

## RARE VOICES AUSTRALIA

### KEY FINDINGS FROM THE 2014 RVA NATIONAL ROADSHOW ON RARE DISEASES: PRINCIPLES AND OBJECTIVES TO PROGRESS A NATIONAL PLAN FOR RARE DISEASES

#### Background

- Rare Voices Australia (RVA) undertook a national Roadshow in 2014 to progress a national plan for rare diseases.
- The Roadshow involved round table discussions with representatives from patient, healthcare, research, government and industry organisations with sessions in Queensland, Victoria, South Australia, New South Wales and Western Australia. A list of Roadshow participants and background information is in Attachment A.
- The discussions focussed on the need, barriers and drivers for coordinated action on rare diseases and for a national plan.
- Key findings on the principles and objectives to progress a national plan have been synthesized from the state-based discussion records. The findings will be presented at a National Summit on Rare Diseases in March 2015 for consideration and action.
- State-based discussion records from the Roadshow and further details on the National Plan and Summit are at [www.rarevoices.org.au](http://www.rarevoices.org.au).
- Peoples' willingness to share their knowledge, experience and information through the round table discussions is gratefully acknowledged by RVA.

#### Five principles and six objectives to progress a national plan for rare diseases

1. **Rare diseases need to be recognised as a national health priority.** The burden of rare diseases, while largely hidden due to inadequate information systems, is unacceptably high for patients, families and the community.<sup>1</sup> People living with a rare disease have shared concerns and needs that warrant national recognition, leadership and action.
2. **More equitable and timely access to diagnostics, treatments, services and coordinated care for people living with a rare disease.** It is in the national and state interest to establish leadership, direction and action towards more effective national systems and approaches to rare diseases. Appropriate service models should be identified and harmonised across states and territories and developed for regional and remote areas.
3. **Incentives are required to drive a coordinated and collaborative action on rare diseases. A national approach should be established, backed by the Australian government.** There is no shortage of opportunities to improve diagnostics, treatments, services and coordinated care. A challenge is to reach agreement on how best to focus effort. Incentives should bridge efforts at local, state, national and international scales, and support integrative approaches which leverage knowledge, skills and capacity.

<sup>1</sup> An estimated 1.2 -2 million Australians live with a rare disease.

4. **Leaders from patient, healthcare, research, and government and industry organisations need to work collaboratively to champion a National Plan.** The Plan needs endorsement from of the Australian, state and territory governments. A National Plan requires agreement among the key stakeholders.
5. **The National Plan should identify a set of objectives and enabling strategies to drive action.** Six objectives are proposed under key themes.
  - i. **National coordinated and collaborative approach**  
Establish a national platform for dialogue, information exchange, and coordinated and collaborative action on rare diseases.
  - ii. **Data collection and use**  
Identify and enable approaches to data collection and use that better meet the needs of people living with a rare disease.
  - iii. **Coordinated care**  
Identify and enable approaches to coordinated care that better meet the needs of people living with a rare disease.
  - iv. **Equitable access to services**  
Ensure Australia’s health system enables timely, equitable access to appropriate services for people living with a rare disease.
  - v. **Equitable access to diagnostics and treatments**  
Ensure Australia’s health system enables the development of diagnostics and treatments for rare diseases and facilitates timely, equitable access for patients.
  - vi. **Nationally coordinated research**  
Establish a nationally coordinated program of research on rare diseases that is informed by patients and carers.

### Key messages supporting the proposed objectives for a national plan

Key messages raised in the round table discussions are presented for each objective. Potential enabling strategies or initiatives are in italics.

1. **National coordinated and collaborative approach**  
Establish a national platform for dialogue, information exchange, and coordinated and collaborative action on rare diseases.
2. **Data collection and use**  
Identify and enable approaches to data collection and use that better meet the needs of people living with a rare disease.
  - a. Rare diseases collectively are as common as diabetes. A barrier to rare diseases being recognised as a national priority is lack of data on rare disease prevalence, and the social and economic costs and impacts of living with a rare disease for people and their families. A dilemma is that health departments require data to determine priorities for action and there is limited data on rare diseases.

