FAIR FOR RARE

A FAIR GO FOR AUSTRALIANS
LIVING WITH RARE DISEASE

CALL FOR A National Rare Disease Framework

6 Strategic Priorities

Effective Rare Disease Policy Transforms Patients' Lives

rare voices
AUSTRALIA

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CALL FOR A NATIONAL RARE DISEASE FRAMEWORK

Effective Rare Disease Policy Transforms Patients' Lives

Rare Voices Australia (RVA) is Australia's non-profit, national peak organisation advocating for all who live with a rare disease - a strong common voice to advocate for health policy and a healthcare system that works for those with rare disease. RVA works with patients, key peak bodies, governments, researchers, clinicians and industry to promote rare disease research, diagnosis, treatment and services.

OUR VISION: Australians with rare diseases will have extended and improved lives.

People with rare disease often have very complex needs and face unique challenges:

- A rare disease is uncommon (less than 1 in 2000 people affected).
- There are approximately 7000 different rare diseases.
- Although individually rare, they are collectively common.

Yet they have much more in common than just small patient numbers.

Rare Diseases are often:

- life threatening or chronically debilitating;
- complex, often affecting multiple body systems and requiring specialised and coordinated care that comes at considerable cost to families and the health system;
- genetic (80%) and therefore not readily preventable;
- incurable, many with no effective treatment and symptoms often worsen over time.

People, often children, living with a rare disease fight against their disease each day. Unfortunately, in many cases the health system is not equipped to respond to the needs of those living with rare diseases, being overwhelmingly geared towards supporting diseases that affect large numbers of people. This presents an opportunity for collaboration to improve the health system and health outcomes for people with rare diseases.

The burden of rare disease remains unacceptably high for patients, families, communities and the health system. Collectively rare diseases affect more Australian patients than diabetes. Similar prioritisation by the health system will not only make a difference to the lives of families affected by rare disease but will help reduce the economic cost to the health system. There is an opportunity to address the current imbalance in Australia's healthcare system and make it also **Fair for Rare**.

> "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

To ensure the most equitable, effective, efficient and coordinated approach to Rare Diseases and to place Australia in line with the EU, UK, and other parts of the world, Australia needs a **National Rare Disease Framework** that provides:

- A system that is equitable, accessible, consistent, transparent, timely and person-centred;
- Collaborative action that includes input from patients, healthcare, government, research, and industry;
- An opportunity for all Australian governments to provide bi-partisan support and work in partnership towards a national coordinated and collaborative approach.
- Most importantly, effective rare disease policy to transform patients' lives.

Without a Rare Disease Framework – Australia is falling behind many countries including the UK, France, Germany, Canada, Taiwan and more.

RVA calls for a **National Rare Disease Framework** with six strategic priorities.

1. DIAGNOSIS
2. ACCESS TO TREATMENTS
3. DATA COLLECTION
4. COORDINATED CARE
5. ACCESS TO SERVICES
6. COORDINATED RESEARCH

Long-term, a comprehensive rare disease framework is required. A staged implementation approach would ensure greater sustainability and still enable much needed immediate policy reform in critical areas. This booklet identifies the 6 Strategic Priorities of a National Rare Disease Framework.

RVA calls for all Governments to work together to develop a strategy for rare disease.

Proposal:
That the Commonwealth Department of Health establish and lead a Working Party to:

1. Map the gaps/opportunities/challenges in rare disease policy;
2. Identify the system issues in adequately addressing these;
3. Develop a road map of actions that can be taken (by both Commonwealth and State Governments); and
4. Identify an implementation timeline.

RVA membership on this Working Party would bring the vital consumer perspective as well as facilitate broad stakeholder engagement (patient groups/ researchers/ clinicians).

DIAGNOSIS

The early and accurate diagnosis of rare diseases will lead to better outcomes for patients, and long-term economic benefits to the Australian healthcare system. Early diagnosis enables the best clinical care, treatment options, access to services, support, increased reproductive confidence and participation in clinical trials (research). For the patient, diagnosis provides a medical explanation, prognosis and management plan that benefits them and their families physically, psychologically, emotionally and financially.

