Email from Chris Higgins (CE RDNZ) to Ministry of Health Principal Policy Analyst sent 7 December 2023

Kia ora [Principal Policy Analyst MOH]

Thanks again for the extension of time for RDNZ's feedback which is attached by way of a covering letter from the RDNZ Board and detailed commentary on, plus proposed amendments to, the draft strategy itself. In both documents we continue to strongly recommend that a rare and undiagnosed disorders centre of expertise be provided for in the strategy, and the latest iteration of our well researched and widely supported proposal is therefore also attached.

Ngā mihi

Chris

Chris Higgins
Chief Executive
Rare Disorders NZ

[Name]
Principal Policy Analyst, System Enablers
Strategy, Policy and Legislation
Manatū Hauora, 133 Molesworth Street
Thorndon, Wellington 6011

7th December 2023

Dear [Principal Policy Analyst]

Rare Disorders Strategy (RDS) feedback

Thanks for the opportunity to comment on the most recent 27th November 2023 draft of the RDS, and for the timeframe extension which has enabled us to include feedback from the RDNZ Board as discussed at its formal meeting today. This letter therefore should be understood as the RDNZ Board's formal response to the draft RDS.

Headline points that the Board wishes to emphasise include:

1. There are many aspects of the draft which represent a significant advancement on the status quo which has prevailed for the last two decades and more, and the Board is pleased with the progress that is being made.

2. The meanings of pae ora and equity for people with rare disorders should be more clearly articulated, and the vision for the RDS should be more clearly expressed as people with rare disorders living as well as possible for as long as possible

3. There are acknowledged gaps in the draft RDS and for this reason we are asking that we are given a further opportunity to review the final and complete draft.

4. RDNZ has previously submitted that the RDS should include a statement to the effect that people with rare disorders will have access to the medicines they need in a way that reflects or exceeds international best practice. This has not been included in successive drafts and we think it would be extraordinary were this not to appear in the final document given that production of the RDS was one of the recommendations of the 2022 Pharmac Review.

5. We continue to recommend the provisional Health of Disabled People Strategy as a useful model for the RDS, particularly its specificity. Manatū Hauora has indicated that our previously submitted four "essential elements" for inclusion in the RDS are too specific for a strategy document, yet the draft RDS includes a number of statements which are more specific than RDNZ's recommendations. RDNZ continues to submit therefore that the RDS should firmly signal both the need for a rare and undiagnosed disorders centre of expertise, and the importance of including RDNZ as a co-designing partner and collaborator in the

RDS's implementation. The RDS content relating to data is close enough to RDNZ's earlier recommendations to be satisfactory, albeit not ideal.

6. There has been effective use of quotes sourced from many of those who have engaged in the RDS consultation sessions, with the exception of those who engaged in the clinical consultation exercises. Inclusion of quotes from this group will enhance the document.

Pleased see attached separately our detailed feedback on the draft which comprise tracked changes and comments on the 27th November draft.

Yours sincerely

James McGoram
Chairperson
Rare Disorders New Zealand

Chris Higgins
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Draft

Aotearoa New Zealand rare disorders strategy

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. People and whānau living with rare disorders have the potential to be part of pae ora in their communities but often face many difficulties.

People with rare disorders have high health needs

Rare disorders have major impacts on people's lives and can come with very high health needs – for some among the highest health needs of all. Added to these direct health effects are the significant extra burden, social isolation and economic hardship that people, whānau and carers often face. Effective health care for people and whāanau living with rare disorders can make a positive difference for life beyond addressing their health needs.

"Yeah, like we had some people come in and going, but nobody knew, you know, like it was all up in the air. So, we could try this, or we could do this, or you could see this person and then they didn't know. So, we were still left once seeing someone, we were still left without any information. Basically, just here, give this a go … I don't know six to 10 times, maybe to the doctors and nobody listened to me…Look at [tamariki] like just someone needs to do tests or something. Like it's just not right, you know? So, we fought. We fought for a good year and that that was the stressful time."

