

Email from Chris Higgins (CE RDNZ) to [Ministry of Health Principal Policy Analyst working on the Rare Disorders Strategy] 19 October 2023

Kia ora [Principal Policy Analyst working on the Rare Disorder's Strategy]

Thanks again for sharing the outline document. While we think the overall structure works satisfactorily for the most part, we have suggested some points for consideration for the content as attached, which were identified at a meeting between [the RDNZ team]. The comments represent the outcomes of our meeting, and I've added some elaboration on a couple of points.

Our one concern with the structure of the outline is that there's no section (or sections for each of the priority areas) which sets out next steps/actions/recommendations which will create expectations and/or requirements for other health entities such as Te Whatu Ora, Pharmac and others. Other Pae Ora strategies include lists of what things will look like in the future and what needs to change, turning strategies into action and monitoring outcomes. It will be good to see similar headings in the RDS, and this is where many of us will expect to see our input and submissions reflected.

[Chief Clinical Advisor Ministry of Health] shared the evidence brief with those who'll be invited to participate in the clinician/expert consultation sessions, including ourselves. Although there's a couple of points that we'll share with him directly, overall this is an excellent document, and we think that the outline could have drawn from it more extensively and directly than it has.

We'll look forward to seeing the next iteration.

Ngā mihi

Chris

Chris Higgins
Chief Executive
Rare Disorders NZ

Draft outline for an Aotearoa New Zealand rare disorders strategy

We are interested in your feedback on any points in this draft strategy outline.

This outline represents current thinking from development to date of a rare disorders strategy for Aotearoa New Zealand. Further input and ideas are welcomed as we work towards completing a draft strategy for the Minister of Health before the end of 2023.

The outline has been developed by a Manatū Hauora | Ministry of Health team working closely with Rare Disorders New Zealand and Te Aka Whai Ora | the Māori Health Authority. It draws on input from:

- the rare disorders community, in particular from support group leads from the many organisations that support people and whānau with rare disorders*
- whānau Māori living with rare disorders*
- a reference group that includes a wide range of clinical, academic and health service delivery advisors and experts*
- staff of New Zealand agencies with a key role to play, such as Te Whatu Ora | Health New Zealand, Pharmac, the Health Quality and Safety Commission and Whaikaha and the Health and Disability Commissioner's office*
- international expert informants.*

This outline also draws from:

- evidence and insights from New Zealand and international research, analysis and observation*
- input provided in the development of other strategies, policies and reports as diverse as, and including, the Pae Ora strategies, the Long Term Insights Briefing on Precision Health and the Waitangi Tribunal Health Services and Outcomes Inquiry (Wai 2575)*
- discussions with health officials in other countries and international organisations.*

Introduction

We aim for a health system that enables all New Zealanders to achieve pae ora, to live healthy lives and participate in their communities.

One substantial group of New Zealanders who face additional difficulties in achieving pae ora are people and whānau living with rare disorders. They too often struggle to get the support they need from the health system, even though rare disorders have major impacts on people's lives and come with very high health needs. Not only are the disorders rare – ability to provide effective care is also rare.

Effective care being rare is not only bad for people with rare disorders, it results in major waste of health care efforts, dissatisfaction for those working in the system and strain on health (especially hospital) resources. Keeping people with rare disorders healthy and out of hospital has clear benefits all round.

Now, global knowledge on rare disorders is growing fast. Our health system is becoming more coordinated and aligned. Tools and technology to support knowledge transfer and enable better care are being built. Ways to make rare treatments more affordable are being investigated.

There are opportunities now to improve how healthcare is arranged and delivered so that rare will no longer be a barrier to effective health care. Now we can work towards a health system where:

- people and whānau with rare disorders can have better health and live better lives
- people working in the health system can deliver better care for all patients
- all New Zealanders can be confident that the health system is working for them, no matter what their health need is or how rare it is.

What is a rare disorder?

A rare disorder is a medical condition with a specific pattern of clinical signs, symptoms and findings that affects fewer than or equal to 1 in 2000 persons in Aotearoa New Zealand.