The value of a diagnosis is undeniable and cannot be underestimated, even in the absence of an effective treatment.[1]

A timely rare disease diagnosis means:

- Treatments can begin early, often preventing unnecessary deterioration in progressive diseases, and ensuring best possible patient outcomes;
- Easier access to health and disability services;
- Families can make informed decisions about reproduction;
- Easier access to peer support and connections with others with the same/similar diseases; for rare diseases where clinical knowledge is limited, sharing anecdotal wisdom is crucial;
- Reducing the number of medical specialist and unnecessary expensive and invasive investigations during the lengthy diagnostic odyssey provides efficient and cost-effective healthcare delivery.

Rare disease diagnosis challenges:

- Delayed or incorrect diagnosis contributes to the diagnostic odyssey. This has physical, psychological, emotional and financial costs for the patient, family and the health system due to multiple hospital admissions, tests and procedures;
- Clinicians cannot be expected to recognise all known rare genetic diseases, since many have not been fully identified and characterised;
- Where a diagnosis is not found, the undiagnosed rare disease patient group needs to be recognised as a distinct group with different needs from those with a diagnosis.

- **30% of people with a rare disease are impacted by a diagnostic delay of more than 5 years**[2]**.**
- **50% had at least 1 misdiagnosis**

RVA calls for a National Rare Disease Framework that supports:

- The endorsement of the Newborn Screening (NBS) National Framework proposal when it is presented to the Australian Health Ministers Advisory Council (AHMAC.) This is vital to ensure an effective mechanism to add to screening panels and early diagnose a greater amount of rare disease. In the absence of this policy, Australia has not added any new disease test to the NBS in the last 15 years despite major progress in diagnostics and treatment options in that time. It is important that screening and diagnostic programs and services are up to date and accessible.

- The National Genomics Health Framework to address the current inequities and gaps in the health system for rare genetic diseases. Genomic technology has an enormous potential to reduce the diagnostic odyssey commonly experienced by those with rare diseases.

- Clinical interfaces that capture phenotypic information and that are aligned to clinical flow. Phenotypic information (i.e. describing manifestations of disease) is the cornerstone of medicine, including genomic medicine. Use of such approaches can significantly increase the diagnostic accuracy of genomic tests. Australia is leading the way in this area.

- A focus on diagnosis, ensuring patients with suspected but undiagnosed rare disease are systemically identified.

- the development of an Australian Undiagnosed Rare Disease Guideline, to support clinicians in the management of the suspected undiagnosed RD patient to aid diagnosis and effective pre- and post-diagnosis management.

- Undiagnosed rare disease patients to have priority access to a specialised and expert diagnostic response, e.g. specialist testing technology (genomics), specialist diagnostic experts (Interdisciplinary Undiagnosed Diseases Programs and Centres of Excellence), genetic counsellors and peer support.

Some rare diseases may be compatible with a normal life if diagnosed early and properly managed, enabling patients to maintain an active life and employment

http://www.eurordis.org/sites/default/files/publications/why_rare_disease_research.pdf

1 Dudding-Blyth T. RACGP – A powerful team: the family physician advocating for patients with a rare disease [Internet]. RACGP.org.au. 2015 [cited 9 May 2017] Available from: www.racgp.org.au/afp/2015/september/a-powerful-team-the-family-physician-advocating-for-patients-with-a-rare-disease/

2 Molster C, Urwin D, Di Pietro L, Fookes M, Petrie D, van der Laan S et al. Survey of healthcare experiences of Australian adults living with rare diseases. Orphanet Journal of Rare Diseases. 2016:11(1)

ACCESS TO TREATMENTS

Australians need a health system that provides equitable access to treatments for rare disease. There are limited treatment options for many rare diseases and existing rare disease treatments are often available in other countries but not accessible to Australians.

- Treatments need to be assessed under criteria that are appropriate for rare disease, characterised by small patient populations, progressive disease, and limited data.
- The reimbursement pathway needs to be equitable, consistent, transparent and timely. Currently, treatments for rare diseases can only potentially be considered for the Life Saving Drugs Program once they have been rejected for funding on the PBS due to cost-effectiveness.
- Patients and their families are increasingly required to advocate for access to treatments, whilst dealing with the daily management of the disease.

