



Government of **Western Australia**
Department of **Health**

WA Rare Diseases Strategic Framework 2015–2018



© Department of Health, State of Western Australia (2015)

Copyright to this material produced by the Western Australian Department of Health belongs to the State of Western Australia, under the provisions of the *Copyright Act 1968 (C'wth Australia)*. Apart from any fair dealing for personal, academic, research or non-commercial use, no part may be reproduced without written permission of the Office of Population Health Genomics, Western Australian Department of Health. The Department of Health is under no obligation to grant this permission. Please acknowledge the Western Australian Department of Health when reproducing or quoting material from this source.

Suggested citation

Department of Health, Western Australia. WA Rare Diseases Strategic Framework 2015–2018. Perth: Office of Population Health Genomics, Public Health Division, Department of Health, Western Australia; 2015.

Important disclaimer

All information and content in this Material is provided in good faith by the Western Australian Department of Health and is based on sources believed to be reliable and accurate at the time of development. The State of Western Australia, the Western Australian Department of Health and their respective officers, employees and agents, do not accept legal liability or responsibility for the Material, or any consequence arising from its use.

Dedication

In memory of Conor Murphy and Kyle Scolari, two incredible young men who lived with muscular dystrophy.

Acknowledgements

The WA Rare Diseases Advisory Group was established to inform the development of this strategic framework. The group included representatives drawn from clinicians, researchers, health service planners, policy-makers, people living with rare diseases, carers, family members and community services organisations. The members of the advisory group are listed below. The Health Consumers Council of WA is also thanked for its valuable input and endorsement of the strategic framework.

Alan Kuipers-Chan	Princess Margaret Hospital
Barry Lewis	Princess Margaret Hospital
Beth Martino	Princess Margaret Hospital
Brad Jongeling	Joondalup Child Development Centre
Catherine Stoddart	Nursing and Midwifery Office
David Henley	Sir Charles Gairdner Hospital
David Joske	Sir Charles Gairdner Hospital
Dianne Bianchini	Chief Health Professions Office
Donna Turner	Carers Association of WA
Eamon McGrath	Complex Needs Care Team, Royal Perth Hospital
Fiona Payne	Disability Health Network
Gareth Baynam	Genetics Services of Western Australia
Hooi Ee	Sir Charles Gairdner Hospital
Jacquie Garton-Smith	Cardiovascular Health Network
Jennifer Mace	Princess Margaret Hospital
Jim Codde	South Metropolitan Health Service
Kate Bullow	Complex Needs Care Team, Royal Perth Hospital
Kate Gatti	WA Child and Youth Health Network
Kathy McCoy	Neurological Council of WA
Lesley Murphy	Rare Voices Australia
Maina Kava	Princess Margaret Hospital
Melissa Evans	Senses Australia
Merrilee Needham	Fremantle Hospital
Mitch Messer	Genetic and Rare Diseases Network
Nick Gottardo	Princess Margaret Hospital
Phillipa Lamont	Royal Perth Hospital
Robyn Timms	Fremantle Hospital
Shanti Balasubramaniam	Princess Margaret Hospital
Sharon Van Der Laan	Genetic and Rare Diseases Network
Stephanie Dowden	Princess Margaret Hospital
Sue Peter	Princess Margaret Hospital
Susanna Wills-Johnson	Consumer representative

Foreword

It is with great pleasure that I present to you the *WA Rare Diseases Strategic Framework 2015–2018*. As the first strategy for rare diseases in Australia, it is a significant landmark for WA Health and for Western Australians living with a rare disease, their carers, families and the clinicians, researchers and policy-makers who support them.

I hope this framework will give you an insight into the broad scope of work being undertaken by WA Health to contribute to the best possible health and wellbeing of Western Australians living with a rare disease. More importantly, I hope it will show you that every effort is being made to ensure people living with a rare disease receive timely, accurate diagnosis and appropriate, coordinated and integrated care. Initiatives such as the creation of local-level patient registries should enhance patients' access to clinical trials, and therefore new treatments. Data collected on their healthcare experiences and service use will help better inform policy-makers, service providers and researchers, enabling them to further plan and refine services to ensure they best meet the needs of the rare diseases community.



Under this framework, Western Australians living with a rare disease – along with their carers and families – are at the centre of health care delivery and decision-making and are set to benefit from the provision of information and connections to networks of people and organisations that can support them and help them to access appropriate services. This framework acknowledges the importance of family members and carers in the provision of rare disease care and recognises the unique knowledge, experience and perspective they can bring to the development of services and policies.

Health professionals will similarly benefit from the *WA Rare Diseases Strategic Framework 2015–2018*, particularly through measures designed to raise their awareness of rare diseases and the issues and challenges they pose to patients and those who care and support them. These professionals will be encouraged to consider the potential of a rare disease diagnosis for the patient who displays symptoms for which no other explanation can be found. To assist in this process, they will be given access to information and best-practice guidelines that can support them to make referrals to services and specialists, and to diagnose, treat and manage rare diseases.

Encompassing more than 50 initiatives, the *WA Rare Diseases Strategic Framework 2015–2018* is an outstanding accomplishment that unites the many activities being undertaken throughout WA Health in responding to the needs of people living with a rare disease.

Tarun Weeramanthri

Assistant Director General, Public Health
WA Health

Table of contents

Executive summary	1
Introduction	1
Strategic framework development	1
Strategic framework structure	1
Section 1: Overview	2
Section 2: Background	3
Definition of a rare disease	3
Common features of rare diseases (RD)	3
Common health system needs	3
Impact on the health system	4
Process of strategic framework development	4
Section 3: The strategic framework	5
Vision	5
Aim	5
Priorities, objectives and initiatives	5
Recognition of the need for evidence	5
Priority 1: Advance RD planning in WA and Australia	7
Objective 1: Adopt a coordinated, collaborative approach	8
Priority 2: Promote a person-centred approach throughout WA Health for people living with RD	9
Objective 2: Engage with people living with RD, their carers and families	10
Objective 3: Promote active participation of people living with RD in their healthcare	11
Objective 4: Promote care coordination for people living with RD	12
Objective 5: Facilitate access for people living with RD, their carers and families to support networks and information on RD	13
Priority 3: Contribute to a high quality health system for people living with RD	14
Objective 6: Champion integration and partnerships in the delivery of healthcare for RD	15
Objective 7: Build on existing WA Health services for screening and diagnosis of RD	16
Objective 8: Encourage the use of evidence-based, best practice guidelines to deliver healthcare for RD	17
Objective 9: Identify emerging technologies to enhance the delivery of healthcare for RD	18
Objective 10: Facilitate access by health professionals to information, education and training on RD	19
Priority 4: Foster world class research on RD	20
Objective 11: Build epidemiology and health system evidence for RD	21
Objective 12: Strengthen clinical and translational research in RD	22
References	24

Executive summary

Introduction

The purpose of the *WA Rare Diseases Strategic Framework 2015–2018* is to provide a framework for the coordination of WA Health initiatives for rare diseases.

Rare diseases are a public health priority. There are approximately 190,000 Western Australians living with a rare disease, including 63,000 children. Rare diseases are commonly life-threatening or chronically debilitating, cannot be prevented, are incurable, have no effective treatments and are associated with significant pain and suffering. People living with rare diseases are likely to have multi-systemic dysfunction and hence require complex health care and access to a wide range of health services across tertiary, secondary and primary sectors.

This strategic framework draws together a range of initiatives for rare diseases that WA Health intends to deliver in 2015–2018. It provides coordination, coherence and a clear direction for activities that WA Health will undertake in the area of rare diseases. As such it is an opportunity to benefit the health and wellbeing of Western Australians living with rare diseases, their carers and families, and to support clinicians, researchers, policy-makers and WA Health to respond effectively to rare diseases.

Strategic framework development

The Office of Population Health Genomics (OPHG), Public Health Division, Department of Health WA led and coordinated the development of this strategic framework. A WA Rare Diseases Advisory group was established to provide advice to OPHG on the structure and content of the strategic framework. A series of stakeholder consultations were undertaken involving the advisory group and broader consultation open to all stakeholders in the rare diseases sector. These consultations directly and substantively informed the development of the strategic framework.