Rare disorders include, but are not limited to, rare genetic disorders, rare cancers, rare infectious disorders, rare poisonings, rare immune-related disorders, rare idiopathic disorders, and rare undetermined conditions.

An ultra-rare disorder is one that affects fewer than or equal to 1 in 50,000 persons in Aotearoa New Zealand.

Commented [I1]: There should be explicit acknowledgment that people with rare disorders comprise a "priority" or "vulnerable" population, leading to considering the needs of the rare disorder community in the context of other areas of national focus (e.g., mental health, chronic diseases, transitional care, Māori health)

What do we know about rare disorders in New Zealand

Summary of knowledge, sources and knowledge gaps, potential sources

Assumptions based on overseas information

Voices of people and whānau living with rare disorders

Māori whānau living with rare disorders

Insert
(current
Healthify
wording,
may need
to revise)

[Key points about rare disorders](https://healthify.nz/health-a-z/r/rare-disorders/) – <https://healthify.nz/health-a-z/r/rare-disorders/>

- A rare disorder is a health condition that affects very few people. However, about 300,000 New Zealanders have a rare disorder because there are many different types.
- They can be hard to diagnose but most rare disorders are genetic and can affect other family/whānau members.
- Each disorder has a different set of signs and symptoms and unless the disorder has been well researched, eg, Huntingtons or cystic fibrosis, there may be a lack of understanding about how the disorder will progress.
- Many rare disorders are life-long and debilitating. About half of them affect children and 30% of those are terminal diseases.
- Support is available from Rare Disorders NZ, genetic services, disability services, and disability and rare disorder support groups.

Health system experiences of people with rare disorders

Many people have good experiences with the New Zealand health system, of life-saving care, prevention and early intervention.

However, we hear from too many people that they have struggled to have their or their children’s health or developmental needs recognised. They may not get the assessment, tests or expert input that would lead to a diagnosis of a rare disorder – people say this often takes many years, and can mean the condition gets worse before preventive care can be started.

Even when people have a diagnosis, they may struggle to get information on what can help. There may not be people with experience or expertise in their condition in their region or even in the country. Where there is a treatment, or specialist equipment, it may be hard to source, unavailable or unaffordable.

...

These experiences are very similar to those described in countries around the world ... other strategies and plans ... UN resolution ...

Commented [I2]: In this section we would like to see more of a whole of life approach and reflection of the WHO definition of health. Health is more than diagnosis and treatment.

Commented [CH3R2]: WHO: Health is a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity.

Commented [I4]: Noting NZ is a signatory to this

International experience with improving rare disorders outcomes and experiences

From evidence brief

Approaches adopted to improve rare outcomes and experiences include programmes to:

- continuously improve understanding and knowledge of rare disorders, their effects and how people can be supported to live well
- make it easy for health practitioners, people and communities to include and respond well to people and whānau living with a rare disorder
- build on collective working with rare disorders communities
- learn from and contribute to international advances for people and whānau living with rare disorders

Strategy goals

The strategy will be successful if the lives of people and whānau living with a rare disorder improve, and if there is corresponding improvement in how the health system responds and relates to people whose needs or situation are uncommon, unusual or unclear.

Goals:

- People living with rare disorders and their whānau achieve pae ora – healthy futures
- Māori, Pacific peoples and disabled people living with rare disorders and those living in rural and other diverse communities achieve pae ora – healthy futures
- The health workforce is able to deliver high-quality care for people and whānau living with rare disorders
- The New Zealand health system works fairly to promote health and address health needs for all, including for those with rare or unknown disorders.

People and whānau living with rare disorders

All people deserve to be respected, listened to, heard and supported when they seek health care. People and whānau living with a rare disorder need these things even more - their situation or needs are not usual and may often be unclear.

Commented [I5]: People with a rare disorder achieve equity within the health system and see an improvement in their and their whānau's quality of life.

Commented [CH6R5]: Mere improvement can probably be achieved without a specific strategy. The RDS should be aiming for something significantly more ambitious.

