Disease Centres of Excellence

In 2013, Rare Disease United Kingdom's (RDUK) *Centres for Excellence for Rare Diseases* report, identified the following list of key characteristics to define Centres of Excellence in the UK[100]:

- **Coordinated care** – the provision of a named care coordinator for every person living with a rare disease. This care should involve coordination with local health care providers.
- **Adequate caseload** – expertise to support the sustainability of care provision and the facilitation of research.
- **Not be dependent on a single clinician** – vital to sustainability.
- **Arrangements for transition from children to adult services** – a key issue for those with childhood onset rare conditions.
- **Information hub and location for peer interaction** – engage with people from rare diseases and work with rare disease organisations.
- **Engagement with people living with a rare disease** – this includes working closely with rare disease organisations.
- **Research active** – record and share information collected either through their own registry or through participation in a national registry.
- **Education and training for medical professionals** – strong links being made between education providers and Centres of Excellence.
- **Membership of international networks of excellence** – work collaboratively to facilitate best practice.

European Reference Networks (ERNs)

ERNs have achieved global recognition in facilitating the delivery of expert care, despite geographical boundaries and distances. Virtual ERNs are comprised of health care 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 health care professionals across borders, giving people access to the expert knowledge they need even if it's not available in their own country or region[101].

Customisation for the Australian context

Similar implementation mechanisms in Australia should be led by a national peak organisation that builds capacity in the rare disease sector. RVA is the current national peak organisation advocating for people living with a rare disease. Having demonstrated strong partnerships and linkages with the rare disease sector throughout its history, including in the development of the Action Plan, RVA is well positioned to lead its implementation in line with international exemplars. RVA is also best placed to build on, and utilise, existing relationships with international rare disease networks such as EURORDIS, IRDiRC and the more newly-developed Asia Pacific Alliance for Rare Disease Organisations (APARDO).

It is critical that the national peak organisation (RVA) is resourced to raise the profile of rare diseases, both in Australia and internationally. RVA must also continue to collaborate with governments and the rare disease community to lead action for rare diseases. Through further resourcing and development, RVA, as the national peak organisation, could lead the:

- provision of an accessible multi-purpose digital repository of information and resources for rare diseases, including available care and support services. This portal would provide access to a wide range of information. Examples could include articles, research and lists of specialists for a particular rare disease. This would be targeted at people living with a rare disease, health professionals, researchers and the general public, and consolidate existing information and resources;
- continued assistance connecting stakeholders with one another, including leaders of rare disease organisations with health professionals and researchers;
- provision of a platform for rare disease stakeholders to enhance communication, collaboration and engagement within the sector;
- the collection and coordination of rare disease expertise that will inform the development of nationally consistent, evidence-based recommendations for areas such as diagnosis, clinical and social care, referral pathways, effective data collection and use;
- provision of a platform for state and national partnerships;
- pursuit of international partnerships to maximise benefits to the Australian rare disease community; and
- continued collaboration with all governments to improve services and policies that impact on people living with a rare disease.

To cover the breadth of Australia, the national peak organisation would provide facilitatory and secretariat support to:

Centres of excellence located throughout Australia that act as localised points of contact. They may be comprised of research groups or institutes, clinics, hospitals and rare disease organisations. This mechanism builds on existing strengths in the sector, increasing its sustainability in the long-term. Further investment would respond to existing critical funding gaps and build workforce capacity, ensure person-centred collaboration and co-design, while allowing for specialisation and reducing duplication.

Appendix 1

Glossary

Chronic conditions is a term used interchangeably with 'chronic diseases', 'noncommunicable diseases', and 'long term health conditions'. Chronic conditions have complex and multiple causes; may affect individuals either alone or as co-morbidities; usually have a gradual onset, although they can have sudden onset and acute stages; occur across the lifecycle, although they become more prevalent with older age; can compromise quality of life and create limitations and disability; are long-term and persistent, and often lead to a gradual deterioration of health and loss of independence. While not usually life-threatening, are the most common and leading cause of premature mortality[102].