Strategic framework structure

The *WA Rare Diseases Strategic Framework 2015–2018* is structured around four priorities, 12 objectives and over 50 initiatives that are intended to respond to the needs of Western Australians living with rare diseases and support clinicians, researchers and policy-makers. The strategic framework is founded on the recognition that further evidence, in the form of up-to-date, relevant data and information, is needed about rare diseases in Western Australia. The four priorities of the strategic framework are:

1. **To advance rare diseases planning in WA and Australia.** This strategic framework represents a coordinated approach to rare diseases planning and is the inaugural state strategic framework for rare diseases in Australia.
2. **To promote a person-centred approach throughout WA Health for people living with rare diseases.** This requires being respectful of and responsive to the needs of people living with rare diseases, actively involving them in decision-making, promoting their care coordination and providing access to information and support.
3. **To contribute to a high quality health system for people living with rare diseases.** This involves progressing equitable and integrated health care for people living with rare diseases and supporting health professionals to deliver quality care for rare diseases.
4. **To foster world class research on rare diseases.** This will provide much needed evidence for rare diseases in areas such as epidemiology, health system use, clinical and translational research.

Section 1: Overview

Vision

The best possible health and wellbeing for Western Australians living with rare diseases (RD)

Aim of the WA Rare Diseases Strategic Framework

Provide a framework for the coordination of WA Health initiatives for rare diseases for 2015–2018

Priority 1

To advance RD planning in WA and Australia

Objectives

1. Adopt a coordinated, collaborative approach to RD planning.

Priority 3

To contribute to a high-quality health system for people living with RD

Objectives

6. Champion integration and partnerships in the delivery of healthcare for RD
7. Build on existing WA Health services for screening and diagnosis of RD
8. Encourage the use of evidence-based, best-practice guidelines to deliver healthcare for RD
9. Identify emerging technologies to enhance the delivery of health care for RD
10. Facilitate access by health professionals to information, education and training on RD.

Priority 2

To promote a person-centred approach throughout WA Health for people living with RD

Objectives

2. Engage with people living with RD, their carers and families
3. Promote active participation of people living with RD in their healthcare
4. Promote care coordination for people living with RD
5. Facilitate access for people living with RD, their carers and families to support networks and information on RD.

Priority 4

To foster world-class research on RD

Objectives

11. Build epidemiology and health system evidence for RD
12. Strengthen clinical and translational research in RD.

Section 2: Background

Rare diseases (RD) are a public health priority. In WA they collectively affect up to 190,000 people, including 63,000 children. There are common features across the range of RD and common health care needs expressed by those living with RD. There is a significant need for more evidence to demonstrate the impact on Western Australians living with RD, their carers and families, the wider community and the healthcare system [1].

Definition of a rare disease

Countries have different definitions of a rare disease, although most definitions refer to low prevalence in the population. It has been proposed by the rare diseases community that Australia adopts the European Union consumer endorsed definition which refers to both prevalence and severity of burden [2]. This definition indicates that RD are “life-threatening or chronically debilitating diseases which are of such low prevalence (1 in 2,000 people) that special combined efforts are needed to address them” [3, 4].

There are 5,000–8,000 known RD which when combined are estimated to affect up to 6–8% of the population [5]. This equates to 1.2 million Australians and is the best available estimate of the prevalence of RD in Australia. There is little data in Australia to accurately identify the number of new cases (incidence) and proportion of people living with a RD (prevalence).

Common features of rare diseases

Many RD onset during childhood and continue throughout life, although some do not become evident until adulthood. Around 80% of RD have a known genetic association. Most cannot be prevented, are complex with multi-system dysfunction, disabling, incurable and have no effective treatment [6, 7]. European studies show that 50% of RD are associated with motor, sensory or intellectual impairment, 30% of RD lead to an incapacity which reduces autonomy and 35% of deaths that occur before the age of one year can be attributed to RD [8].

RD are often associated with significant pain and suffering. Studies from Europe and the United States (US) show that people living with rare diseases, carers and families experience the burden of RD in terms of an impact on health, social, financial and emotional states [7, 9, 10]. Further investigation is required into the experiences of people in WA, which anecdotally are described as similar to experiences internationally. An increase in local evidence will inform the further development of health and social policy for RD.

Common health system needs

Consultation with the RD community indicates that common features across the range of RD lead to common needs from the health system [11]. People living with RD are likely to require complex case management and access to a range of government and non-government services and programs. This includes allied health professionals, clinical specialists, general practitioners, and disability and social support services. People with rare diseases want to be partners in the provision of care and want services to be patient-centric, flexible, integrated and coordinated to provide “seamless” care across the whole-of-lifetime. This includes transitions from paediatric to adult to aged care and changes in location of residence which result in services being accessed in another region (e.g. patients who move interstate or from rural to metropolitan settings or vice versa).

People living with RD have reported experiences of a diagnostic journey involving delayed or inaccurate diagnosis and treatment. An early, accurate diagnosis can impact on disease progression, reduce complications and co-morbidities and improve quality of life [12, 13]. Once diagnosed, patients, carers and families need useful, reliable and timely information to

inform their decision-making about ongoing care and treatment. Health professionals need access to information about RD to support them to provide early and accurate diagnosis. They also need best practice guidelines, such as models of care and/or clinical pathways that focus on the commonalities across RD.

People living with RD want their information shared among health professionals and others involved in their care, using modern technologies. They also need access to health professionals who are aware of, experienced with and knowledgeable about RD. They need health professionals to question whether the people they see, whose symptoms they can't explain, have a rare disease and to have a good understanding of the services and specialists to which they may refer patients. This requires that health professionals are supported and provided ongoing access to information, education and training about RD.

Most RD have no effective treatment [14]. Hence many people living with RD want access to clinical trials of drug treatments and other therapies that are being developed. Because there are relatively few people with each rare disease in Australia, as in other countries, there is a need for people with RD to gain access to multi-country, international clinical trials. Infrastructure, such as patient registries, is required to facilitate the involvement of Australian patients in such trials. Research to investigate the causes of and treatments for RD is also critical to enhancing healthcare and outcomes for people living with RD. Local and national partnerships are needed to facilitate research opportunities that translate to benefits in clinical care and public health.

Impact on the health system

While evidence is limited, the complexity of care requirements means it is likely that RD have a significant impact on the health system. This claim is supported by a Western Australian study that shows rare genetic disorders result in increased hospital admissions and longer lengths of stay, for both adults and children [15, 16]. This suggests that the economic burden of RD on the health system is disproportionately high compared to other patients.

Process of strategic framework development

The Office of Population Health Genomics (OPHG), Public Health Division, Department of Health WA led and coordinated the development of this strategic framework. The phases involved in strategic framework development included:

- a comprehensive review of international and national literature relating to RD
- the establishment of an expert advisory group to provide advice to OPHG on the structure and content of the strategic framework
- stakeholder consultations, including:
 - a workshop with the expert advisory group to explore key strengths, opportunities, aspirations and potential results for a state RD strategic framework.
 - broad consultation on a draft strategic framework through a stakeholder survey and call for written submissions on the strategic framework. These avenues of consultation were open to all stakeholders in the RD community.
 - final input to the strategic framework from the expert advisory group, following updates being made to incorporate feedback from the earlier stages of consultation.

The strategic framework has been endorsed by the WA State Health Executive Forum (SHEF) and the Director General of Health.

OPHG will coordinate the implementation, monitoring and evaluation of this strategic framework. Evaluation will occur mid term and at the end of the four year period of the strategic framework.

Section 3: The strategic framework

Vision

The vision of the *WA Rare Diseases Strategic Framework 2015–2018* is to facilitate the best possible health and wellbeing for Western Australians living with RD.

People living with RD typically have severe conditions with complex care requirements and as such are a population with a great need for care. One of the four pillars of WA Health's *Strategic Intent 2010–2015* is caring for those who need it most. This involves working to ensure that health services are available as needed and serving to improve the health and wellbeing of those for whom need is greatest [17].

This strategic framework expresses WA Health's intentions for specific measures in relation to RD. As such it is an opportunity to benefit the health and wellbeing of the estimated 190,000 Western Australians living with RD, their carers and families, and to support clinicians, researchers, policy-makers and WA Health to respond effectively to RD.

Aim

The aim is to provide a framework for the coordination of WA Health initiatives for RD for 2015–2018.

WA Health already delivers a range of initiatives for people living with RD and further initiatives are planned for the next four years. This strategic framework enables these initiatives to be drawn together into a cohesive framework, and promotes the coordination of activities for RD across the Western Australian health system.

This strategic framework builds on what already exists and provides a clear direction and coherence to the different initiatives that are being and will be undertaken. It also acknowledges what currently works well in the health system and identifies opportunities for the future. It enables a collective view to be taken of the commonalities among RD which is more efficient and effective than individual strategic responses to the 5,000–8,000 RD that are known to exist.

A coordinated approach to RD, through this strategic framework, is desired and has been informed by people living with RD, their carers, families and support organisations, clinicians, researchers and policy-makers.

Priorities, objectives and initiatives

This strategic framework is structured around four priorities, 12 objectives and over 50 initiatives that are intended to respond to the needs of Western Australians living with RD.