- *Māori whānau on accessing services for tamariki living with a rare disorder*

It can be difficult to find and provide effective health care for rare disorders

Even with these very high health needs, people living with rare disorders too often struggle to get the support they need from the health system. Not only are the disorders rare – the ability to provide effective care may be rare as well. No health professional, or even group of health professionals, can be across the wide breadth of rare disorders. In some cases, people and whanau living with rare disorders know a lot more about their rare disorder than the health professionals they encounter.

Lack of effective care 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. We also miss out on the value people living with rare disorders bring to our society. Keeping people with rare disorders as healthy as possible and out of hospital has clear benefits all round.

Commented [l1]: "part of pae ora" needs expansion to clarify what pae ora means. "Healthy futures" is vague. It already briefly states what pae ora means in the first sentence, so its use in the second is potentially confusing.
prefer "With the support of the health, education and social systems in Aotearoa, people and whānau living with rare disorders are able to lead as healthy and fulfilling lives as possible, but face many difficulties" or could use wording from the UN resolution which refers to achievement of "optimal potential development and the right to the highest attainable standard of physical and mental health"

It should also be noted that the achievement of pae ora expressed in these terms is an investment as people with rare disorders who receive appropriate and timely clinical interventions will have a better chance of being able to productively contribute to the country's economic well being. Newborn screening and timely therapies for conditions such as SMA and infantile onset Pompe disease, funding of Trikafta for cystic fibrosis, and education strategies for parents and teachers of fragile X childrenare all examples of how people can have opportunities to enjoy productive lives as a direct result of appropriate interventions.

We would like to see it said outright that the strategy is about achieving equity in health outcomes by eliminating barriers for people with rare disorders, their whānau, and for Māori

Perhaps include a definition of what pae ora and equity means for people living with rare disorders up front?

Commented [l2]: People with rare disorders despite the variable health care system, do bring value to our society. Suggest that this sentence is changed to something like: "*We do not unlock the full potential value people living with rare disorders can bring to our society.*"

Rare disorders and unknown disorders

Until a diagnosis can be arrived at, people with unclear or unusual health needs may live some time with significant effects but no knowledge of the cause. In order that people with rare disorders can receive more timely diagnosis and care, improvements are needed for a wider group of people whose health problems are, presently, unknown.

Commented [I3]: The important issue here is that delay in diagnosis where effective treatment is available may result in higher long term costs for the individual and the health system. E.g. SMA and nusinersen. (Rosemary)

Improvements

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
- people and whānau are enabled to be involved and promote their health
- 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.

What do the words mean? disease, disorder, condition, syndrome …

In December 2021, the United Nations adopted the first-ever Resolution on "Addressing the Challenges of Persons Living with a Rare Disease and their Families".

Aotearoa New Zealand definition is adopted from United Nations definition for rare disorders and aligns with the assumed number of people living with a rare disorder in many countries.

Globally, rare 'disease' and 'disorder' are often used interchangeably. We use disorder in Aotearoa New Zealand to recognise that undiagnosed disorders may fall under rare with continuous improvements in diagnostic tools and technology and with improvements in the knowledge base of rare disorders among health professionals.

A **disorder** might indicate that a specific disease is possible but there is not enough clinical evidence for diagnosis.

A **disease** is a medical condition that has a clear cause and can be diagnosed by a healthcare provider.

A **condition** is an abnormal state of health that interferes with the usual activities or feeling of wellbeing.

A **syndrome** is a certain group of symptoms that occur together which can help a health professional to identify a condition as a disorder or a disease.

Commented [I4]: There are many different definitions - Suggest:
A **syndrome** is a ~~certain~~ group of *signs and* symptoms that occur together which can ~~help a health professional to identify a condition as~~ *indicate a specific* disorder or a disease.
Addition of signs is important here e.g. people with Down Syndrome know that they look different but would not see this as having symptoms. Inclusive language is important.