A **disability** is defined by *The Disability Discrimination Act 1992 (Cth)* as[103]:

- total or partial loss of the person's bodily or mental functions;
- total or partial loss of a part of the body;
- the presence in the body of organisms causing disease or illness;
- the malfunction, malformation or disfigurement of a part of the person's body;
- a disorder or malfunction that results in the person learning differently from a person without the disorder or malfunction; and
- a disorder, illness or disease that affects a person's thought processes, perception of reality, emotions or judgment, or that results in disturbed behavior.

Epidemiology is the study of how often diseases occur in different groups of people and why. Epidemiological information is used to plan and evaluate strategies to prevent illness and as a guide to the management of patients in whom disease has already developed[104].

Fundamental discovery research is defined as the creation of new knowledge and/or the use of existing knowledge in a new and creative way so as to generate new concepts, methodologies and understandings. This could include synthesis and analysis of previous research to the extent that it leads to new and creative outcomes[105].

Genetics is the study of heredity[106]. As defined by the Cambridge Dictionary, it is 'the study of how, in all living things, the characteristics and qualities of parents are given to their children by their genes'[107].

Genomics is the study of genes and their functions, and related techniques, and 'addresses all genes and their inter relationships in order to identify their combined influence on the growth and development of the organism'[108].

Health technology is, according to the World Health Organization (WHO), 'the application of organized knowledge and skills in the form of devices, medicines, vaccines, procedures and systems developed to solve a health problem and improve quality of lives.'[109]

Health Technology Assessment (HTA) is a multidisciplinary field of policy analysis studying the medical, economic, social and ethical implications of the development, diffusion and use of health service delivery, and associated technologies, in a systematic, transparent, unbiased and robust manner. HTA encapsulates a range of processes and mechanisms that use scientific evidence to assess the quality, safety, efficacy, effectiveness and cost effectiveness of health services[110].

Incidence – The incidence of a disease is the rate at which new cases occur in a population during a specified period[111].

Interdisciplinary care refers to an approach to care that involves team members from different disciplines working collaboratively, with a common purpose, to set goals, make decisions and share resources and responsibilities. The person being cared for, and their family and carers, are central to discussions and decision-making. This is in contrast to a multidisciplinary approach, which is less collaborative, may not involve common objectives, and sees discipline-specific care plans implemented simultaneously but independently of one another[112].

Orphan drugs 'are so called because they are intended to treat diseases so rare that sponsors are reluctant to develop them under usual marketing conditions…[113] The indications of a drug may also be considered as 'orphan' since a substance may be used in the treatment of a frequent disease and not have been intended for another, more rare indication'[114].

Peer support 'is a system of giving and receiving help founded on key principles of respect, shared responsibility, and mutual agreement of what is helpful. Peer support is not based on psychiatric models and diagnostic criteria. It is about understanding another's situation empathically through the shared experience of emotional and psychological pain. When people find affiliation with others they feel are 'like' them, they feel a connection. This connection, or affiliation, is a deep, holistic understanding based on mutual experience where people are able to 'be' with each other without the constraints of traditional (expert/patient) relationships'[115].

Person-centred and family-centred care are terms used throughout the Action Plan to refer to care that sees an equal partnership between the person receiving care, their family (if appropriate) and health professionals to ensure that health care decisions are respectful of and responsive to the preferences, needs and values of people, and that they have the education and support they need to make decisions and participate in their own care.

Phenotype is a term that refers to 'the observable physical properties of an organism… [including its] appearance, development and behavior. Examples of phenotypes include height, wing length, and hair color. Phenotypes also include observable characteristics that can be measured in the laboratory, such as levels of hormones or blood cells'[116].

Precision medicine is an emerging approach for disease prevention and management that tailors care to account for an individual's variations in genes, environment, and lifestyle[117].

Prevalence – The prevalence of a disease is the proportion of a population with a disease at a point in time (point prevalence) or over a specified period of time (period prevalence)[118].

Primary health care – 'For most patients, a primary care clinician will be their first point of contact in the Australian health system. A primary care clinician may be a doctor, dentist, nurse, allied health professional or a pharmacist. This level of care may be provided in a general practice, community or allied health centre or Aboriginal and Community Controlled Health Services. It may also include health promotion, health education or prevention. Depending on the person's health condition, they may be referred on to secondary or tertiary care'[119].