Recognition of the need for evidence

To provide policies, programs and practices that effectively respond to the needs of Western Australians living with RD, it is imperative that these be built on a solid foundation of evidence and be monitored and evaluated. Evidence-informed decision-making is a structured, objective way to build a holistic understanding of the context in which relevant, efficient, cost-effective policies, programs and practices can be developed and implemented. It helps ensure that policies, programs and practices are responding to the real needs of the community and that improved health and wellbeing outcomes are being achieved.

At present, there are gaps in the evidence related to RD in WA (as in the whole of Australia). Baseline data and information on the RD landscape is required, which includes the need to systematically assess the healthcare experiences of Western Australians living with RD and the impact of RD on the health system. A greater understanding of existing and potential policies, services, programs, research, resources and organisational structures for RD will provide a more comprehensive picture of the way in which WA Health can and does respond to the needs of Western Australians living with RD. This will identify elements of the healthcare system that support RD, areas of strength and opportunities for improvement and a better understanding of the interactions of system components. Mechanisms are needed to record and report epidemiology and economic burden of disease, health outcomes and the quality and equity of access to clinical services.

This strategic framework is built on the recognition of the need for more up-to-date, relevant data and information about RD in WA. Building the evidence base will improve the visibility of RD in the health system and improve the ability of policy makers, service planners and service providers to make evidence-informed decisions in relation to RD. The data and information gathered through the initiatives of this strategic framework will be analysed and integrated to inform the direction of future planning for RD in Western Australia. The evidence accrued as a result of this strategic framework will:

- support the government and healthcare system to plan and respond to RD
- support those living with a rare disease, their family and carers
- strengthen data available on RD
- provide the basis for an evidence-informed approach to RD in WA and Australia
- enable more accurate service planning and policy development
- support clinicians providing care
- identify best practice approaches.

Priority 1: Advance RD planning in WA and Australia

WA is the first state in Australia to develop a RD strategic framework. This provides the opportunity for WA to lead the way in responding to the needs of people living with RD, their carers and families and to provide support for clinicians, researchers and policy-makers.

WA has also been integral to national efforts related to RD planning, including the development in 2013 of a *Scoping paper on the need for a national plan for RD*, for the Australian Health Minister's Advisory Council [18]. This was preceded in 2011 by the organisation of the first national RD symposium called *Awakening Australia to rare diseases: Global perspectives on establishing a coordinated approach to a national plan*. This brought together nearly 200 stakeholders including people living with RD, their carers and families, advocates and patient support groups, medical specialists, allied health practitioners, social and disability services representatives, industry representatives (e.g. pharmaceutical, biotechnology and medical device companies), researchers, and state and commonwealth policy-makers.

During the symposium a series of stakeholder consultation workshops were conducted to determine perspectives on a range of issues including: strategic planning for RD; patient empowerment; patient care, support and management; research; networks; and partnerships and collaboration. The outcomes of these workshops [2, 11] included the need for:

- a national peak body for people living with RD, which led to the establishment of Rare Voices Australia (www.rarevoices.org.au)
- a coordinated planning approach to RD
- collaboration and stakeholder networks within and across clinical care and research
- information, data and evidence to inform strategic planning, funding allocation and research. This includes a standard definition of RD, publicly available information on RD, awareness and education programs and audits that identify the “current situation” and good practice models
- coordinated, whole-of-lifetime care where there are no gaps in service delivery across the lifespan of people living with RD
- information sharing between patients, carers, families and health professionals, and systems that enable the sharing of clinical and other health information within and across health professionals and services
- support for health professionals to provide effective services, including resources and tools (e.g. referral guidelines, education programs), primary healthcare partnerships and access to information on RD
- support for people living with RD and their carers and families, including access to support groups and information on RD.

Similar views were expressed by the WA Rare Diseases Advisory Group and other stakeholders during consultations specifically for the development of this strategic framework. As such the majority of these issues are addressed by the priorities and objectives of this strategic framework.

Objective 1: Adopt a coordinated, collaborative approach

This strategic framework brings a range of WA Health initiatives for RD together into a single framework. It aligns with other WA Health documents which have elements relevant to RD planning. These include the *WA Chronic Conditions Framework 2011–2016*, *Our Children Our Future – A Framework for Child and Youth Health Services in Western Australia 2008–2012* (currently being updated) and *Palliative Model of Care*. It is important to encourage future WA Health policies, strategies, frameworks and models of care to consider RD and align with this strategic framework.

Coordinated planning for RD begins by taking a collective view of RD. This recognises that RD have commonalities and in total affect up to 6–8% of the population, thus representing a significant proportion of Western Australians who use WA Health services. A collective view should raise the profile of RD in a health system that is oriented towards more common diseases and is more efficient than if planning efforts were duplicated many times over for the 5,000–8,000 individual RD.

Effective implementation of the initiatives in this strategic framework must involve collaboration, networking and partnerships with local, national and international RD stakeholders. This recognises that links between people living with RD, their carers and families, healthcare providers, researchers, industry and policy makers are vital to facilitate the best possible health and wellbeing for people living with RD. National and international collaborations and partnerships will build on strong local networks. Extending beyond local borders enables WA Health to draw on the international experiences of countries that are further progressed with RD planning and facilitates the coordination of local access to national and international resources for RD.

In addition to state initiatives, it is imperative that coordinated planning for RD occurs at a national level. This will bring Australia in line with other countries which have adopted national plans and initiatives to respond to the needs of people living with RD. WA Health has already made significant contributions at a national level and will continue to engage in and sponsor initiatives that promote the need for RD planning at the national level.

Initiatives

1. Encourage the consideration of RD in WA Health policies, strategies, frameworks and models of care.
2. Represent RD on relevant WA Health Networks.
3. Promote a collective view of rare diseases and the adoption by WA Health of a standard definition of RD.
4. Promote sustainability for RD planning in WA.
5. Foster collaboration, networking and partnerships with local, national and international stakeholders.
6. Build the capacity of community service organisations in the rare diseases sector to develop and contribute to networks, collaborations and partnerships, including those with WA Health.
7. Establish a WA RD advisory group.
8. Support initiatives to advance RD planning at the national level.

Priority 2: Promote a person-centred approach throughout WA Health for people living with RD

WA Health is committed to a person-centred approach to healthcare for all Western Australians, including those living with RD. This means being respectful of and responsive to the values, preferences and needs of people living with RD, their carers and families. WA Health encourages and supports participation in decision-making and recognises that people living with RD, their carers and families have a central role in:

- individual care, which requires a respectful and collaborative partnership between healthcare users and providers in which both are partners in planning, goal-setting, developing and assessing care to make sure it is most appropriate for their needs [19]
- health system and service planning and delivery, including program and policy development, quality improvement, patient safety initiatives and healthcare design [13, 20, 21].

There is evidence that a person-centred approach is sustainable and can lead to improved healthcare quality and better health outcomes including improved safety, cost effectiveness and patient, family and staff satisfaction [19, 22-25]. For example, the Australian Commission on Safety and Quality in Health Care cites a body of evidence illustrating links between person-centred care, also called patient-centred care, and decreased readmission rates and healthcare acquired infections; improved delivery of preventive care services; reduced hospital stays and enhanced compliance with treatment regimens [25].

A range of principles and practices are relevant to a person-centred approach [26-29] and several of these are directly addressed by this strategic framework as follows:

- engagement of people living with RD, their carers and families in service and system-level decision-making (see Objective 2)
- active involvement and choice in their own care for people living with RD (see Objective 3)
- care coordination (see Objective 4)
- information for people living with RD, their carers and families to make informed decisions and access to support (see Objective 5)
- integrated care across healthcare providers (see Objective 6).

Objective 2: Engage with people living with RD, their carers and families

Engagement is a process by which the aspirations, concerns, needs and values of people living with RD, their carers and families can be incorporated in WA Health decision-making [30]. Engagement recognises that people have the right to be included in the decision-making processes that affect their healthcare and that people living with RD, their carers and families have unique knowledge and experiences that can contribute to health system decision-making. Engagement with people living with RD, their carers and families should occur in all areas of the health system including: the development of policies and strategies; service planning, design/redesign, delivery and evaluation; quality improvement; and the development of education resources [21, 31-33]. The *WA Health Consumer Carer and Community Engagement Framework 2011–2016* [30] establishes engagement as an integral part of core business for WA Health. The Framework must be accompanied by the development of a culture of engagement so that people living with RD, carers and families are genuinely involved in decision-making.