What do we know about rare disorders in New Zealand

People affected by rare disorders

We currently do not have good information to know the effect of rare disorders on people and the health system in Aotearoa New Zealand. Our understanding of rare disorders mostly comes from international findings.

Based on overseas studies, we assume that around 300,000 people in New Zealand have rare disorders - many of these are children. Around 80% of rare disorders are genetic and, of these, some are inherited and others not. Rare disorders can occur in any family or population group in our country.

Commented [I5]: The subtleties of this are complex and people may not understand what this means. Suggest stop at genetic.

Over 7,000 different rare disorders have been identified. A person may be the only one in New Zealand with their rare disorder, or there may be up to ~~250~~ others spread across the country and across age groups. For most people, it will be somewhere in between. There will often be other New Zealanders with the same rare disorder, but not often in the same age group or living in the same region.

Commented [CH6]: 2500?

Commented [I7R6]: 1/2000 of 5.123 million is 2,562

Rare Disorders New Zealand sponsors a biennial, national 'voice of rare disorders survey' to collect information about people and whānau living with rare disorders in Aotearoa New Zealand. This survey provides a good snapshot of the New Zealand context and points to the impacts rare disorders have for people and their whānau.

We do not have much specific information on how many people and what disorders are most prevalent in New Zealand. Very little rare disorders information is easily retrievable from health or statistical data. The information we do have (often about particular rare disorders) indicates that, overall, New Zealand is likely to have similar proportions of people affected by rare disorders as in comparable countries.

Individuals with rare disorders often have complex needs that may be unique from each other. All of them may have complex needs, but their needs are often very different. Setting up a health system that is responsive to everyone living with rare disorders is not simple.

Māori whānau affected by rare disorders

Commented [I8]: Change to Whānau Māori (throughout)

Commented [I9]: It would be good to see te Tiriti referred to as a key underlying factor for rare whānau Māori

People and whānau living with rare disorders have significantly poorer health outcomes than the whole population. This is often further compounded for Māori whānau.

Māori whānau face barriers to access culturally competent and safe health services, often having to fight to access available services, experience not being~~en~~ seen, heard or respected leading to feeling isolated within the health system.

The nature of some rare disorders means that there are one or two disorders that may affect Māori whānau more than other population groups. [include the stomach cancer example here]

Our kōrero with whānau living with rare disorders and those who care for someone with a rare disorder said services that are whānau centred and embedded within te ao Māori would make a positive impact on their and others journey across the health system.

Whānau also spoke consistently of having to fight for access to information relating to diagnosis, funding entitlements, and access to support avenues within the health system and wider social services.

Whānau provided many examples of being given a diagnosis with little, if any, guidance and support from the health system. Whānau spoke of not being listened to and treated with respect and wanting to contribute to the solutions to meet their health needs.

Many whānau, particularly parents of tamariki living with a rare disorder, experience feelings of loneliness and isolation. Whānau often felt they had to become the specialist of their rare disorder while health professionals were not open to listen to the knowledge whānau held in relation to their health.

"And so, there's a huge sort of urgency to ignore Māori ways of doing things based on what they [health professionals] might feel is more important. We experience this pretty much every time."

"So, what's important to me for my own hauora is be taken seriously. How I'm to be seen is, you know a strong Māori woman, not a symptom, not a disease. I'd like to be seen as a person. That's really important to me now."

"Knowledge is strength. The more I learn the more I can put procedures in place for my whānau, but if I'm just me learning through Facebook or Doctor Google, that's not right either. I need the knowledge properly."

Whānau also talked about good experiences within the health system when care was provided with respect and understanding for Māori culture.

"The day we arrived for the day of [tamariki] surgery the nurse we had was phenomenal, Māori herself with Tamariki in kohanga. She made sure that we had everything that we needed she asked us if we needed to have karakia and made sure that there was a space available for us. She ensured that we were able to do it our way before [tamariki] went into surgery which was great because I got to oriori [tamariki] to sleep before the surgeons came so [tamariki] fell asleep in my arms and when [tamariki] woke up after the surgery [tamariki] woke up in their mother's arms. So, she was amazing so only mum could go in after surgery I think that was a space thing, so the nurse came in and she sat with us because she knew that I wanted to be in there too, she was very comforting, and she used the bit of reo that she did know. So culturally she was fantastic she was right for us."