- i. In costing rare diseases, we need to cost the delay in diagnosis, unnecessary treatments and operations associated with misdiagnosis, the impacts on siblings, the opportunity cost when time is spent on the child with a rare disease, and the impact of rare diseases on marriage breakdown and being a single parent.
  - b. Rare diseases require recognition as a disease category alongside other common disease categories in health and research information systems. Lack of recognition of rare diseases as a disease category limits capacity to develop improved services, diagnostics and treatments.
  - c. Varying estimates have been published on the number of people with a rare disease which is confusing. The data needs to be tightened and a national definition agreed. <sup>2</sup>
  - d. People living with a rare disease have limited capacity to contribute genotypic and phenotypic and other data on their rare disease. Most people are not on treatments, do not access hospital services and are therefore not 'plugged into registries'. How can we expand participation in data collection to support the development of diagnostics, treatments, early interventions and services?
  - e. Australia has multiple rare disease data registries with duplicated effort across the different registries. Data registries need to be harmonised nationally and internationally. More efficient systems and tools are needed to collect, share and analyse patient data, and to manage data entry, access, consent and privacy.
  - f. International experience is that patient groups are a key driver to overcoming the barriers to establishing rare disease registries.
  - g. *A feasibility study could assess the merits of a national rare disease registry, and or a system of joined up registries and information systems. Many questions need answers to progress with reference to scope, costs, benefits, access, usability, quality control, recruitment, consent, privacy and long term resourcing.*
  - h. *One or more initiatives could be established to demonstrate innovative approaches to data registry development and use that address rare disease patient needs.* <sup>3</sup>
- 3. Coordinated care**  
Identify and enable approaches to coordinated care that better meet the needs of people living with a rare disease.
- a. It is not uncommon for 10 or more specialists to be involved in patient care.

<sup>2</sup> It is estimated that only 3.5% of rare disease are coded using the International Classification of Disease (ICD) codes. The other rare diseases are listed as 'other' and therefore are invisible in the health system (nationally and internationally). Orphan codes will be incorporated into ICD codes in 2017 or later using a code for all rare diseases.

<sup>3</sup> For example a project could be initiated to enable rare disease patient groups with very small numbers to benefit from the systems and tools for data collection that have been developed by the large patient organisations or international organisations.

- b. While transdisciplinary clinics and coordinated and integrated approaches to care exist for some rare diseases where a treatment option is available, these services are required to meet the complex needs of all people with a rare disease.
- c. Patients want care closer to home by people who are informed about their rare disease.
- d. Patients require support to transition from paediatric to adult services. The lack of coordination among specialists during the switching phase can result in major negative consequences for young people.
- e. It is recognised that there are pressures on health services to move away from coordinated care models because of scale, critical mass and resourcing challenges. People living with a rare disease are generally disadvantaged by the absence of coordinated care.
- f. General practitioners should be involved in developing coordinated care models, approaches and tools. They are the first point of contact and are more likely to be involved in life-long patient management and care.
- g. *There is significant potential to apply successful coordinated care models, approaches and tools across different types of rare diseases, across paediatric and adult health services, and across health conditions (for example by applying what has been learned from the development of services for cancer patients).*
- h. *There is significant potential to extend coordinated care beyond the medical model to include for example how young people living with a rare disease interact with health, education, training, disability and other services. This could be progressed through a pilot initiative as part of the National Disability Insurance Scheme.*

#### 4. Equitable access to services

Ensure Australia's health system enables timely, equitable access to appropriate services for people living with a rare disease.

- a. People living with a rare disease seek equitable access to quality health services which narrow the gap between how they experience the health care system compared to people who have more common, complex health conditions and irrespective of where people live.
- b. Patients desire a nationally coordinated approach to genetic testing and counselling that is more equitable, efficient and effective than the current system.
  - i. Social and economic costs associated with delays in testing can be huge.
  - ii. The cost of and access to genetic testing varies depending on where people live.
  - iii. There is currently no national framework for genetic testing.

- iv. Expansion of neonatal and familial testing should be supported where this can be shown to be cost beneficial.
  - v. There is a large unmet demand for genetic counsellors, clinical geneticists and laboratory scientists.
  - vi. *Expand and strengthen state-based clinical genetic services and develop “one stop” rare disease clinics.*
- c. Patient and carer identified aims are to improve consistency of care and consistency in cost.
  - d. *Develop national guidelines for professional practice and service delivery targeted to rare diseases.*
  - e. *Develop national guidelines to assist rare disease patient organisations to establish and develop their governance structures, leadership skills and organisation strategies.*
- 5. Equitable access to diagnostics and treatments**
- Ensure Australia’s health system enables the development of diagnostics and treatments for rare diseases and facilitates timely, equitable access for patients.
- a. The ‘diagnostic odyssey’ is a principal concern for people living with a rare disease, many of whom experience delays in diagnosis of years or decades. Meanwhile people and their families experience the profound social, health and economic burden of misdiagnoses, and inappropriate treatments, care and disease prevention. Delays in diagnosis can lead to shortened life expectancy.
  - b. The advent of molecular testing for multiple diseases is a major breakthrough that will reduce the time to diagnosis and enable screening with profound benefits for individuals, families and communities.
  - c. People living with a rare disease seek reforms to government systems to enable timely and affordable access to new treatments and to overcome barriers to testing and approvals due to small patient numbers in Australia and globally.