This strategic framework commits WA Health to engaging people living with RD, carers, families and community service organisations in the RD sector by way of: their involvement in key organisational committees and appointments to advisory and governing structures (e.g. the RD advisory group); partnering to implement the initiatives of this strategic framework; and adopting mechanisms to ensure their views and needs are understood by WA Health. Throughout these processes consideration will be given to ways of involving and addressing the needs of harder-to-reach groups such as Aboriginal Australians, people from culturally and linguistically diverse (CALD) backgrounds and people living in rural/remote areas.

Emerging evidence indicates that engagement contributes to improved health outcomes at the individual, service, network and systems level [22, 24]. It results in a more responsive and integrated health system and helps to achieve long term sustainability [34] and equitable and effective decision-making for health services planning [35]. In general a service designed with an understanding of the views and needs of those who use it is more likely to effectively target these needs.

Initiatives

1. Include people living with RD, carers, families and their representatives on the state RD advisory group and other committees, advisory groups and working groups that are established to implement this strategic framework.
2. Consider ways to involve and address the needs of harder-to-reach populations, such as Aboriginal, CALD and rural/remote populations.
3. Build the capacity of community service organisations in the rare diseases sector to investigate and represent to WA Health the views and needs of people living with RD, their carers and families.

Objective 3: Promote active participation of people living with RD in their healthcare

Self-management refers to what a person living with a disease does to be actively involved in their own healthcare [36]. It involves the person knowing about their condition, sharing decision-making about their healthcare, following agreed care plans, monitoring and managing symptoms of their condition, managing the impacts of their condition on physical, emotional and social life, and having confidence to access community support services [36].

Self-management support is what carers, healthcare providers and systems do to increase the capacity of people living with diseases to actively participate in their own healthcare. Support can be invaluable to: link people to personal, medical, disability and community resources, including psychological and allied health services; provide strategies for care planning and negotiating the health system; and address medication management, pain control, risk reduction, behaviour change and learning to interpret changes in the disease [13, 36, 37].

Optimising self-management is considered essential for people with long term, chronic health conditions [13]. As such, self-management is a guiding principle of the *WA Chronic Health Conditions Framework 2011–2016* [12]. Further, WA Health has developed the *WA Chronic Conditions Self-Management Strategic Framework 2011–2015* [36] which aims to: support system and practice changes to incorporate self-management into the management of chronic conditions; train healthcare professionals to assist people living with chronic conditions to actively self-manage their health; and develop and implement chronic conditions self-management programs and services [36]. RD are chronic conditions and therefore need to be considered and included in the implementation of these activities.

For some RD, such as cystic fibrosis, there are WA models of care that incorporate principles of self-management and self-management support [38]. However, further studies are required to determine the relevance and effectiveness of the self management approach across the broad spectrum of RD. There is little evidence of whether self-management for people living with RD achieves outcomes similar to those achieved for chronic conditions such as diabetes, heart disease and obesity. These outcomes include better health service utilisation, uptake of behaviours that maximise health, wellbeing and quality of life [13, 26, 39], improved quality of care and clinical outcomes and reduced health system costs [13, 36, 40–44].

Initiatives

1. Represent RD in the implementation of the *WA Chronic Conditions Self Management Strategic Framework 2011–2015*.
2. Investigate the relevance and effectiveness of self-management and self management support to the care and management of RD from the perspective of all stakeholders.
3. Map self-management supports that are being or should be implemented in the WA health system for people living with RD.

Objective 4: Promote care coordination for people living with RD

People living with RD often have complex and long term care needs. As such, they often see multiple clinicians across the spectrum of primary, secondary and tertiary healthcare in the private and public sectors, as well as accessing disability and social services [45]. Ideally their care is coordinated across this range of health and community settings [46] so that: continuity and communication between health care providers is maintained; health service users can navigate the system; appointments and visits are well scheduled; health professionals are able to share information to ensure timely diagnosis, early intervention and ongoing care; tests are not unnecessarily repeated; and patients know who to contact for advice and the support they need.

Coordinated care has been shown to: improve disease management; improve access to services and the quality and consistency of healthcare provision; and reduce unnecessary hospitalisations, resulting in cost savings and a more efficient health system [12, 13, 20, 34, 42, 47–54]. WA Health has demonstrated a commitment to care coordination in the *WA Chronic Health Conditions Framework 2011–16* [12] and *Our Children Our Future – A Framework for Child and Youth Health Services in WA 2008–2012* [45] and in several models of care developed for specific RD (e.g. cystic fibrosis, motor neuron disease and coeliac disease).

A number of approaches to care coordination have been proposed including individualised care plans [55, 56], care coordinators [57, 58] and having access to multidisciplinary clinics. In Australia, general practitioners (GPs) often hold a coordinating role, particularly for people who live further away from major centres of healthcare. The role in care coordination of primary care providers, such as GPs, needs further exploration in the context of RD. This should involve collaborations with relevant organisations and professional bodies such as the Royal Australian College of General Practitioners (RACGP) and primary healthcare networks and organisations.

Internationally, multi-disciplinary centres of expertise in RD are promoted as mechanisms for care coordination. Such centres bring together health professionals from a range of medical and allied health disciplines, to provide coordinated, team based care, including diagnosis, follow-up and management. They are also a focal point for medical training, research in RD and information sharing [5, 59-61]. The extent to which this model would be applicable in WA is unknown and will be explored as an initiative within this strategic framework.

Initiatives

1. Gather and publish evidence of the experiences of care coordination in WA among people living with RD, identifying the core elements of care coordination and opportunities for improvement in care coordination.
2. Map existing services that provide care coordination for people living with RD.
3. Scope the need for new or expanded services for care coordination for people living with RD, across WA Health and the broader health system for RD.
4. Collaborate with relevant organisations to explore the care coordination role of primary care providers, such as GPs, in the context of RD.
5. Develop a WA Health policy on the need for WA centres of expertise in RD.

Objective 5: Facilitate access for people living with RD, their carers and families to support networks and information on RD

Access to the right information at the right time is essential for effective healthcare. It enables people to navigate the health system and to make informed decisions about how to manage their own health [12, 13, 20, 34]. Access to information is a critical component of a person-centred approach and self-management. It is essential before, during and after diagnosis and should be a significant part of ongoing care. This includes access to information on support networks and groups.

Consultations with people living with RD and their carers and families show they want information about RD to be publicly available through avenues such as websites. They want this information to be centralised, reliable, accurate and easy to understand [11]. The types of information that are desired include: treatment and management options; a directory of specialists; local medical and social services; entitlements (e.g. financial); and disease-specific patient support groups [11].

Orphanet is a leading international web portal of information on RD and orphan drugs (www.orpha.net). It is for all audiences and aims to help improve the diagnosis, care and treatment of patients with RD. Orphanet contains directories of information including an: inventory of RD; inventory of orphan drugs; assistance to diagnose tool; and directories of national specialist clinics, medical laboratories, research projects, clinical trials, patient/clinical registries and patient organisations. Originally Orphanet contained European information only. In more recent years it has expanded to include information for non-European countries and WA Health has joined Orphanet as the country coordinator to provide Australian information to Orphanet. This will include information on clinics, laboratories, research projects, clinical trials, registries and patient organisations in WA.

Support groups and networks typically play an important role in providing access to information and education, for those directly affected by RD as well as health professionals and the general public. Support groups may also encourage and fund research and treatment, engage in advocacy and provide social and emotional support and a sense of community through opportunities to share experiences, knowledge, coping strategies and skills with others who are in a similar situation. In Australia there are support groups for specific RD as well as umbrella groups for a range of diseases. Links are needed between support organisations and people living with RD. Many RD do not have a support group and initiatives are required to promote access to information and support for people living with these diseases.

Initiatives

1. Raise awareness among the RD community of the internationally recognised Orphanet web-based portal of information on RD.
2. Populate the Orphanet databases with WA content.
3. Build the capacity of community service organisations in the RD sector to provide increased access for people living with RD to resources, support and information on health and other services.

Priority 3: Contribute to a high quality health system for people living with RD

WA Health offers high quality universal healthcare to all Western Australians and is part of a broader healthcare system that delivers services for people living with RD. Most people living with RD have chronic, long term conditions with complex care requirements. As a result, many need to access a wide range of services within WA Health, private health, disability and non-government sectors [45]. This includes primary care (e.g. general practitioners), secondary care (e.g. medical specialists and allied health professionals such as physiotherapists, occupational therapists, speech therapists, podiatrists etc) and tertiary care (e.g. hospital-based services).

Consultation with the RD community indicates a desire for features of the healthcare system to include:

- high visibility of RD in the healthcare system
- equitable access to healthcare for all people living with RD
- timely and appropriate diagnosis of RD
- no gaps in service delivery across the lifespan of people living with RD
- coordinated healthcare across the spectrum of services accessed
- partnerships between primary, secondary and tertiary service providers
- sharing of clinical and other health information within and across health professionals, services, settings and locations (e.g. across States)
- development of resources and tools to support health professionals in providing care for people living with RD (e.g. referral guidelines, education programs, access to information on RD).