Prevention refers to both actions aimed at avoiding the manifestation of a disease and early detection when this improves the chances for positive health outcomes[120]. Examples include:

- **Pre-conception vaccination** against chicken pox (varicella) to avoid development of the rare disease congenital varicella syndrome in infants;
- **Awareness and education** targeted at families of pregnant women on risks of passing whooping cough, a rare disease, onto newborns, and measures to reduce those risks (i.e. vaccination);
- **Folic acid fortification and supplementation** to lower the incidence of rare neural tube defects, such as spina bifida, and;
- **Prenatal** and **newborn screening programs** to enable early detection of rare diseases.

Rare diseases – according to Orphanet, the portal for rare diseases and orphan drugs, 'Rare diseases are diseases which affect a small number of people compared to the general population and specific issues are raised in relation to their rarity. In Europe, a disease is considered to be rare when it affects 1 person per 2000.'[121] While Australia does not have an explicit legislated definition of rare diseases, the Therapeutic Goods Regulations 1990 states that, in order for a medicine to be designated as an orphan drug, it must be intended to treat a condition that affects less than five in 10,000 Australians at the time of application, or to prevent or diagnose a condition that would not be likely to be supplied to more than five in 10,000 Australians each year.

Rare disease organisations is the term used throughout to refer to peak/leading, individual patient and peer support organisations, including those related to undiagnosed rare diseases. Their activities can vary from connecting people living with a rare disease for peer support to funding research.

Rare disease registries aim to promote patient-centred best practice, encourage uniformity around key principles and commit to further developing a growing understanding of the national rare diseases picture[122].

Surveillance is defined as an ongoing, systematic collection, analysis and interpretation of health-related data essential to the planning, implementation, and evaluation of public health practice[123].

Telehealth allows health care professionals to examine, diagnose, and treat patients using technology like a phone, computer, or other connected device[124].

Transition is the term used throughout the Action Plan to refer to the transition of one's care from one health service to another. This may be due to a change in age, level of needs, providers, geography or other factors. The transition from paediatric to adult health services is commonly cited as a challenge for people living with a rare disease. Examples of Australian paediatric to adult transition services, for people with rare diseases, include:

- The Transition Support Service, which operates at the Royal Children's Hospital Melbourne in Victoria[125]; and
- Two transition clinics in Western Australia at Perth Children's and Sir Charles Gairdner Hospitals[126].

Undiagnosed rare diseases is the term used throughout to refer to the two groups of undiagnosed diseases, which are classified amongst rare diseases and comprise of[127]:

- 'Not yet diagnosed' refers to a patient who lives with an undiagnosed condition that should be diagnosed but hasn't been because the patient has not been referred to the appropriate clinician due to common, misleading symptoms, or an unusual clinical presentation of a known rare condition'; and
- 'Undiagnosed ('Syndromes Without A Name' or SWAN) refers to a patient for whom a diagnostic test is not yet available since the disease has not been characterised and the cause is not yet identified.This patient can also be misdiagnosed as his/her condition can be mistaken for others.'

Undiagnosed Disease Program (UDP) – Australia's first UDP is currently underway in Western Australia, known as the UDP-WA. The UDP-WA aims to provide a definitive diagnosis for people with complex and long-standing medical conditions. The program was announced in late 2015. It incorporates a team of doctors from a broad range of specialties working together in partnership with researchers. The UDP-WA officially commenced in March 2016, when a cross-disciplinary Expert Panel met to consider its first case. This expert panel is made up of doctors from a range of medical specialties, depending on the individual case requirements. These may include genetics, general paediatrics, endocrinology, neurology, gastroenterology, dermatology, oncology and respiratory medicine[128].

Appendix 2

Acknowledgments

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Appendix 3

Working Towards Rare Disease Care and Support that is Integrated

Rare disease care and support that is integrated streamlines patient journeys through often fragmented and complex health care and social systems. It ensures optimal, continuous and effective whole-of-life care.