Commented [I7]: We would like to see what this means expanded on. For instance this will look like early and accurate diagnosis planned pathways for clinical care, equitable access to modern medicines, a coordinated and funded program of research and workforce development etc.

Commented [I8]: We would like the strategy to acknowledge that the wellbeing needs of people living rare disorders are not confined to the health sector, and that while the scope of the strategy will not be extended to other sectors including disability/whaikaha, education, justice, social welfare etc, there is important work to be done here too.

Commented [I9]: Can we define what pae ora is for people with rare disorders- perhaps along the lines of above "people and whānau with rare disorders can have better health and live better lives", in the SGL feedback people were not happy with the term pae ora or healthy futures as for many that is not attainable.

A child, for example, who is disabled from an early age as a result of a life shortening rare disorder will not necessarily be looking forward to a healthy future, and neither will their family. The UN resolution refers to achievement of optimal potential development and the right to the highest attainable standard of physical and mental health, and RDNZ recommends that pae ora be articulated in these and similar terms

This strategy outlines how the health system, health practitioners and others can work so that all people can feel heard, respected and supported. In this way, people can live better even while waiting for further assistance, follow-up or tests. Their wait for these may be shortened. Whether or not a rare disorder is identified, advice and support can help with living well and supporting child development.

People living with rare disorders have much to contribute to their communities' life, economy and wellbeing. By reducing negative impacts that rare disorders can have, their contributions are enabled and enhanced.

Health practitioners and workers

...

Health system

...

Principles that underpin the strategy

- Stands on the shoulders of the Pae Ora strategies, the New Zealand Disability Strategy and the Child and Youth Wellbeing Strategy
- Upholds te Tiriti o Waitangi, and works towards achieving equity for Māori
- Gives voice to people and whānau with rare disorders
- Supports health practitioners and providers to provide quality care
- Is informed by and seeks out evidence
- Is collaborative and built on partnerships
- Supports pae ora for all while focusing on the health system challenges that come with rarity

More about these principles and what they mean

Commented [I10]: should include a reference to and acknowledgement of the specific role of Rare Disorders New Zealand and rare disorders support groups in: supporting people with rare disorders to navigate their way through health, education, justice and other social support systems; providing information and advice to people with rare disorders, their whanau/family, their health professionals, other cross-sectional professionals, and cross-sectional policy makers; and facilitating implementation of the strategy upon its completion

Commented [I11]: Stands on the shoulders of the pharmac review also.

We would like to see what quality care actually means expanded

Who are the collaborations and partnerships proposed to be with?

Priorities

1. Putting people and whānau at the heart
2. Equipping the health workforce to support pae ora
3. Gearing the system for good and timely care
4. Learning and sustaining progress
5. Joining up internationally to achieve more

Priority 1. Putting people and whānau at the heart

What will it look like?

When people and whānau are at the heart of health interactions, attention is directed to what they say is most important. Services adapt to their cultures and circumstances, preferences and choices. A holistic view of wellbeing is the starting point and health and wellbeing are supported even when there is no diagnosis or treatment.

People have their own health records. Their expertise and knowledge is valued and informs the care that is suggested. They can bring in other expertise and decide on their care. What is most important to them drives the priorities of health providers. Whānau needs are met so that they can nurture whānau members with or without a rare disorder.

New Zealand Health Strategy ten-year ambitions:

- People, whānau and communities will have greater control and influence over their health and the services they need
- People have the best possible experience of health services and the health system
- Health services will be flexible to people's range of needs and their cultural expectations

What do we hear now?

More about people and diverse needs ... Equitable outcomes ... Services close to home .. support for rural communities to access services at a distance

Commented [I12]: We are concerned this may push too much responsibility onto individuals and their whānau, we agree their voice is important but they also need guidance that comes from reference to standards of care and clinical pathways

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Having people and whānau at the heart of health interactions and care relationships is very important. Equally important is that the voices of people and whānau are heard in policy, design, commissioning, service delivery, progress and outcome monitoring, research and improvement programmes at all levels. Accommodating rare disorders in how the system is set up will mean people can be met with a level of understanding as the starting point when interacting with different parts of the system.