The initiatives under this priority have been developed to address the needs expressed by the RD community. They strive to develop a clearer understanding of the range, coordination and integration of services accessed by people living in WA, which may identify gaps in service delivery and inform the services offered by WA Health (see Objective 6); support health professionals to provide timely and accurate diagnosis (see Objective 7); provide health professionals with better access to best-practice clinical guidelines (see Objective 8); explore the use of e-technologies for innovative approaches to service delivery and the sharing of information between health professionals (see Objective 9) and assist health professionals with education and training in RD (see Objective 10).

Implementing these initiatives is intended to generate higher visibility for RD in the health system and health professionals will have greater awareness of RD, a better understanding of the services that people living with RD use and are referred to and are better supported to provide evidence-based care. For people living with RD, it is intended that these initiatives will contribute to progress in equitable access to healthcare and integrated healthcare for people living with RD.

Objective 6: Champion integration and partnerships in the delivery of healthcare for RD

The complexity of RD means that most people living with them require access to multiple services within the healthcare system, across the spectrum of primary, secondary and tertiary care both within WA Health and in the private health, disability and non-government sectors. Ideally patients experience “seamless” interfaces, that is, integrated care across the different services, settings, agencies and sectors they access and over time [12, 13, 20, 26].

A number of ways of achieving greater integration have been proposed including integrated care pathways, models of care and a partnership approach [45]. A key integration issue is the need for effective transitions between care settings. This is both between health services, such as hospital and community-based providers, and across phases of care (e.g. transition from child to adult services and to end-of-life services). *The Paediatric Chronic Disease Transition Framework* [62] provides a guide to transition planning from paediatric to adult healthcare services in WA, which is particularly relevant for people living with RD. As such it is important that RD are represented in the implementation of this framework.

In line with the *WA Primary Health Care Strategy* [63] this strategic framework recognises the importance of partnerships and integration between public hospital services, specialist services and primary healthcare providers (e.g. GPs) to creating a seamless care pathway for health service users. It complements a range of frameworks and models of care, including the *WA Chronic Health Conditions Framework 2011–16* [12], *Our Children Our Future – A Framework for Child and Youth Health Services in WA 2008–2012* [45], and the *Paediatric and Adolescent Palliative Care Model of Care* [64] that also recognise strengthened links across primary, secondary and tertiary care are needed to reduce duplication, improve efficient resource use and improve continuity of care and the management of people with chronic and/or life-limiting conditions, including RD.

Initiatives

1. Map existing WA Health services accessed by people living with RD and how these interface and link with the wider health system.
2. Explore integration of healthcare services for RD from the perspective of healthcare providers in primary, secondary and tertiary care.
3. Build the capacity of community service organisations in the RD sector to link people living with RD, their carers and families, to existing healthcare and other services.
4. Build evidence of the healthcare experiences of people living with rare diseases, their carers and families in relation to integration of healthcare services.
5. Represent RD in the implementation of the *Paediatric Chronic Disease Transition Framework*.

Objective 7: Build on existing WA Health services for screening and diagnosis of RD

Diagnosis is an important element of the patient journey. An accurate and early diagnosis provides those living with a rare disease and their families and carers an understanding of future healthcare requirements. In addition, early diagnosis supports early treatment, which can improve health outcomes by delaying disease progression, reducing complications and premature mortality, and improving quality of life [12, 13, 65]. Early diagnosis and treatment may also avoid unnecessary hospital admissions and related complex and expensive treatments [12, 13, 65].

There are multiple pathways to the diagnosis of RD in WA, including: the Newborn Screening (NBS) Program; referral from a community health nurse to the Statewide Child Development Service; and referral from a GP to Genetic Services WA or a medical specialist. Sometimes multiple specialists, in multiple locations including interstate and overseas, must work together to collectively arrive at a diagnosis. The full range of diagnostic options is not well understood and initiatives within this strategic framework will enable this to be more fully investigated. This strategic framework will also investigate a proposal from RD stakeholders that a RD clinic is required to increase capacity in referral pathways for RD from primary and community care providers to specialists.

NBS is an important tool that enables early diagnosis and treatment of more than 25 RD. In Australia, NBS programs are implemented by each state/territory government. The WA NBS program is coordinated by the Department of Clinical Biochemistry, Princess Margaret Hospital (PMH) and publically funded. This program currently screens more than 99% of newborns [18]. There are calls to expand NBS programs to incorporate additional RD. A decision framework, which supports consistent consideration of the benefits and harms of screening, is essential to informing any decisions to change the scope of newborn screening, particularly for RD, for which there are often no cures and a lack of treatment options [18].

Screening is not available for all RD, and where it is available, requires follow-up diagnostic testing. Diagnosis can be made through avenues including clinical assessment (e.g. MRI scans, X-rays, dysmorphology) and diagnostic testing (e.g. genetic, metabolic and immunological tests). Evolving approaches to testing, such as next generation sequencing, are increasingly important for RD diagnosis. Diagnostic tests are analysed by laboratories. It is important for clinicians to know the appropriate tests to use and which laboratories analyse which diagnostic tests.

Initiatives

1. Evaluate current referral pathways for the diagnosis of RD, including ways to increase access to interstate/overseas experts in RD for diagnosis.
2. Scope the need for and feasibility of mechanisms (e.g. a RD clinic) to facilitate referral pathways from primary/community care for diagnosis of RD.
3. Generate a list of diagnostic tests and Australian laboratories that perform these tests and make information on these publicly available in Orphanet.
4. Promote a consistent, nationwide approach to the provision of NBS for RD and the development of a decision-making framework to support the consideration of conditions for inclusion in the newborn screening panel.

Objective 8: Encourage the use of evidence-based, best practice guidelines to deliver healthcare for RD

The RD community in Australia has recognised a need to develop models of care and/or best-practice guidelines specific to RD [11]. These guidelines should aim to increase access to evidence-based and person-centred care for people living with RD across general practice, acute and community care.

In the clinical context, best practice guidelines facilitate the implementation of healthcare strategies and aid in the improvement of overall disease management as well as ensuring consistency across the provision of clinical care [66, 67]. Guidelines are essential for effective disease management, early identification, self-management and service integration [42]. Documented referral pathways are also important to ensure that patients requiring complex and multidisciplinary care do not get lost in the system [33, 66]. It has been shown that when evidence-based guidelines are available and integrated into everyday practice there are improved health outcomes for patients [42].

WA Health has a strong approach to the development of evidence based models of care for diseases, condition or population groups. Since its establishment in 2006, WA Health Networks has coordinated the development of over 60 models of care. These outline the principles and directions that apply to the provision of healthcare services to deliver the right care, in the right place, at the right time by the right team [68]. In particular they focus on the systemic structures and strategies to improve service delivery. There currently is no overarching model of care for RD and the need (or otherwise) for one requires consideration.

WA Health is a collaborating partner in the RARE-Bestpractices project (www.rarebestpractices.eu). This four year international project, which commenced in 2012, will develop a sustainable global networking platform, supporting the collection of standardised and validated data and the exchange of knowledge and reliable information among 14 countries. The main goal of RARE Bestpractices is to improve clinical management of patients living with RD and to narrow any gaps in the provision of high quality healthcare. This will be achieved by identifying, evaluating and disseminating best practice guidelines and sharing this knowledge globally.

Initiatives

1. Identify best practice documents for RD (e.g. clinical guidelines, models of care).
2. Scope the need for a WA model of care for RD.
3. Identify mechanisms for best practice delivery of healthcare for people with RD in rural and remote areas.
4. Contribute to state, national and international platforms to develop best practices for RD.

Objective 9: Identify emerging technologies to enhance the delivery of healthcare for RD

In recent decades significant advances in the information and communication technology sector have led to a number of emerging technologies aimed at promoting health system improvement. Telemedicine and e-health are two areas that have the potential to significantly impact on the delivery of equitable, cost-effective, efficient, person-centred and integrated care for people living with RD.

e-health is defined as 'the combined use of electronic communication and information technology in the health sector' [69]. In more practical terms, e-health is a means for providing the right health information, to the right person, at the right place and time, in a secure, electronic form for the purpose of optimising the quality and efficiency of healthcare [69]. Implementation of various e-health solutions across the health system will help to integrate care, reduce potential medication errors and duplication of services, support the delivery of quality primary healthcare services and improve patient outcomes [69]. These potential benefits are particularly relevant to people living with RD whose complex and long term conditions require a range of health and social care delivered by a variety of health professionals.