People with a rare disease often require large interdisciplinary teams of doctors, nurses and allied health professionals who work in different settings (primary care, hospital, emergency departments, community allied health) to manage the multiple medical problems and disabilities experienced. Their health needs can change throughout the course of their lifetime, and these changes can be rapid and critical. Additionally, they often have complex support needs that extend beyond health to incorporate disability, social/welfare, mental health, education, employment and housing. It is not just the person living with a rare disease who needs support. Their family and carers also have high and significant support needs.

Rare disease care and support should meet the needs of patients and professionals while taking into account local contextual factors, such as existing services and structures; resources, including funding and workforce; workforce expertise, and; the preferences of people living with a rare disease and their families and carers.

To embed into the health care and social support systems, and to maximise impact for all stakeholders, strong collaboration and consultation with all stakeholders is needed. Working towards a rare disease care and support model that is integrated must include:

- *Formative evaluation* – scoping local context, resources and the needs of all stakeholders;
- *Model development* – person-centred models collaboratively developed by all stakeholders, building on both the formative evaluation results and Australian and international best-practice;
- *Trials of the developed model in the real world* – to determine person and family, service provider and system outcomes, including patient reported outcomes and experiences;
- *Thorough evaluation* – to facilitate understanding of the implementation process and factors that help or hinder implementation and sustainability in different contexts;
- *Economic evaluation* – including incremental cost effectiveness ratios and incremental health utility ratios or savings per quality-adjusted life year (QALY); and
- *Development of long-term recommendations and plans for scaling-up and spreading successful models of care to other settings.*

Integrated rare disease care and support models tend to be complex and need to be flexible to meet individual needs. They are reliant on a suite of enablers, including, but not limited to:

- Care and support coordination delivered by specialist coordinators;
- Interdisciplinary clinics where services from teams of different medical, allied health and social support professions work together to meet the needs of the person with a rare disease and their families and carers;
- Interconnected clinical networks and communities of practice to support shared learning;
- Alternative care and support delivery platforms, including telehealth and mobile apps, to increase equity of access;
- Supported communication through shared care and support management plans, shared electronic records and case notes, and case conferencing; and
- Capacity building among the workforce to build knowledge, skills and confidence to look after patients with a rare disease.

RVA thanks Dr Yvonne Zurynski for her valuable input into this piece.

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Appendix 4

Health Technology Assessment

According to the WHO, 'a health technology is the application of organized knowledge and skills in the form of devices, medicines, vaccines, procedures and systems developed to solve a health problem and improve quality of lives'[129]. Diagnostic tests, blood products and public health interventions also fall within this definition.

What is Health Technology Assessment (HTA)?

HTA is a multidisciplinary field of policy analysis studying the medical, economic, social and ethical implications of the development, diffusion and use of health service delivery, and associated technologies, in a systematic, transparent, unbiased and robust manner. HTA encapsulates a range of processes and mechanisms that use scientific evidence to assess the quality, safety, efficacy, effectiveness and cost effectiveness of health services[130].

Many different types of health technology are used in rare diseases, and many more are emerging. There are a number of different HTA pathways for health technologies for rare diseases.

- HTA Access Point, which is the team responsible for assisting potential applicants and to facilitate the assessment of co-dependent or hybrid technologies to ensure their complexity does not hinder timely access to reimbursement[131].
- The Pharmaceutical Benefits Advisory Committee (PBAC) for pharmaceuticals to be funded under the Pharmaceutical Benefits Scheme (PBS) and vaccines to be funded under the National Immunisation Program[132].
- The LSDP for pharmaceuticals that the PBAC has deemed as being clinically effective but not cost-effective, but are considered life-saving for life-threatening and very rare diseases[133].
- The Medical Services Advisory Committee for medical services involving new procedures or health technologies to be funded under the Medicare Benefits Schedule or for other programs (for example, blood products or screening programs)[134].

Limited data is inherent in rare diseases. This creates uncertainties that present specific challenges for HTA processes.

