Australian Government

Department of Health

National Strategic Action Plan for Rare Diseases

February 2020

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

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Foreword

Nicole Millis
Chief Executive Officer – Rare Voices Australia

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

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

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

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

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

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

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

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

Nicole Millis

rare voices
AUSTRALIA

Executive Summary

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

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

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

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

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

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

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

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

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

Plan on a Page

VISION

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

CRITICAL ENABLERS

Multi-stakeholder involvement and engagement

Collaborative governance and leadership

State, national and international partnerships

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

PILLAR 1: AWARENESS AND EDUCATION

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

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

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

PILLAR 2: CARE AND SUPPORT

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

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

Priority 2.3: Facilitate increased reproductive confidence.

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

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

PILLAR 3: RESEARCH AND DATA

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

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

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

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

THESE PRINCIPLES ARE THE FOUNDATION FOR THIS ACTION PLAN

Person-centred

Equity of access

Sustainable systems and workforce

Contents

About the Action Plan

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

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

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

Introduction

What is a rare disease?

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

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

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

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

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

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

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

Rare diseases: the international approach

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

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

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

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

Rare diseases: the Australian context

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

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

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

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

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

Priority Populations

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

Australians living with a rare disease

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

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

Australians living with an undiagnosed rare disease

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

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

This population can be considered in three categories:

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

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

Aboriginal and Torres Strait Islander people

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

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

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

People living in regional, rural and remote areas

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

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

People from CALD backgrounds

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

People experiencing socio-economic disadvantage

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

Partnerships

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

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

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

Pillar 1: Awareness and Education

Why is this important?

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

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

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

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

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

What we hope to achieve:

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

Priority 1.1

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

Action 1.1.1

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

Implementation

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

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

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

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

Action 1.1.2

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

Implementation

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

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

Action 1.1.3

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

Implementation

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

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

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

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

Priority 1.2

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

Action 1.2.1

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

Implementation

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

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

Action 1.2.2

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

Implementation

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

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

Priority 1.3

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

Action 1.3.1

Develop a national rare disease workforce strategy.

Implementation

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

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

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

Action 1.3.2

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

Implementation

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

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

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

Pillar 2: Care and Support

Why is this important?

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

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

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

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

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

What we hope to achieve:

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

Priority 2.1

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

Action 2.1.1

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

Implementation

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

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

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

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

Action 2.1.2

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

Implementation

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

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

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

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

Action 2.1.3

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

Implementation

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

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

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

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

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

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

Action 2.1.4

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

Implementation

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

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

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

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

Action 2.1.5

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

Implementation

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

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

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

Priority 2.2

Ensure diagnosis of a rare disease is timely and accurate.

Action 2.2.1

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

Implementation

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

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

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

Action 2.2.2

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

Implementation

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

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

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

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

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

Action 2.2.3

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

Implementation

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

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

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

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

Action 2.2.4

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

Implementation

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

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

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

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

Priority 2.3

Facilitate increased reproductive confidence.

Action 2.3.1

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

Implementation

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

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

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

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

Action 2.3.2

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

Implementation

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

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

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

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

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

Priority 2.4

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

Action 2.4.1

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

Implementation

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

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

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

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

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

Action 2.4.2

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

Implementation

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

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

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

Action 2.4.3

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

Implementation

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

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

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

Priority 2.5

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

Action 2.5.1

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

Implementation

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

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

Action 2.5.2

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

Implementation

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

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

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

Action 2.5.3

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

Implementation

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

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

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

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

Pillar 3: Research and Data

Why is this important?

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

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

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

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

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

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

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

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

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

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

What we hope to achieve:

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

Priority 3.1

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

Action 3.1.1

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

Implementation

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

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

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

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

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

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

Action 3.1.2

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

Implementation

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

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

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

Action 3.1.3

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

Implementation

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

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

Action 3.1.4

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

Implementation

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

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

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

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

Priority 3.2

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

Action 3.2.1

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

Implementation

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

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

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

Action 3.2.2

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

Implementation

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

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

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

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

Action 3.2.3

Support collaborative research into rare diseases in Australia and internationally.

Implementation

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

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

Action 3.2.4

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

Implementation

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

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

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

Action 3.2.5

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

Implementation

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

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

Priority 3.3

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

Action 3.3.1

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

Implementation

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

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

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

Action 3.3.2

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

Implementation

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

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

Priority 3.4

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

Action 3.4.1

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

Implementation

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

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

Action 3.4.2

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

Implementation

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

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

Achieving Progress

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

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

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

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

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

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

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

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

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

Theme #4
The need to measure rare diseases

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

Theme #5
The need for sustainable systems and workforce

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

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

Theme #6
The need for stakeholder collaboration

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

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

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

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

Theme #8
The need to progress early implementation wherever possible

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

Implementation Mechanisms

International Exemplars

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

The Genetic and Rare Diseases (GARD) Information Center

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

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

Rare Disease Centres of Excellence

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

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

European Reference Networks (ERNs)

ERNs have achieved global recognition in facilitating the delivery of expert care, despite geographical boundaries and distances. Virtual ERNs are comprised of health care professionals and are spread throughout Europe. The ERNs' objective is to tackle complex or rare diseases and conditions that require highly specialised treatment and a concentration of knowledge and resources. ERNs facilitate the exchange of knowledge between health care professionals across borders, giving people access to the expert knowledge they need even if it's not available in their own country or region[101].

Customisation for the Australian context

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

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

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

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

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

Appendix 1

Glossary

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Appendix 2

Acknowledgments

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Appendix 3

Working Towards Rare Disease Care and Support that is Integrated

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

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

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

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

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

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

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

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

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Appendix 4

Health Technology Assessment

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

What is Health Technology Assessment (HTA)?

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

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

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

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

Acronyms

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

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