Commented [I10]: Huntington's Disease is another good example of a disorder more prevalent in Māori

Commented [I11]: Suggest rephrasing. It is not so much the "nature" of the disorder that causes these prevalence variations, e.g. may be due to founder effects etc.

Suggest: Some rare disorders are more prevalent in different ethnic groups than other population groups. For Māori an example of this is [stomach cancer eg]

Commented [I12]: "Whānau also spoke consistently of having to fight for access to information..." Improvements here will be good for a large number of health service users.

Health and wellbeing of people with rare disorders

Rare disorders can have a widespread effect on the lives of people and whānau living with rare disorders, including difficulties with daily activities. Some people have relatively mild impacts of their rare disorder, while for others the impacts can be incredibly severe and life-limiting. For some whānau their experience of a rare disorder involves prenatal diagnosis and others are diagnosed much later in life. From the time of diagnosis their lifespan may be very short or it may be something they live with for a long time. For many, living with a rare disorder or ~~having to care~~ caring for someone with a rare disorder often decreases their capacity to work, creating barriers to employment and education.

A wide range of social support is often required for the management of rare disorders. Emotional and psychological well-being of the individual and/or family members, especially if there is a genetic component or life limiting diagnosis, can be vital for positive long-term outcomes.

Voices of people and whānau living with rare disorders plays a vital role in the health system due to the expertise and knowledge of the disorder they often hold.

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.

International efforts to improve rare disorders outcomes

In 2021, the UN member states unanimously adopted the first-ever UN Resolution on "Addressing the Challenges of Persons Living with a Rare Disease and their Families". It encourages countries to promote public policies, patient-centred programs, and initiatives that would strengthen health systems and calls for action to address the specific challenges faced by people and whanau living with rare diseases.

Commented [l13]: Please make it explicit that NZ is a signatory to this

There is little evidence internationally as to how best to improve rare disorders outcomes. Many countries have also developed strategies and plans to guide their health and social sector to support people living with rare disorders. They have taken diverse approaches to improve rare disorders services and outcomes.

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.

Current evidence to inform epidemiology and cost analyses is limited. There is limited literature about the costs and cost effectiveness of health interventions or policies for rare disorders.

Orphanet

A portal based in 40 European countries for high-quality information on rare diseases and orphan drugs which includes directories of experts, registries, biobanks, research activities and technical infrastructure.

More to come …

Commented [l14]: It would be great to see content on virtual networks, centres of expertise and action plans here

Commented [l15]: Vision- 'Share' is very vague- how much of a share?

Commented [l16]: Strategy Goals also need discussion. How will the achievement of these goals be measured given the lack of data currently?

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Strategy vision

- People living with rare disorders and their whānau can attain equity ~~share~~ in pae ora – healthy futures and are supported to achieve the best health and life possible
- Māori whānau living with rare disorders can share equitably in pae ora
- Disabled people living with rare disorders and those living in rural and other diverse communities (including Pacific peoples) can attain equity ~~share~~ in pae ora – healthy futures.

Strategy goals

- People and whānau living with rare disorders can have ~~better~~ best possible health and live best possible lives
  ~~better lives~~
- Māori whānau living with rare disorders are provided with resources and tools to be able to exercise tino rangatiratanga
- 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.

The strategy has been developed on the recommendation of the independent review of Pharmac, reported in 2022. The Government agreed that more can be done to improve the lives of people with rare disorders and to make it easier for people, practitioners and organisations to get the information and support that would help.

The Minister of Health asked the Ministry to lead development of a strategy that will lead to better, more timely services and more equitable support and outcomes for people and whānau with rare disorders. The Government also noted that many of the Panel's recommendations for rare disorders aligned with changes being brought in through the Pae Ora Bill.