Rare Disease patients should have equitable and timely access to the best available, current treatment options.

"Australians are generally waiting anywhere from 2–4 years longer for access to government-funded treatments for rare diseases than in comparable countries."[1]

RVA calls for a National Rare Disease Framework that provides:

- A formal response to the Life Saving Drugs Program Review that commenced in 2014. The rare disease community engaged strongly with this Review and are greatly concerned by the lack of transparency.
- Improved access to treatments. RVA welcomed the Life Saving Drugs Program Review and the opportunity to participate. As part of this Review, RVA and many other organisations called for the following reforms:-

  A new and separate fit-for-purpose model to be developed for the reimbursement and management of therapies for rare and very rare disease on the Pharmaceutical Benefits Scheme (PBS) – i.e. A new Section 200 – Rare and Very Rare Disease Therapies. A 'fit-for-purpose' assessment to be developed in the adoption of these therapies that recognises the key principles and issues for rare diseases. The assessment approach should include a range of workable criteria, including: the rarity of the condition, its nature and severity, the existing options for treatment, and the impact of the condition on patients in terms of life expectancy but also quality of life. A new Rare Disease Subcommittee of the PBAC to be formed with the specific responsibility of assessing submissions for this Section 200 listing, including representatives of the Pharmaceutical Benefits Advisory Committee (PBAC), clinical experts with rare disease expertise, and consumer representation from the rare disease community.
- Increased and earlier consumer involvement in the reimbursement process.

1 McKell Institute (2014) Funding Rare Disease Therapies: Ensuring equitable access to healthcare for all Australians p 29

DATA COLLECTION

The healthcare system should enable approaches to data collection and use that better meet the needs of people living with a rare disease. The greatest challenge in responding to rare disease is lack of data. Yet despite this, currently the healthcare system does not systematically code (identify) rare disease.

"If you can't measure it, you can't improve it." – PETER DRUCKER

Lack of data in rare disease has a direct negative impact on:

- Diagnosis
- Patient care
- Support
- Development of treatments
- Government's reimbursement of these treatments (access)
- Quality improvement
- Improving Outcomes

RVA calls for a National Rare Disease Framework that supports:

- the healthcare coding system to automatically identify and measure rare disease in Australia
- rare disease data collection for clinical care and translational research that improves quality of life, quality of care and survival for patients
- development of evidence-based rare disease policy based on data that quantifies the collective impact of rare diseases on healthcare, service planning, clinical guidelines and research.
- development of an integrated rare disease registry strategy for Australia

Registries are critical clinical tools and powerful cost-effective instruments to support clinical trials and translational research that improves quality of care, quality of life and survival. Registries are critical for rare diseases with low prevalence and propensity for variation in treatment and outcomes.[1] RVA supports the concept of a National Rare Disease Registry – to increase evidence-based knowledge and encourage research.

RVA is currently developing a **National Alliance of Rare Disease Registries** to encourage collaboration, shared knowledge and best practice. The National Alliance of Rare Disease Registries will:

- bring together existing rare disease registries across Australia
- establish national rare disease registry standards to support interoperability and cohesion across registries
- develop strategies for sustainability to maximise the utility of existing rare disease registries and support the development of new registries
- develop standards and systems for data collection that are appropriate and sustainable for healthcare personnel

Government support is important to unlock the full potential of this for the health system and the rare disease patient community it serves.

One of the greatest challenges in rare disease is lack of data.

1 Jaffe A, Zurynski Y, Beville L, Elliott E. 'Call for a national plan for rare diseases.' Journal of paediatrics and child health, (2010), 46:2-4

COORDINATED CARE

Providing a coordinated approach to care will contribute to an equitable Australian healthcare system and better health outcomes for people living with rare diseases. These services should provide continuity of care across service providers, and deliver whole-of-life services specific to the individual patient. This type of approach is already a feature of the National Disability Insurance Scheme and the Australian Digital Health agency.