To draw out in examples:

- Governance of data, genetic samples – Maui Hudson work
- Pharmac rare disorders committee
- Disability national identifier

Fit with how the health system is changing – eg, better links between primary and secondary care

Priority 2. Equipping the health workforce to support pae ora

Health practitioners and health and care service workers, overwhelmingly, are motivated to help people and to do excellent work. When they are alert to the possibility of a rare disorder and can easily get information, guidance and help, practitioners and others can make a sizeable difference to the experiences of people and whānau living with rare or unknown disorders.

Being alert to the possibility of a rare disorder is important for health practitioners – rare disorder examples incorporated into much of their learning can help, especially when integrated with broader learning along with diverse consumer preferences and needs. Education and care and support workers can also benefit from such integrated learning about rare disorders.

Easy, automated and on-line access to guidance and tools for decision support, referral pathways or clinical care help enormously when busy practitioners and service workers care for people with needs outside their own experience. Expert help and advice is important backup for the care providers who are accessible to people where they live. This expert advice is not only from specialists in a rare disorder (including medical, nursing, allied health, care and support), but also practitioners such as allied health and other therapists who can translate general wellbeing guidance to fit the lives and needs of people and whānau living with rare and unknown disorders. This fit includes cultural, practical and community-based elements.

New Zealand Health Strategy ten-year ambitions:

- The workforce will reflect our diverse communities and have the skills and capabilities required to meet their needs
- The workforce will feel valued, recognised and respected and will be supported and motivated to deliver high-quality care
- Flexible learning and working environments will give more room for growth and development

Working with diverse communities to build, maintain and enable strong foundations for health and wellbeing, and for including all community members, is another skill area that can be grown. Communities can support people and whānau in everyday activities and connections that promote their wellbeing.

To draw out in examples:

- Metabolic disorders, neuromuscular disorders, congenital anomalies registries
- Cystic fibrosis care standards
- Health pathways
- Kidz First, South Auckland

Priority 3. Gearing the system for good and timely care

While learning, tools and back-up can help practitioners and others to provide quality care, it is not enough. The wider health system needs to be geared up to provide good, timely, safe and life-affirming care for people and whānau of all needs, all stages of a health journey, and all including the rare and ultra-rare conditions.

All elements of the health system, and the system as a whole, need to be able to respond to rare needs. The elements need to be connected up so that people can move through it easily, being connected to the next services that can help. Information and feedback needs to flow and be easily accessible. Pathways for practitioners (such as referral and advice) and for care coordinators, whānau and patients (such as navigation) need to be well signalled and supported. While some elements exist, many are approximate, present in some places or for some age groups, or vulnerable to blockages elsewhere (like availability of tests or certain expertise).

New Zealand Health Strategy ten-year ambitions:

- The health system will develop standards for high-quality care that support all services to improve
- The health system will be more productive and efficient, to make the best use of public money
- The health system will lead and influence across government to improve health and wellbeing outcomes

Having the information and data on which decisions can be made is one vital element that, for rare disorders, is often disconnected or missing from our system. Our current medical condition data capture is based on older classifications, and plans for adopting newer ones need to allow also for continual updating of world knowledge about rare disorders. Other data of equal importance, like disability information, is in the planning stages.

Commented [I13]: Reference the July 2023 Australian recommendations for a national approach to rare disorders data. Also currency HIRA work in NZ

Ensuring the right governance, protections, access and ownership of system data is of foundational importance, especially for Māori. Rare voices, along with other consumer, cultural and minority voices, need to be at the table where all such system foundations are discussed and decided. Participation and a degree of choice will be needed for citizens to be confident in in-built protections.

Equally foundational are the relative priorities given to different parts of the system and how investment is decided across these. Prioritisation across products, services and infrastructure needs to account for all, including rare, needs and for lifelong needs alongside emergency and acute needs.

To draw out in examples:

- Auckland biobank
- Child cancer network and Children’s Oncology Group
- Genomics Aotearoa research alliance
- Efficiencies from Pharmac device purchasing eg dressings for EB

Priority 4. Learning and sustaining progress

Not only does the system need to be geared towards good and timely care for people with rare and unknown, or medically non-defined, disorders. It needs to learn about, track progress in and spread knowledge about people’s health and the care and outcomes people are

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experiencing. This is needed for rare disorders even more than for others as there is such a deficit of information now.