Telemedicine is the use of advanced telecommunication technologies to exchange health information and provide healthcare services across geographic, time, social and cultural barriers [70]. Telemedicine aims to improve access to medical services for patients who have difficulty getting to a specialist and/or live in rural and remote areas [71]. Therefore telemedicine has the potential to improve equity in healthcare access for people living with RD, particularly those who are poorly mobile or living with a disability, are living in outer metropolitan, rural and remote areas or who have RD for which clinical expertise is geographically disperse.

A program currently using telemedicine technologies to improve service delivery for RD patients is the metabolic telehealth clinic. This clinic provided by the metabolic team at PMH allows families living in rural WA to have their appointments by videoconferencing, rather than attend PMH in person. Since it was established in May 2011, over 80 patients have accessed the metabolic telehealth clinic with very positive client feedback and significant cost savings for both families and WA Health [72]. This suggests the use of e-health and telemedicine warrants further investigation in the RD environment.

Initiatives

1. Scope the need for and availability of e-health solutions that could enhance the sharing of RD patient information.
2. Scope the need for and availability of telemedicine/e-health as a means of delivering services for people living with RD, including in rural/remote settings.

Objective 10: Facilitate access by health professionals to information, education and training on RD

A key to the success of WA's healthcare system is that patients have access to a highly skilled and motivated medical workforce employed in general practice, community and hospital settings.

International evidence indicates that RD pose a challenge to health professionals across all sectors of the health system [1, 73–76]. They can experience difficulties managing patients with RD due to lack of opportunities to learn about the vast number of RD and lack of easy access to information, expert advice, management guidelines and referral pathways. Primary care clinicians and paediatricians believe there is insufficient knowledge about RD and want access to evidence-based information for themselves and their patients about diagnosis, management and specialised referral clinics [1, 73, 76, 77].

There is a need to provide appropriate education, training and information resources to support health professionals to diagnose and manage RD. With more than 5,000 known RD and an estimated 190,000 people in WA living with a RD, it is likely that health professionals will come across some patients with RD. While it is clearly not possible for health professionals to know about every rare disease, it is possible for them to have greater awareness of RD as a collective group and to question whether the people they see, whose symptoms they find difficult to explain, have a rare disease. Health professionals might also benefit from greater access to information on the services and specialists to whom they might refer RD patients.

Education, information and training will raise awareness of RD and the issues and challenges they pose for people living with RD, their carers and families. It will also support health professionals to effectively diagnose and provide ongoing information, advice and clinical management for people living with RD. It is critical that the perspectives of both health professionals and people living with RD, their carers and families are understood during the development of education, information and training tools.

Initiatives

1. Identify specific information, education and training needs in RD among healthcare professionals.
2. Engage with professional bodies to explore opportunities to incorporate training in RD in professional development and continuing education programs.
3. Build the capacity of community service organisations in the RD sector to develop and disseminate information to healthcare professionals on RD that is relevant in the WA context.
4. Raise awareness among healthcare professionals of the internationally recognised Orphanet web-based portal of information on RD.

Priority 4: Foster world class research on RD

For many years WA has produced and attracted highly skilled and world class scientific and medical researchers. This rich and successful history in medical and health research has resulted in a healthier population and led to innovations that have boosted the efficiency and effectiveness of our healthcare system. Yet there is more to discover and RD are one area where new knowledge is required to improve prevention, diagnosis and treatment.

The field of rare disease research provides a multitude of opportunities. For many RD basic knowledge, such as the cause of the disease, pathophysiology, natural course of the disease and epidemiological data is limited [78]. Scientific and medical research is vital for increasing our knowledge of these RD. As more is learned about the underlying biology of an illness, that understanding can be applied to developing diagnostic tests that help individuals and their families who are living with RD. Research into RD can also give insights into common conditions and health generally [79].

This strategic framework includes initiatives that look to build upon WA's high quality academic and research sector and existing contribution to RD research on the international stage. It will build on the foundation of successful research models and local needs, innovation and networks. Momentum for clinical and translational research in RD has grown, with numerous disease specific projects currently underway. Existing research projects are located across the state in universities, hospital and health services, government departments and in the private sector. This could potentially benefit from a more coordinated approach to make the most impact for RD.

In addition to local initiatives, national and international research collaborations and networks are seen as a primary enabler for progress in RD research. Such collaborations are essential to provide adequate case numbers for meaningful studies and to bring together specialised multidisciplinary expertise [78]. Currently, WA Health is involved in a number of initiatives that are national or global in scope. These initiatives aim to bring about collaboration and integration of research tasks through the provision of robust tools for large scale data and sample sharing across multiple research projects [18].

Significant efforts are being made by state and federal governments, tertiary institutions and industry to further the understanding of RD. However, unmet opportunities for research remain. There is a need for concerted actions to advance the science of medicine and improve the effectiveness of healthcare [18].

Most of the opportunities described in this section relate to the provision of integrated platforms for connecting registries, biobanks and clinical informatics and the harnessing of these and other platforms to deliver advances in diagnosis and treatment.

Objective 11: Build epidemiology and health system evidence for RD

Effective monitoring and surveillance are an essential component in health service planning and resource allocation. Building efficient and integrated systems of care are reliant on providing the appropriate level and mix of services to meet population needs and this requires the availability of both population and service level data for planning, monitoring and reporting service need, use and effectiveness [13, 20, 65].

WA has a research community well equipped to assist with and undertake epidemiological studies and collect and analyse health service information. However data and information on RD is limited. Studies and mechanisms are needed to record and report data on prevalence, incidence, morbidity, mortality, economic burden of disease, health outcomes and clinical services. This would contribute to understandings of the collective impact of RD, which would improve the ability of policy-makers and service providers to make evidence-based decisions and build evidence-based policies and services for people living with RD.

Coding and classification of RD have been described as a major limitation for epidemiological surveillance and monitoring for RD. The international reference for classification of diseases and conditions is the International Classification of Diseases (ICD), coordinated by the World Health Organisation. The current version of this classification system, ICD-10, is used in both public and private health settings in WA. Unfortunately, ICD-10 does not support comprehensive reporting of RD, because currently codes exist for only about 3.5% of RD [60].

Limitations with the current coding of RD is likely to result in significant under reporting of RD and limits the ability of WA Health to collect quality data for analyses of the: individual and collective prevalence and incidence of RD; impact and burden of RD on individuals, carers, families and the healthcare system; quality and equity of access to healthcare services; and the impact of future changes in policy and service implementation on health and societal outcomes.

Shortfalls of current coding are to be somewhat addressed through the next release of ICD codes, ICD-11. Orphanet has developed a new set of classifications for RD that will be formally adopted in ICD-11. This means that there will be an internationally accepted, comprehensive data classification system that supports RD. Given that WA Health currently uses ICD-10, it can therefore be expected, that as ICD-11 is gradually rolled-out, there will be the capacity to more effectively record and report local RD data.

Initiatives

1. Promote the use of the best available coding/classification for rare disease within WA Health.
2. Gather evidence of health system use from the perspective of RD patients and carers.
3. Conduct epidemiological studies to investigate health system use by people living with rare diseases in WA.

Objective 12: Strengthen clinical and translational research in RD

Supporting and developing capacity in clinical and translational research is critical to providing optimal healthcare for people living with RD in WA. Clinical research focuses on the development of diagnostic tools and therapeutic solutions while translational research accelerates the transfer of knowledge from basic “bench-side” research into clinical “bedside” applications resulting in the adoption of best clinical practice for RD. These studies help enlighten solutions towards better standards of care and treatment and a higher quality of life for RD patients.

Patient registries are sets of data collected, stored, retrieved and disseminated in an organised, systemic manner [80]. They are important tools for: clinical planning and treatment strategies; public health surveillance; studying disease aetiology, the distribution (for example, incidence and prevalence) and determinants of disease; service planning, operation and evaluation; and diagnostic classification [80]. They are also important for clinical research, particularly since they can overcome some of the limitations of small numbers of cases of each RD in WA, primarily by increasing access to local, national or international clinical research trials and access to novel therapeutics. Further, research has shown that patient registries have the potential to provide significant return on investment [81].

WA Health has a strong interest in supporting the development and maintenance of patient registries for RD. This includes support for a national RD registry which would incorporate all RD, since strong calls have been made in the RD community regarding the need to develop such a registry. Registries primarily aim to facilitate the recruitment of Australians into international clinical trials. For example the Australian Neuromuscular Disorders Registry is an overarching database that currently includes disease-specific registries for conditions such as Duchenne muscular dystrophy (DMD), spinal muscular atrophy and myotonic dystrophy. The DMD Registry, launched in 2010, was the first registry that WA Health facilitated and was driven by patient support. It is linked with the TREAT-NMD (Translational Research in Europe for the Assessment and Treatment of Neuromuscular Disease) global network of national registries enabling registered patients to be included in international clinical trial enquiries.