**6. Nationally coordinated research**

Establish a nationally coordinated program of research that is informed by patients and carers.

- a. Timely access to overseas research on diagnostics and therapeutics is particularly important where there are small patient numbers and limited services in Australia.
- b. Patients express a desire for research to extend beyond diagnostics and therapeutics (which is an ongoing priority) to research that informs improved, joined up services which better support people living with a rare disease.
- c. It is in the public interest to understand how different health care models, approaches, tools and technologies impact on people living with a rare disease, the associated costs and benefits, and how the options that work best for people living with a rare disease can be further developed and applied across Australia.

- d. There are models for research that bring patients, carers, clinicians and researchers together that could be replicated in Australia such as the US Patient-Centered Outcomes Research Institute and the UK James Lind Alliance.

**Key messages on the barriers to a national plan for rare diseases and how they might be overcome**

1. Lack of data on the social and economic costs and impacts of living with a rare disease for people and their families.
  - a. A dilemma is that health departments require data to determine priorities for action and investment and there is limited data on rare diseases.
  - b. A feasible national system or road map for data collection and use needs to be articulated and promoted for rare diseases.
2. Lack of levers to progress a National Plan
  - a. Highlight evidence of inequity in service access and disadvantage for people living with a rare disease.
  - b. Provide examples of how national plans for rare diseases in other countries have led to health system reforms and benefitted people living with a rare disease.
  - c. Identify and promote approaches to health care that are benefitting people living with a rare disease and how these approaches could be more broadly applied.
3. Lack of champions working together to progress a National Plan
  - a. A leading health bureaucrat is required to champion a National Plan from each of the states and territories and nationally. This is not necessarily the heads of Department in the hospitals but other people in the system. They need to be open and transparent.
  - b. At least one influential State health Minister is required, in addition to the national political support that is being garnered.
  - c. Patient organisations could establish a coordinated advocacy campaign to write a joint letter to health ministers and the Chair of the Australian Health Ministers Advisory Council that communicates their support for a National Plan. The letter should indicate the size of membership for each patient organisation signatory.
  - d. Seek advice from advocacy experts in the cancer community on how to strategize a National Plan.
4. Clear objectives, enabling strategies and a handful of feasible demonstration initiatives should be articulated.
  - a. It is important to propose solutions and not problems to government, for example solutions based on models of care. Look to build on what has been done already.



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A U S T R A L I A

- b. Bring some big ideas to the fore using a staged approach. Highlight the diagnostic odyssey, the burden of the disease on the community and the need to solve access and equity issues. Focus on the patient journey and access to diagnostic testing, coordinated clinical services and life-long management of patients.
- c. Identify 4-6 national projects or initiatives for investment which would enable of the order of 50 organisations to come together to achieve shared outcomes. Investment could be from government, private and or benevolent sources.
- d. Clarify the scope and feasibility of a National Registry for Rare Diseases, and alongside other pathways to address patient organisation concerns about data collection and use.



**ATTACHMENT A: LIST OF ROADSHOW PARTICIPANTS AND  
A COPY OF BACKGROUND INFORMATION  
PROVIDED TO THE PARTICIPANTS**

**List of Roadshow participants**

**New South Wales**

Association for the Wellbeing of Children in Healthcare (AWCH)
Australasian Tuberous Sclerosis Society
Australian Leukodystrophy Support Group (ALDS)
Australian Pompe Association
BioMarin
Cancer Council in ACT
Children's Tumour Foundation of Australia
ConnecTed Foundation
Cystic Fibrosis
Duchenne Foundation
Fragile X Association
HAE Australasia Ltd
IDFA - Immune Deficiencies Foundation Australia
LAM Australasia Research Alliance (LARA)
LeapFrog Ability
Lymphoma Australia
NSW Office for Health and Medical Research
Research Australia
Russell Silver Syndrome
Sanfilippo Children's Foundation
SCHN - Sydney Children's Hospital Network
The Abinism Fellowship of Australia
The Australian Mitochondrial Foundation

**Queensland**

The University of Queensland
The Department of Health, Qld
The Human Genetics Society of Australasia, Qld
Institute for Molecular Bioscience, The University of Queensland
Mater Health Services
Nephrologist – Royal Brisbane Hospital
Institute for Molecular Bioscience, The University of QLD