Acronyms

APARDO	Asia Pacific Alliance for Rare Disease Organisations
APEC	Asia-Pacific Economic Cooperation
AIHW	Australian Institute of Health and Welfare
CALD	Culturally and linguistically diverse
ERNs	European Reference Networks
EURORDIS	European Organization for Rare Diseases
GARD	Genetic and Rare Diseases
HTA	Health Technology Assessment
ICD-11	International Classification of Diseases 11th Revision
IRDiRC	International Rare Diseases Research Consortium
LSDP	Life Savings Drug Program
MRFF	Medical Research Future Fund
NBS	Newborn Bloodspot Screening
NCAR	National Congenital Anomalies Register
NCATS	National Center for Advancing Translational Sciences
NDIS	National Disability Insurance Scheme
NIH	National Institutes of Health
OHTA	Office of Health Technology Assessment
PBS	Pharmaceutical Benefits Scheme
RDUK	Rare Disease United Kingdom
RVA	Rare Voices Australia
SMAC	Scientific & Medical Advisory Committee
SWAN	Syndromes Without A Name
TEP	Trials Enabling Program
TGA	Therapeutic Goods Administration
UDP-WA	Undiagnosed Disease Program Western Australia
UK	United Kingdom
US	United States
WARDA	Western Australian Register of Developmental Anomalies
WHO	World Health Organization

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National Strategic Action Plan for Rare Diseases

February 2020

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.

RARE METABOLIC DISEASE WORKFORCE WHITE PAPER

Towards a Strengthened Rare Disease Workforce for Australia

Current Status in New South Wales (February 2022)

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.

Executive Summary

Rare metabolic diseases include over 1,600 rare disorders, which result from defects in the breaking down and conversion of carbohydrates, proteins and fats.[1] The variability and complexity of rare metabolic diseases, together with the rarity of each condition, causes overwhelming medical and social burden on families.[2] This complexity means that patients require care from a variety of specialists. Rare metabolic conditions are a highly heterogenous group of complex, multisystemic conditions that are poorly understood due to low patient numbers.

The findings presented in the *Rare Metabolic Workforce White Paper–Towards a Strengthened Rare Disease Workforce for Australia* (White Paper)[3] gathered expertise and experience from patients, carers, healthcare professionals and industry. Despite growing demand due to improvements in diagnosis and more rare metabolic patients surviving into adulthood, the service is under resourced, and staff are struggling to keep up with current demand–this is being felt by patients.

Urgent Key Actions

- Address current gaps to ensure all patients in New South Wales (NSW) living with a rare metabolic condition have their needs met
- Increase staffing capacity to meet current needs and future demand—critical staff shortages are being felt across all roles, including diagnostic scientists
- Invest in and embed care coordination, multidisciplinary care, outreach services, metabolic specialist training, data collection processes (including registries) and telehealth into clinical care
- Revise the current funding model—significant amounts of activity across NSW metabolic services are not tallied
- Invest in resources and infrastructure to embed clinical trials and new therapies into clinical care.

It is vital for governments and key stakeholders in NSW to invest in rare metabolic services to ensure optimal care for rare metabolic patients now and into the future. Recommendations for immediate action and future investment into New South Wales' metabolic services, together with proposals for a national approach to rare disease care for Australia, are outlined in this summary.

About the Research

Australia's first nationally coordinated effort to address rare diseases—the National Strategic Action Plan for Rare Diseases (the Action Plan)[4]—called for the development of a national rare disease workforce strategy that responds to current and future demands, including the impact of genomics. The 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.

The White Paper 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 looked 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. Issues across metabolic care in NSW are 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.

Key Findings (National)

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.

These 5 key findings highlight the 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. This state summary outlines the strengths and gaps across the metabolic workforce in NSW in relation to 4 baseline 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.
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.

The Situation in New South Wales

35% of rare metabolic patients and carers surveyed and residing in NSW are not having their needs met by existing services

As the largest state by population 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 of these services also provide support to patients and diagnostic services to people living in the Australian Capital Territory. Gaps remain in service coverage, and 35% of patients surveyed in NSW reported that their needs were not being fully met.

How Does New South Wales Measure Up?