The strategy will be successful if:

- people and whānau living with a rare disorder have a better quality of life
- the health system responds and relates equitably to people whose needs or situation are uncommon, unusual or unclear.
- it is able to be utilised and built upon to develop action and implementation plans with measurable outcomes for people with rare disorders
- it leads to consideration of people with rare disorders being a routine part of future policy development, design, commissioning and service delivery conversations across health, education, and social services.

Commented [I17]: Ideally we would like to see people with rare disorders recognised as a priority population

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For 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.

This strategy outlines how the health system, health practitioners and others, together with peak bodies such as Rare Disorders New Zealand, 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.

For health practitioners and workers

…

For the health system

…

For New Zealand communities

…

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

Six Pae Ora | Healthy Futures Strategies set the long-term vision and direction for our health system that will be equitable, accessible, cohesive and people centred. The strategies include:

- the New Zealand Health Strategy
- the Provisional Health of Disabled Peoples Strategy
- Pae Tū: Hauora Māori Strategy
- Te Mana Ola: The Pacific Health strategy
- the Women's Health Strategy
- the Rural Health Strategy.

These strategies will inform decision-making and government priorities for the health system. Each strategy has a vision of achieving Pae Ora for Aotearoa New Zealand and their communities. The priority areas outlined in the Pae Ora Strategies closely link with what we have heard as priorities for Rare Disorders community, including:

- supporting access for all: accessible health services delivered closer to home
- building workforce capacity and capability
- cross-sector and cross-government collaboration to address broader drivers of poor health and wellbeing (social determinates to health)
- community voices at the centre of health service design and delivery
- ensuring timely and high-quality services that adapt to the needs and expectations of people's health
- ensuring whole-of-government commitment and accountability for Māori health and wellbeing.

The Provisional Health of Disabled Peoples Strategy has also acknowledged the commonalities between people living with rare disorders and people with disabilities and how the strategy will help direct the health system to better coordinate health and wellbeing for these communities. These include investments in identifying unmet need, early diagnosis and intervention, clearer pathways to accessing specialist diagnoses and a robust health system data and evidence base that identifies the diverse disabled population and their health and wellbeing needs.

Commented [I18]: Not sure this is quite the right word, a person can have both a rare disorder and a disability simualtaneously

Five priorities for action

There are five priorities where we need action and change over the next ten years. Action in these priorities will drive progress to improve health and wellbeing for people and whānau living with rare disorders.

Priority 1: Putting people and whānau at the heart

Priority 2: Equipping the health workforce for quality rare disorders care

Priority 3: Gearing the system for quality and timely care

Priority 4: Learning and sustaining progress

Priority 5: Joining up internationally to achieve more

Priority 6: Collection of data that will measure achievement.

Commented [I19]: Without data, how can we measure success? Either needs to be a stand alone priority or extensively incorportated in priority 2, 3 and 4

Something akin to priority 5 from the interim disability strategy ("Increase the visibility of disabled people in health data, research and evidence as part of an active learning system")

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The changes we need to see, and why, are described under each priority. People and organisations throughout the health system have a role in making the changes.

Priority 1. Putting people and whānau at the heart

What we've heard

"I just want to say that it'll be good to have a rare disorders voice." [RD Support Group Lead]

"I am unique and special – not rare" [Whānau Māori Voices Engagement]

"Being heard, everything is so transactional. Needs to be more about knowing her [tamariki]." [Whānau Māori Voices Engagement]

"A holistic approach to care is really needed, it's not just, you know, symptom based as we are treated, we actually have to be treated as a whole." [RD Support Group Lead]

"You have to be your own advocate and when you are battling and already exhausted this is hard to do." [RD Webinar Engagement]

"It's changed our lives completely. I've gone from someone that was really like, I don't know, confident, you know, working hard. Making a living, you know, and we really were very social people. Like our lives were completely different." [Whānau Māori Voices Engagement]

"Eventually we managed to push, push, and push again and I had to get assertive again and I would say, "I need something done and I need it done now." Then I feel bad having to talk about something like that but then I think about [tamariki] and she needs this, and I advocate for her.