Biobanks refer to organised collections of human biological samples and any related information stored for one or more purposes. These resources are maintained collaboratively by clinicians and researchers and are a valuable resource for RD research. They involve a delicate balance between health policy objectives, academic research, the public good and community trust in the benefits of biobanks and privacy protection. International cooperation is important for sharing limited numbers of samples from people with RD and reaching a critical mass of patients and samples [18].

In 2010, WA Health developed *Guidelines for human biobanks, genetic research databases and associated data* [82]. These guidelines provide principles and best practices for the establishment, governance, management and use of human biobanks, genetic research databases and associated data used for research purposes. These guidelines are due for review in 2015.

There are various reasons that clinical and translational research on RD is difficult, namely the: high number and variety of RD; lack of suitable experimental models for most RD; poorly defined endpoints; small number of patients; and, above all, limited resources [83]. Therefore in the field of rare disease research maximising scarce resources and coordinating research efforts is a necessity. There is a strong need to foster collaborative programs at local, national and international levels, since research, no matter where it is conducted, will ultimately benefit people living with RD in WA. In particular, collaboration in the performance of clinical trials is essential to reach a population size which provides sufficient statistical power to undertake studies [83].

WA Health is involved in a number of collaborations at a national and international level that are aimed at building capacity in RD research. One important global program is the International Rare Diseases Research Consortium (IRDiRC) which was launched in 2011. It is an initiative of the European Commission and the US National Institutes of Health. The aim is to foster international collaboration in RD research. The goal is to pool resources and work beyond borders to get a better understanding of RD and find adequate treatments [78].

Membership to IRDiRC extends beyond Europe and the US, including Australia represented by WA Health [18]. The IRDiRC links researchers and organisations investing in RD research. It has two main objectives: to deliver 200 new therapies for RD; and the means to diagnose most RD by 2020 [84].

Another international project that WA Health is a partner in is RD-Connect. This project aims to enable progress of the IRDiRC goals by developing an integrated platform that connects databases, registries, biobanks and clinical bioinformatics for RD research. It recognizes the need for global collaboration and harmonized infrastructure to make optimal use of resources for RD research.

Initiatives

1. Investigate mechanisms that support the translation of research to clinical care and treatment of RD in WA.
2. Investigate the priorities for clinical and translational research from the perspective of people living with RD, researchers and funders.
3. Investigate RD that have successfully attracted research funding in Australia (e.g. through a case study) and identify key success factors that are transferable to other RD.
4. Support the development of an Australian RD Registry.
5. Generate a list of existing patient registries, clinical trials and biobanks for RD patients in WA and make this information publicly available in Orphanet.
6. Develop a state policy on the development of registries by WA Health.
7. Support the development and integration of patient registries and biobanks for WA patients.
8. Review WA Health's *Guidelines for human biobanks, genetic research databases and associated data*.
9. Foster linkage of WA registries and biobanks with national and international registries and biobanks.
10. Contribute to local, national and international networks that promote research in RD (e.g. IRDiRC, RD-Connect).

References

1. Zurynski, Y., H. Leonard, and E. Elliott, *Rare childhood diseases: How should we respond?* Archives of Disease in Childhood, 2008. 93: p. 1071–1074.
2. Dawkins, H., et al., *Awakening Australia to rare diseases: Symposium report and preliminary outcomes.* Orphanet Journal of Rare Diseases, 2011. 6: p. 57.
3. European Commission. *Rare diseases – what are they?* 2013 5 July 2013 [cited 2013 7 July]; Available from: http://ec.europa.eu/health/rare_diseases/policy/index_en.htm.
4. Orphanet. *The portal for rare diseases and orphan drugs. About rare diseases.* 2012 [cited 2012 11 Jan]; Available from: <http://www.orpha.net/consor/cgi-bin/EducationAboutRareDiseases.php?lng=EN>.
5. Aymé, S. and C. Rodwell, *Report on the state of the art of rare disease activities in Europe of the European Union Committee of Experts on Rare Diseases – Part I: Overview of rare disease activities in Europe and key developments in 2010.* <http://www.eucerd.eu/upload/file/Reports/2011ReportStateofArtRDAactivities.pdf>, 2011.
6. Jaffe, A., et al., *Call for a national plan for rare diseases.* Journal of Paediatrics and Child Health, 2009. 46: p. 2–4.
7. European Organisation for Rare Diseases, *Rare diseases: understanding this public health priority.* 2005, EURORDIS.
8. Republique Francaise, *French national plan for rare diseases 2005–2008: Ensuring equity in the access to diagnosis, treatment and provision of care.* 2004.
9. Limb, L., S. Nutt, and A. Sen, *Experiences of Rare Diseases: An Insight from Patients and Families.* 2010, Rare Disease UK.
10. Shire Human Genetic Therapies, *Rare disease impact report: Insights from patients and the medical community 2013,* Shire.
11. Molster, C., et al., *Key outcomes from stakeholder workshops at a symposium to inform the development of an Australian national plan for rare diseases.* Orphanet Journal of Rare Disease, 2012. 7: p. 50.
12. Department of Health Western Australia, *WA chronic health conditions framework 2011–2016.* 2011, Department of Health Western Australia: Perth WA.
13. National Health Priority Action Council, *National chronic disease strategy.* 2006, Australian Government Department of Health and Ageing: Canberra ACT.
14. Rohn, J., *Billions spent on rare diseases.* Nature Biotechnology, 2013. 31(5): p. 368.
15. Dye, D., et al., *The impact of single gene and chromosomal disorders on hospital admissions of children and adolescents: a population-based study.* Public Health Genomics, 2011. 14(3): p. 153-161.
16. Dye, D., et al., *The impact of single gene and chromosomal disorders on hospital admissions in an adult population.* Journal of Community Genetics, 2011. 2: p. 81–90.
17. Department of Health Western Australia, *Working Together. WA Health strategic intent 2010–2015,* Department of Health Western Australia. 2010: Perth.
18. Department of Health Western Australia, *Scoping paper on the need for a national rare diseases plan for Australia.* 2013, Government of Western Australia: Perth.

19. National Ageing Research Institute, *What is person centred care? A literature review*. 2006.
20. National Health and Hospital Reform Commission, *A healthier future for all Australians final report 2009*, Commonwealth of Australia: Canberra ACT.
21. Australian Commission on Safety and Quality in Health Care, *Australian safety and quality framework for health care*. 2010, Australian Commission on Safety and Quality in Health Care: Sydney.
22. Consumer Focus Collaboration, *The evidence supporting consumer participation in health 2001*, Commonwealth of Australia: Canberra ACT.
23. Gregory, J., *Consumer engagement in Australian health policy: Final report of the AIHPS research project*. 2008: Melbourne VIC.
24. Health Consumers Queensland, *Consumer and community engagement: Evidence of improved individual health outcomes, services and systems*. 2012: Brisbane QLD.
25. Australian Commission on Safety and Quality in Health Care, *Patient-centred care: Improving quality and safety by focussing care on patients and consumers*. 2011, Australian Commission on Safety and Quality in Health Care: Sydney.
26. The King's Fund, *From vision to action – Making patient-centred care a reality*. 2012, The King's Fund: London.
27. The King's Fund, *Seeing the person in the patient*. 2008, The King's Fund: England.
28. Australian Commission on Safety and Quality in Health Care, *Patient-centred care: Improving quality and safety by focusing care on patients and consumers – Discussion paper*. 2010, Australian Commission on Safety and Quality in Health Care.: Sydney.
29. Bensberg, M., *Patient centred care – Literature review*. 2007, Dandenong Division of General Practice: Dandenong.
30. Department of Health Western Australia, *WA Health consumer carer and community engagement framework: For health services, hospitals and WA health following consultation across WA Health*. 2007, Department of Health Western Australia: Perth WA.
31. Health Consumers Queensland, *Consumer representative program: Consumer handbook*. 2009, Queensland Government: Brisbane QLD.
32. Queensland Health, *Disability Service Plan 2011–2014*. 2011.
33. NSW Health, *NSW Health Policy 2011–2016: Healthy bodies, healthy minds, vibrant futures*. 2010.
34. Queensland Health, *Queensland strategy for chronic disease 2005–2015*. 2005, Queensland Health: Brisbane QLD.
35. Alexander, K. and N. Hicks, *Sailing without radar: An excursion in resource allocation*. Australian Health Review, 1998. 2(2): p. 76–99.
36. Department of Health Western Australia, *WA chronic conditions self-management strategic framework*. 2011, Department of Health, Western Australia: Perth WA
37. Holman, H. and K. Lorig, *Patients as partners in managing chronic disease - partnership is a prerequisite for effective and efficient health care*. British Medical Journal, 2000. 320: p. 526–527.
38. Department of Health Western Australia, *WA cystic fibrosis model of care*. 2013, Health Networks Branch, Department of Health, Western Australia: Perth.