NSW	Partially meets criteria through service within own jurisdiction	Partially meets criteria through service within own jurisdiction	Partially meets criteria through service within own jurisdiction	Partially meets criteria through service within own jurisdiction

National Comparisons

	Criteria 1 Multidisciplinary Services	Criteria 2 Models of Care	Criteria 3 Emerging Therapies	Criteria 4 Training and Succession
NSW	Partially meets criteria through service within own jurisdiction	Partially meets criteria through service within own jurisdiction	Partially meets criteria through service within own jurisdiction	Partially meets criteria through service within own jurisdiction
VIC	Partially meets criteria through service within own jurisdiction	Partially meets criteria through service within own jurisdiction	Partially meets criteria through service within own jurisdiction	Does not meet criteria
QLD	Partially meets criteria through service within own jurisdiction	Fully meets criteria	Partially meets criteria through service within own jurisdiction	Does not meet criteria
SA	Partially meets criteria through service within own jurisdiction	Partially meets criteria through service within own jurisdiction	Partially meets criteria through service within own jurisdiction	Partially meets criteria through service within own jurisdiction
ACT	Partially meets criteria through formal arrangements with other jurisdiction/s	Partially meets criteria through formal arrangements with other jurisdiction/s	Partially meets criteria through formal arrangements with other jurisdiction/s	Does not meet criteria
TAS	Partially meets criteria through informal arrangements with other jurisdiction/s	Partially meets criteria through informal arrangements with other jurisdiction/s	Does not meet criteria	Does not meet criteria
NT	Does not meet criteria	Does not meet criteria	Does not meet criteria	Does not meet criteria
WA	Does not meet criteria	Does not meet criteria	Does not meet criteria	Does not meet criteria

Next Steps for New South Wales

Address Service Gaps

The main specialist centres for rare metabolic conditions in NSW provide access to multidisciplinary teams and transition services. However, this does not include integrated psychological services and there are gaps in access to allied health professionals across all services. Fewer than 50% of NSW patients surveyed had seen a dietitian in the previous 12 months, and fewer than one third had access to care coordination services, both of which are necessary for managing metabolic patients.

Access to new therapies is essential to metabolic care. While there is some capacity to run clinical trials in NSW metabolic services, there is inadequate resources and infrastructure, including limited physical space, to take on all opportunities and integrate clinical trials into care.

Westmead metabolic centres provide outreach services to regional hospitals and diagnostic services operate state-wide; however, savings for other health services, in terms of both reduced admissions and reduced length of stay for patients, are not counted by Westmead and there is ineffective cross subsidisation occurring.

Key service gaps must be addressed to ensure the best outcomes for people living with a rare metabolic disease in NSW, including:

1. Increasing staffing capacity and resources to meet current needs and cater for future growth—critical staff shortages were identified across all roles; however, some services highlighted shortages in diagnostic and clinical nurse specialists, and inadequate counselling support
2. Further investment in existing care coordination and multidisciplinary care models to ensure resourcing constraints do not undermine service delivery, and the services are able to meet the needs of a growing cohort of patients
3. Investing in resourcing and infrastructure to support access to emerging therapies and technologies–services reported a lack of staff and resources to meet clinical requirements for novel therapeutics and clinical trials
4. Investing in sustainable data collection processes, including registries, particularly given the rapid increase in known rare metabolic conditions—services reported limitations with patient databases
5. Reviewing current funding models—the Genetic and Metabolic Disorders Service at Westmead highlighted that a large proportion of their work is not tallied under the current activity-based funding model
6. Investing in equipment upgrades to ensure access to the best diagnostic equipment

7. Further investment in telehealth—telehealth is not established and underfunded at the Genetic and Metabolic Disorders Service at Westmead
8. Investing in and formalising outreach services to meet challenges of providing clinical care to a widespread geographical region with many regional and remote communities—currently, there is no formal funding or arrangements to cover care provided to regional NSW
9. Increasing funding for formalised and ongoing training pathways and succession planning to ensure services are robust and sustainable—the lack of formalised training was particularly noted for specialised nurses, allied health professionals and diagnostic specialists

A National Approach for Rare Disease Care Into the Future

The White Paper findings should be further leveraged to respond to broader rare disease workforce issues in Australia. Continued work in this area must be prioritised by all jurisdictions to respond to the Australian Government's Action Plan in which workforce is a foundation principle and priority. International models can be adapted 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 Australia will ensure that all Australian rare disease patients have access to the best available care. The Action Plan highlights the importance of state, national and international partnerships. This work can, and needs to, start now.

References

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