It's the wait time when you need it you need it now. If you leave things too late you miss the boat,

she's already got delayed development she doesn't need delayed services. If I didn't push for everything she would have fallen through the cracks." [Whānau Māori Voices Engagement]

"Now that our local [GP] surgery has been taken over by … a Māori organisation and now I walk in and there's Māori nurses, Māori doctors. I honestly just feel so much more comfortable, and they make us feel like we're at home, you know, and it's just a totally different surgery and I love walking in there and them helping [tamariki] has just been amazing." [Whānau Māori Voices Engagement]

"They [health care team] walked in and introduced themselves to our family. They would also say [pāpā] introduce us to your family. If a doctor walked in they would walk in and know our names. He [pāpā] would say 'who are you what do you want' when they just walked in without introducing themselves and asking about us [his whānau]." [Whānau Māori Voices Engagement]

"I've got lovely doctors … my specialist is a lovely joker and good friend. Your stay in hospital is always made better if you have good nursing staff and good doctor. Someone who is passionate about their jobs. There are so many doctors who are basically overworked and underpaid, and it shows in their presentation, and I go, no, no, no, don't fob me off I want more than five minutes of your time. I try not to dwell on the bad, I try to focus on the good." [Whānau Māori Voices Engagement]

Why it matters

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~~ Considering and integrating 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.

For whānau Māori and all with rare disorders ~~in particular~~, being listened to and understood in a welcoming and respectful environment makes a major difference to comfort and trust with services and practitioners.
Building care plans together makes a big difference to health outcomes in the longer term.

Commented [I20]: Stronger and less deficit based language

What the future will look like

People and their whānau will be the most important contributors to their health and care

With people and whānau at the heart of health interactions, attention is on what they say is most important. Services will adapt to their cultures and circumstances, preferences and choices. A holistic view of wellbeing will be the starting point and health and wellbeing will be supported even when there is no diagnosis or treatment. People and whānau with unknown or rare disorders will feel welcome and supported to improve their health and lives.

What is most important to people will drive the priorities of health providers. People and their whānau will get support that helps them understand and participate in their health care as much as they would like. That includes having explained the recommended standard of clinical care and how this translates to their own situation.

People and whānau will have choices and be empowered to make decisions on their care

People will have their own health records. 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.

People's expertise and knowledge will be valued and inform the care that is suggested. They will be welcome to bring in other support and expertise, and make decisions on their care. Whānau needs will be met so that they can nurture whānau members with or without a rare disorder.

People with rare disorders or possible rare disorders will receive high quality care wherever they live

The codes of health and disability consumers' rights and of expectations for health entities' engagement with consumers and whānau for be upheld for people and whānau living with a rare disorder or the possibility of a rare disorder.

No matter where in New Zealand people live, they will be able to have expert input into their assessment, diagnosis and care. When they need to travel for this input, or to visit unfamiliar services, they will be supported. They will be listened to and will no longer feel the need to 'fight' for attention or to be heard.

Voices of people and whānau with rare disorders will be pervasive across the health system

Voices of people and whānau with rare disorders will be heard at all levels and across all functions in the health system. They will be present in policy, design, commissioning and service delivery conversations. They will inform progress and outcome monitoring, iwi Māori partnerships, research and improvement programmes and strategic overview of New Zealanders' health and wellbeing.

Communities will support wellbeing of people and whānau living with rare disorders

The health sector will work with diverse communities to build, maintain and enable strong foundations for health and wellbeing, and for including all community members. Health promoters, occupational therapists, physiotherapists and others will support communities to include people and whānau living with rare disorders. People and whānau will be supported to participate in everyday activities and connections that promote wellbeing. The health sector and health professionals will work with Rare Disorders NZ and rare disorder specific support groups to ensure that people with rare disorders are provided with opportunities to engage with each other and find support.