Australian Rare Chromosome Awareness Group (ARCAN)
Friends of RD Day (Far North Qld)
Cystic Fibrosis Group, Qld
Myasthenia Gravis Association of Qld Inc
Australian Pituitary Foundation Ltd
ozED - Australian Ectodermal Dysplasia Support Group Inc
Congenital Adrenal Hyperplasia Support Group Australia

### South Australia

Epilepsy – Let’s Talk About It (FIRES Group)
Fibromuscular Dysplasia Association of Australasia
Australian Leukodystrophy Support Group
Ehlers Danlos Syndrome
Australian Kabuki Syndrome Association
Sturge Weber Foundation
Diagnostic Laboratory, Department of Health, SA
Novita Children’s Services
Client Services Manager, Muscular Dystrophy SA
Juvenile Batten Disease (BDSRA)

### Victoria

Murdoch Children's Research Institute
GSK
Monash University
Murdoch Children's Research Institute
PWSA - Prader Willi Syndrome Association
University of Melbourne
Angelman Syndrome Organisation
Alpha 1 Association (AAA)
Thalassaemia Australia
PWSA Victoria (Prader Willi Syndrome Association)
Genetic Support Network of Victoria (GSNV)
aHUS (Atypical hemolytic uremic syndrome) Patient Support Group Australia
Fabry Support Group Australia
IDFA – Immune Deficiencies Foundation Australia
GSNV and AusDoCC (Australian Disorders of the Corpus Callosum)
MDS Group

Calvary Health Care Bethlehem
MPS Australia
Alpha 1 Association (AAA)
PNH Support Association Australia.
Syndromes without a Name (SWAN) Australia Inc
Porphyria Association.
Cystic Fibrosis Tasmania
ozED- Australian Ectodermal Dysplasia Support Group

### Western Australia

Living With Hypersomnia
OPHG, Dept of Health WA.
GaRDN
CRC for Spatial Information
Centre for Comparative Genomics, WA
Immune Deficiencies Foundation Australia (IDFA)
Australian Leukodystrophy Support Group
Lyme Disease Association of Australia, WA
Cohen Syndrome
Duchenne Foundation, WA
Australian Pompe Association, WA
NCWA (Neurology Council, WA) and LARA
Haemophilia Foundation, WA
Centre for Comparative Genomics, WA
Myositis Association Australia Inc., WA
Rare Voices Australia
Murdoch University

## Background information provided to Roadshow participants

### Project - 'Rare Disease National Plan Engagement Roadshow February - October 2014'

#### Project Background

Rare Voices Australia is advocating as its number one priority for Australia to adopt a National Rare Diseases Plan. The Office of Population Health Genomics (OPHG) has led a collaborative approach to consider the current rare diseases landscape relevant to Australia. Over the past two years extensive research and information has been gathered through national coordinating committees, international meetings, engagement with those from across the rare disease sector and detailed reviews of literature. This information was used to develop the Scoping paper on the need for a National Rare Diseases Plan (Scoping Paper) as requested by the Australian Health Ministers Advisory Council (AHMAC) in 2011. This paper included one recommendation, that Australia develops a National Rare Diseases Plan.

In September, the Scoping Paper was presented to AHMAC for consideration. The outcome of the discussion was that while AHMAC did not support the development of a National Rare Diseases Plan it requested more information on the status of genetic testing, disease coding and clinical pathways.

WA Health and OPHG remain committed to advancing the understanding of rare diseases and supporting those living with a rare disease. This will be achieved through ongoing national and international networks, continued development and support of registries, considering the data needs at the state, national and international level and developing a WA Rare Diseases Strategy.

The Scoping Paper is available here online.

#### RVA's Position

RVA would like to engage with the jurisdictions to gain further understanding on what is needed in order to give full support to a National Rare Diseases Plan.

#### Focus questions to guide round table and/or focus group discussion:

1. How important is a National Plan?
  - What are the most important reasons to have a Plan?
  - What will success look like in five years time?
2. Have we got the vision for the Plan right?
  - How could it be strengthened, and to make it pragmatic and achievable?
3. What are the barriers to achieving a National Plan and how might they be overcome?
4. In the absence of a National Plan, what initiatives in the Plan should and could be progressed in the coming 12-18 months?
5. What would be the reasonable next steps to progress these initiatives?

6. What can participants in this meeting commit to, over the coming 12-18 months (i) to progress a National Plan and (ii) to progress key initiatives in the Plan?
  7. What are your expectations for RVA's role in progressing a National Plan and key initiatives within the Plan?
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