39. Centre for Research into Disability and Society, *Scoping document. Western Australian state wide chronic disease self-management strategy: Implementation and evaluation*. 2006, Curtin University of Technology: Perth WA.
40. Lambert, S., *Chronic condition self-management: A primary health care change management problem*. Australian Journal of Primary Health, 2005. 11(2): p. 70–77.
41. Williams, A., et al., *Sustaining chronic disease management in primary care: Lessons from a demonstration project*. Australian Journal of Primary Health, 2007. 13(2): p. 121–128.
42. South Australian Deptment of Health Statewide Service Strategy Division, *Chronic disease action plan for South Australia*. 2009.
43. Chodosh, J., et al., Meta-analysis: *Chronic disease self-management programs for older adults*. Annals of Internal Medicine, 2005. 143(6): p. 427–438.
44. Zwar, N., et al., *A systematic review of chronic disease management*. 2006: Sydney NSW.
45. Department of Health Western Australia, *Our children our future: A framework for child and youth health services in Western Australia 2008–2012*. 2008, Department of Health Western Australia: Perth WA.
46. Ouwens, M., et al., *Integrated care programmes for chronically ill patients: a review of systematic reviews*. Int J Qual Health Care, 2005. 17(2): p. 141–6.
47. Battista, R.N., et al., *Genetics in health care: an overview of current and emerging models*. Public Health Genomics, 2012. 15(1): p. 34–45.
48. Rajgopal, R., et al., *Cost-benefit analysis indicates the positive economic benefits of the expanded food and nutrition education program related to chronic disease prevention*. Journal of Nutrition Education & Behavior, 2002. 34(1): p. 26–37.
49. Bjornson, C.L. and I. Mitchell, *Congenital tracheoesophageal fistula and coordination of care: Expectations and realities*. Paediatr Child Health, 2006. 11(7): p. 395–9.
50. Bembi, B., et al., *Management and treatment of glycogenosis type II*. Neurology, 2008. 71(23 Suppl 2): p. S12–36.
51. Australian Health Ministers Advisory Council, *National framework for universal child and family health services*. 2011.
52. Leutz, W.N., *Five laws for integrating medical and social services: Lessons from the United States and the United Kingdom*. Milbank Quarterly, 1999. 77(1): p. 77.
53. Axelsson, R. and S.B. Axelsson, *Integration and collaboration in public health – A conceptual framework*. The International Journal of Health Planning and Management, 2006. 21(1): p. 75–88.
54. Hofmarcher M, Oxley H, and E. Rusticelli, *Improved health system performance through better care coordination*, in *OECD Health Working Papers*. 2007, OECD.
55. National Health Service UK, *Improving care for people with long term conditions: Information Sheet 1 – Personalised Care Planning*. 2011, National Health Service UK.
56. Russell, G., et al., *Beyond fighting fires and chasing tails? Chronic illness care plans in Ontario, Canada*. Ann Fam Med, 2008. 6(2): p. 146–53.
57. Goodwin, N., *Taking integrated care forward: the need for shared values*. Int J Integr Care, 2013. 13: p. e026.
58. Wissel, J., J. Olver, and K.S. Sunnerhagen, *Navigating the poststroke continuum of care*. J Stroke Cerebrovasc Dis, 2013. 22(1): p. 1–8.

59. European Union Committee of Experts on Rare Diseases (EUCERD), *EUCERD workshop report: National centres of expertise for rare diseases & European collaboration between centres of expertise*. 2011, EUCERD.
60. Šimerka, P., *Council Recommendation of 8 June 2009 on an action in the field of rare diseases, in 2009/C 151/02*, Official Journal of the European Union, Editor. p. 7–10.
61. Houyez, F. and S. Lynam, *5th European Conference on Rare Diseases*. EURORDIS, 2010.
62. Department of Health Western Australia, *Paediatric chronic diseases transition framework*. 2009, Health Networks Branch, Department of Health, Western Australia: Perth, Western Australia.
63. Department of Health Western Australia, *WA primary health care strategy*. 2011, Health Networks Branch, Department of Health, Western Australia: Perth.
64. Department of Health Western Australia, *Paediatric and adolescent palliative care model of care*. 2009, WA Cancer and Palliative Care Network, Department of Health, WA: Perth, Western Australia.
65. Council of Australian Governments (COAG), *National disability strategy 2010–2020*. 2010, <http://www.fahcsia.gov.au/our-responsibilities/disability-and-carers/publications-articles/policy-research/national-disability-strategy-2010-2020>.
66. Northern Territory Government, *Northern Territory chronic conditions prevention and management strategy 2010–2020*. 2009.
67. Department of Health and Human Services, *Connecting care: Chronic disease action framework for Tasmania 2009–2013*. 2009.
68. Department of Health Western Australia, *Model of care: Overview and guidelines*. 2007, WA Health Networks: Perth.
69. Victorian Department of Human Services, *National e-Health strategy*. 2008, Australia Health Ministers' Council: Melbourne VIC.
70. Department of Health and Ageing. *Telehealth 2012* 17 April 2012 [cited 2013 12 November]; Available from: <http://www.health.gov.au/internet/main/publishing.nsf/Content/e-health-telehealth>.
71. Carati C and Margelis G, *Towards a national strategy for telehealth in Australia 2013–2018*. 2013, Australasian Telehealth Society.
72. Allen, C., *Moving with the times – The impact of a metabolic telehealth clinic, in WA Health Conference and Awards 2013*. 2013: Perth WA.
73. Knight, A. and T. Senior, *The common problem of rare disease in general practice*. Medical Journal of Australia, 2006. 185: p. 82–83.
74. Zurynski, Y., K. Reeve, and E. Elliott, *International conferences on rare diseases: Initiatives in commitment, patient care and connections*. Medical Journal of Australia, 2007. 187(10): p. 597.
75. Le Cam, Y. *Patients' needs and expectations concerning access to health services – Eurordiscare 3 study, in 4th European Conference on Rare Diseases*. 2007.
76. Phillips, W., *Zebras on the common: Rare conditions in family practice*. Journal of the American Board of Family Practice, 2004. 17: p. 283-286.
77. Dan, B., *The paediatrician's role in support groups for rare diseases*. Acta Paediatrica, 2008. 97: p. 1510–1511.

78. de Vruh, R., E. Baekelandt, and J. de Haan, *Background paper 6.19 rare diseases: Update on 2004 background paper written by S. van Weely and H.G.M. Leufkens, in Priority medicines for Europe and the world: "A public health approach to innovation"*. 2013. p. 46.
79. Rare Disease UK, *Funding support for rare disease research: Raising awareness and increasing transparency*. 2010, Rare Diseases UK London
80. Richesson, R. and K. Vehik, *Patient registries: Utility, validity and inference*. *Advances in Experimental Medicine & Biology*, 2010. 686: p. 87–104.
81. Evans, S., et al., *Development of clinical-quality registries in Australia: The way forward*. *Medical Journal of Australia* 2011. 194(7): p. 360–363.
82. Office of Population Health Genomics, *Guidelines for human biobanks, genetic research databases and associated data*. 2010, Department of Health WA: Perth.
83. Europlan. *Recommendations for the development of national plans for rare diseases – Guidance document*. 2010 10 Jan 2012]; Available from: [http://www.europlanproject.eu/public/contenuti/files/Guidance_Doc EUROPLAN 20100601_final.pdf?bcsi_scan_2C647EB3599034DE=0&bcsi_scan_filename=Guidance_Doc EUROPLAN 20100601_final.pdf](http://www.europlanproject.eu/public/contenuti/files/Guidance_Doc_EUROPLAN_20100601_final.pdf?bcsi_scan_2C647EB3599034DE=0&bcsi_scan_filename=Guidance_Doc_EUROPLAN_20100601_final.pdf).
84. IRDiRC. *Goals*. 2013; Available from: http://www.irdirc.org/?page_id=9.



This document can be made available in alternative formats on request for a person with a disability.

Produced by the Office of Population Health Genomics
© Department of Health 2015

Copyright to this material is vested in the State of Western Australia unless otherwise indicated. Apart from any fair dealing for the purposes of private study, research, criticism or review, as permitted under the provisions of the *Copyright Act 1968*, no part may be reproduced or re-used for any purposes whatsoever without written permission of the State of Western Australia.