Effectively coordinated care provides personalised patient care and contributes to a better quality of life; allows service providers to provide effective services; assists in avoiding unnecessary tests, referrals and hospital admissions; improves the quality and consistency of healthcare provision; results in financial savings; and a more efficient health system[1].

- It is important that care for each rare disease patient is coordinated across a range of health settings as they often have complex and long-term care needs, requiring care and input from a range of specialities[1];
- Coordination of care will support earlier diagnosis, provide access to appropriate interdisciplinary care and enhance the potential for research in these rare and neglected disorders[2];
- Coordinated care is essential to ensure holistic service delivery across the lifespan of people living with rare diseases[1].
- Centres of Excellence are important in care provision as well as increasing rare disease knowledge and expertise.

Current areas for improvement:

- Patients and families have multiple appointments, with a range of specialties and services that are not provided in a fully coordinated manner[3];
- Many services in Australia do not provide adequate transition support for patients and families going from paediatric to adult health services[3];
- Fragmented health systems lead to patients being unable to access adequate ongoing care and disease management, which is crucial for people with rare diseases even where no treatment is available[4].

RVA calls for a national rare disease framework that enables

- Development of rare disease guidelines that establish standards for care coordination and clear referral pathways;
- Centres of clinical and research expertise (including undiagnosed rare disease clinics) to:
  - deliver interdisciplinary services (including mental health) from pre-diagnosis, diagnosis, ongoing care;
  - collaborate with and support the patient's community service and primary care providers;
  - connect patients with relevant patient support organisations;
  - deliver medical training (pre-and post-graduate) in respective fields of expertise;
  - be involved in translational research, data collection, rare disease coding and rare disease registries;
  - provide virtual clinics (telehealth/telemedicine) for rural and regional patients;
  - collaborate with their respective counterparts internationally.
- Improved transitional services, such as child to adult services, change in patient location, and change in service requirements, such as end of life care.
- Promotion and increased uptake of the My Care Health Record

Delivery of coordinated care for patients with rare diseases has significant potential to save costs and improve patient outcomes[5]

1 Department of Health Western Australia. (2015). WA rare diseases strategic framework 2015-2018. Retrieved from http://ww2.health.wa.gov.au/~/media/Files/Corporate/Reports%20and%20publications/PDF/Rare-diseases-strategic-framework.ashx

2 Bushby, K. (2014). Why coordinated care is key for rare disease patients. Health Service Journal. Retrieved from https://www.hsj.co.uk/sectors/commissioning/why-coordinated-care-is-key-for-rare-disease-patients/5067326.article

3 Simpson, A. (2016). The hidden costs of rare diseases: A feasibility study. Retrieved from https://www.geneticalliance.org.uk/media/2502/hidden-costs-full-report_21916-v2-1.pdf

4 International Federation of Pharmaceutical Manufacturers and Associations. (2017). Leaving no one behind: A set of policy principles to meet the global challenge of rare diseases. Retrieved from https://www.ifpma.org/wp-content/uploads/2017/02/IFPMA_Rare_Diseases_Policy_Principles_28Feb2017_FINAL.pdf

5 Australian Bureau of Statistics. (2016). Patient experiences in Australia: Summary of findinfs, 2015-2016. Retrieved from http://www.abs.gov.au/ausstats/abs@.nsf/0/398E27DFBF6DE8E2CA257952001C9AD9?Opendocument

ACCESS TO SERVICES

The Australian health system should provide equitable access to high quality services for all people living with a rare disease, irrespective of the rarity of their condition, a lack of diagnosis, and their location. Services also should be economically accessible to all.

- People living with rare diseases are likely to require complex case management and access to a range of government and non-government services and programs. This includes allied and mental health professionals, clinical specialists, general practitioners, and disability and social support services.
- Equitable access to services reduces health inequities and positively impacts on quality of life and health outcomes for people with rare disease and their families.

The Australian Government recognises the need for equitable access to services and has a framework that provides national direction for child and youth health[1]; specifically Strategic Priority Four: children and young people have equitable access to health care services and equitable health outcomes.