Becoming a learning system means we need people affected by rare disorders to be part of discussions about service design, delivery and outcomes tracking. We need indicators of system performance for rare disorders, and consideration of where opportunities for improvement lie. Given the close links with the disability community, rare disorders indicators are needed in disability indicators as well as health indicators.

We are unique in being able to gather knowledge of pae ora for Māori living with rare disorders, and to contribute to Pacific knowledge. Collaborative research and service design and monitoring with Māori, Pacific and other rare disorder communities is a key opportunity.

New Zealand Health Strategy ten-year ambitions:

- New initiatives will help all parts of the system to harness innovation and put evidence and research into everyday practice
- New national functions will drive continuous improvement and support the development and implementation of new technologies
- Health services will be flexible to people's range of needs and their cultural expectations

New Zealand research into service models and approaches for rare disorders and support for NZ participation in clinical trials...

To draw out in examples:

- CRISPR trial for HAE
- Orion AI partnership
- National clinical trials ecosystem

Commented [I14]: We could give more research examples if you want them e.g.

- Professor Karen McBride-Henry and Dr Tara Officer, Research Trust of Victoria University of Wellington recently HRC funded research project "Invisible inequity: Healthcare insights from people with rare disorders"

Priority 5. Joining up internationally to achieve more

New Zealand is a small country and cannot hold expert knowledge of every rare disorder alone. Considerable attention to rare disorders is building internationally. We can learn much from, and contribute to, collective efforts to improve outcomes for people and whānau living with rare disorders.

Rare disorders programmes, networks, centres and other efforts have existed for some years, and many jurisdictions now have rare disorders strategies, plans or other coordinated

approaches. While these vary, and evidence of which approaches work best is not yet clear, there is much that we can learn.

An evaluation is currently underway, for example, of the European Union' European Reference Networks, virtual networks involving healthcare providers across Europe. They aim to facilitate discussion on complex or rare diseases and conditions that require highly specialised treatment, and concentrated knowledge and resources.

Other models that have been adopted include ...

Guidelines, decision trees, referral and care pathways and similar tools have been promulgated in countries like England, ... They may be based on, and/or themselves generate, evidence of good outcomes for people with rare disorders. Adapting such tools for New Zealand may be an effective early approach to inform better and more widely accessible care here. Rare disorders community voices, along with Māori, Pacific, rural and other voices, will be needed in testing quality and suitability for our context and service users.

Commented [I15]: Suggest covering Perth's rare care centre

New Zealand Health Strategy ten-year ambition:

- Health entities will partner with other sectors to support shared approaches to improving health and wellbeing

We can contribute as well as learn from others. Joining in international collaborations, regional, global and local with our near neighbouring countries, ...

To draw out in examples:

- Orphan & high cost medicines funding
- Orphanet
- Virtual networks

Commented [I16]: It would be helpful for the document to more clearly express what the strategy will and won't do, and to identify which agencies will be accountable for addressing the RDS's implementation, and which agencies will be expected to address anything that the RDS states that it won't do

Commented [I17]: Pharmac review

Primary health care review commission), (Manatū Hauora have been instructed to carry out a primary health care review, this creates an opportunity for future primary health care delivery to have a more focussed approach to delivering services to people with rare disorders. We specifically recommend that the RDS include a comment to the effect that the forthcoming primary health care review will include a focus on rare disorders),

New Zealand Health Research Strategy 2017-2027.

The body of health information work being undertaken by Hira , which encompasses the SNOMED CT National Release Centre

Precision health,

How this strategy fits with the health system and other strategies

Brief description of overall system settings and instruments (consumer charter, pae ora strategies, government policy statement, health plan, iwi partnership boards, focus on equity, disability settings, ...)

How together they are leading to improvements like more patient and whānau centric services, increasing cultural capability, more consistent access to services across the country, ...

Pictorial to come

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