Improved access to services for people with rare and undiagnosed rare diseases also aligns with the Department of Health Strategic Intent 2016-20 that lists as a strategic priority: "...improved access to high quality, comprehensive and coordinated preventive, primary and mental health care for all Australians, with a focus on those with complex health care needs and those living in regional, rural and remote areas, including through access to a skilled health workforce."

Yet challenges persist:

- Rare disease patients may be unaware of the services available.
- Rare disease specialists and services are also rare and, as patients predominantly have complex health care needs, finding appropriate specialists and services can be a challenge.
- Not having a diagnosis is a barrier to accessing services.
- Services that could benefit rare disease patients are only available (or more easily available) for specific conditions, regardless of level of unmet need. Linking service eligibility to a finite list of diseases is problematic for those with a rare disease.
- There is disparity in access to services. There is greater access to services for those who can afford to pay for their care or those able to travel to receive expert advice.

RVA calls for a national rare disease framework that:

- Ensures rare disease is better responded to in all currently funded services and programs, including:
  - Chronic Health
  - Early Intervention
  - Disability and the NDIS
  - Mental Health
- Increases consultation with the rare disease community to identify service gaps and how to better provide equitable service access.
- Enables service providers to understand the specific challenges faced by rare disease patients.
- Provides appropriate services to rare disease patients; including genetic testing services.

One family's story – different experience of rare and common disease:

The family consists of Dad, Mum and four young kids. Two of their children each have a different serious, life-threatening disease. One child has a rare neurological metabolic condition that causes serious & progressive disability. It is fatal with no effective treatment options. The family have had to battle hard for vital services, equipment, funding and respite. Recently one of their younger children was diagnosed with a different (common) life-threatening disease. The family found it a lot easier to access services, equipment, funding for their younger child with the more common disease than for their older child with the rare disease – despite the older child needing more support. They found there were more services available plus they were easier to access. There was also a treatment option for the younger child.

RESEARCH

Australia should establish a nationally coordinated program of research on rare diseases that includes active participation by patients, carers and patient advocacy groups. Ideally research should be embedded into clinic care.

For many rare diseases, there are no active research programs, and no policy incentives to support research into rare conditions. It is very difficult to get grant funding for rare diseases and requires national and international collaboration. Often it falls to patients and their families to join together and fundraise to progress rare disease research.

Rare disease research can increase awareness, improve accuracy and speed of diagnosis, advance treatments and improve the quality of life for individuals with rare diseases. In many cases, research on rare diseases can also help better understand the mechanisms of related common conditions[1].

Patients, families and patient advocacy groups can contribute to research with their disease knowledge and experience.

Challenges of rare disease research:

- As there are fewer patients who have the condition than for common diseases, it can be harder to coordinate statistically robust studies[2];
- Research activities are less common[3] and limits the development of treatments;
- Patients and scientific experts may be widely dispersed;
- Lack of data from natural history studies, which follow a group of people with a specific medical condition over time[4], impacts on their ability to inform trial design.
- Rare diseases are not identified as a National Health and Medical Research Council (NHMRC) National Health Priority Area
- Researchers choose to study more common diseases due to funding opportunities.

RVA calls for improved research efforts and coordination in the field of rare diseases. This would include:

- Greater access and support for Australian patients to participate in both Australian and international clinical trials.
- Prioritised rare disease research through the Medical Research Future Fund (MRFF) and the NHMRC Strategic Plan;
- Active participation by patients, carers and patient advocacy to provide advice on the needs of the rare disease patient community.
- Australian research to contribute to and develop international patient registries and biospecimen repositories; enrolment in studies and trials, monitoring, evaluation and sharing of data;
- Policy and action that recognises the importance of rare disease research including consideration of economic incentives for researchers and pharmaceutical development.

For many rare disease patients, participating in a clinical trial may be their only way to access (any) treatment.

1 https://www.b2match.eu/system/h2020oslo2016/files/AO_presentation_Oslo_May_13__2016_Rare_diseases.pdf?1465394365
2 https://blogs.biomedcentral.com/on-biology/2016/02/26/rare-disease-research-helps-understand-medicine-diseases/
3 Forman et al 2012
4 https://www.nih.gov/news-events/news-releases/nih-funds-research-consortia-study-more-200-rare-diseases

Australia needs a health system that is 'Fair for Rare', guided by a national rare disease plan or framework. The Communique to progress a national plan for rare diseases was reviewed at the 2014 Rare Disease Summit and has been endorsed by the following organisations:

A. Menarini Pty Ltd
aHUS Patient Support Group Australia
Alexion Pharmaceuticals Australasia Pty Ltd
Alpha-1 Association of Australia
AMDF
Amicus Therapeutics
Angelman Syndrome Association Australia
Angelman Syndrome Association of Western Australia
aPSGA
APSU
ARCAN
ausEE Inc
Austin Health
Australia Kabuki Syndrome Association Inc
Australian Addison's Disease Association
Australian College of Children & Young People's Nurses
Australian Cystinosis Support Group
Australian Pituitary Foundation
Australian Pompe Association
Autoimmune Resource and Research Centre
AWCH
Batten Disease Support and Research Association (BDSRA)
Baxalta
BioMarin
Brain Tumour Alliance Australia Inc
Cairns Ehlers-Danlos Support Group
Carers NSW
Carers Qld
CDH Australia
Charcot-Marie-Tooth Association Australia
Charles Bonnet Syndrome Foundation
Children's Tumour Foundation of Australia
Community Neurological Nurses Network Inc
Cowden's Syndrome -Australia
Cystic Fibrosis ACT
Cystic Fibrosis Australia
Cystic Fibrosis NSW
Cystic Fibrosis Tasmania
Dept. of Medical Genetics, University of Sydney
Developmental Disability WA
Duchenne Foundation
Dystrophic Epidermolysis Bullosa Research Association Australia
ECD Global Alliance
Fabry Australia
Fibromuscular Dysplasia Association of Australasia Inc
FOD (Fatty Oxidation Disorders) Family Support Group
Foundation for Angelman Syndrome Therapeutics Australia Pty Ltd
Fragile X Association of Australia Inc
Friedreich Ataxia Network
Gaucher Association Australia
Gelastic Seizure Support Hub
Genetic and Rare Disease Network
Genetic Support Network of Victoria
Genzyme
HAE Australasia
Haemochromatosis Australia
HCU Network Australia
HHE Hemiconvulsion Hemiplegia Epilepsy Syndrome - Rare Connect
HSP Research Foundation Inc
Human Genetics Society of Australasia
Hypersomnolence Australia
Immune Deficiencies Foundation Australia
Indian Organization for Rare Diseases
Institute of Molecular Biology, UQ
Institute for Immunology and Infectious Diseases, Murdoch University
Janssen
Kalparrin
Lung Foundation Australia
Malaysian Rare Diseases Society
MDDA
MdDS Australia
MND Australia
MPS & Related Diseases Society Aust. Ltd
MS Australia
Multiple Sclerosis Limited
Muscular Dystrophy Association of WA
Muscular Dystrophy Foundation
Muscular Dystrophy Queensland
Muscular Dystrophy SA
Myasthenia Gravis Association of Queensland Inc
NBIA Families in Australia
Orpharma
Pain Australia
Pallister-Hall Syndrome (PHS) Support Hub
Parenteral Nutrition Down Under (PNDU)
Pfizer Australia
PKU Association of NSW Inc
PNH Support Association of Australia
Prader-Willi Syndrome Association Australia
Raynaud's Phenomenon/Disease/Syndrome Research Australia
Rett Syndrome Association of Australia
Sanfilippo Children's Foundation
Sanofi-Aventis Philippines Inc.
Self Help Queensland
Senses Australia
Shine for Thomas Foundation Inc.
Syndromes Without A Name (SWAN) Australia
Tarlov Cyst Society of Australia
Telethon Kids Institute
The Australian Phenomics Network
The Centre for Personalised Immunology
The Neurological Council of WA
The Scarlett May Foundation
The Sydney Children's Hospital Network
Trapeze
Tuberous Sclerosis Australia
UNSW
VCFS 22q11 Foundation Inx
WA Register of Developmental Anomalies
WMozzies

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