What needs to change

Voices of people and whānau with rare disorders will be present across the health system

Increasingly, people who can speak to rare disorders experiences will be members of forums across the health system. These forums will include consumer forums, advisory boards and committees, locality groups and partnership boards. They will include ad hoc committees and working groups set up for specific purposes such as developing a guideline or programme or planning an event.

Rare disorders voices will come across in major system reviews, service evaluations, business cases and investment decision-making.

Commented [I21]: What does this actually mean? Electronic access? Easier access through the Privacy Act? Ideally access would be barrier free, simple and all in one place.

Commented [I22]: We aren't sure what this means- is it refering to a rare disorders tag in electronic notes (similar to an allergy alert?) - may need an example

Commented [I23]: Is this talking about ensuring the needs of the whole whānau is met or about families without a lived experience of a rare disorder? If the former then maybe "Whānau will be supported to nurture whānau members with a rare disorder without this impinging on their ability to meet the needs of the rest of the whānau". If it is the later it does not belong in this strategy.

Whānau Māori and tangata whaikaha living with rare disorders will experience manaaki

Whānau Māori will feel empowered after consultation with their health care team. Health care experts will cater to the needs of each whānau with empathy, making them made them feel at ease. They will allow the space for karakia and tikanga Māori and communicate in a way that is clear and captures everything. A comfortable and respectful environment will feel safe for whānau and help build trust.

Whānau Māori will be acknowledged, welcomed and listened to in a collaborative relationship with care providers. They will build connections and work together with their care team towards aspirations and goals. They will be offered choices where possible. Their choices will be accommodated, including for rongoa Māori or mirimiri to restore wellness of mind, spirit, body and emotions.

Community wellbeing efforts will include people and whānau living with rare disorders

Activity in localities and diverse communities will increasingly focus on promoting health and wellbeing for all community members. As activities, programmes and events focused on health and wellbeing increase, so will the focus on inclusion of all community members. People and whānau living with rare disorders will be supported to participate. People who can speak to rare disorders experiences will be included in the planning and implementation of such activities.

Priority 2. Equipping the health workforce for quality rare disorders care

What we've heard

"My GP took a proactive approach to making referrals that led to my diagnosis." [RD Webinar Engagement]

"Improved knowledge of local Paediatricians has helped with my quality of life." [RD Webinar Engagement]

"The other thing that's important to me is having access to doctors that understand the disease you know, and having that knowledge at their fingertips, that's important because otherwise that's what helps create that misdiagnosis and people waiting years before they're actually being diagnosed." [Whānau Māori Voices Engagement]

"There is often constructive curiosity among clinicians about my rare disorder. I'd love to see that fostered through further education about rare." [RD Webinar Engagement]

"Clinicians need a place to go [for information and guidance], this is where so many of us have fallen between the cracks." [RD Support Group Lead]

Commented [I24]: We expect to see clinician voices captured in the quotes here

Why it matters

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.

Commented [I25]: Back this up with quotes in above section from these people

Commented [I26]: what exactly does this mean? Who are the others?

What the future will look like

Health practitioners and workers know about rare disorders and how to find out more

Students of clinical disciplines (medicine, nursing, midwifery and allied health such as pharmacy, physiotherapy and psychology) will learn about rare disorders through examples incorporated into many of their learning areas. Learning about rare disorders will be integrated with broader learning modules so that students become alert to the possibility of rare disorders and learn how to support people while a rare disorder is investigated and care plans developed.

Health practitioners' ongoing learning will be bolstered with integrated rare disorders examples. Options will be available for more detailed supported learning in a range of rare disorders and related topics.

Care and support workers, health promoters and people in education and social care roles will also have learning opportunities that include integrated rare disorders support skills. People working in technical, research and counselling roles will similarly have rare disorders learning opportunities.

Health practitioners and workers will be able to support people and whānau with possible rare disorders

There will be plans and initiatives to create and support a health workforce that mirrors the diverse communities of Aotearoa.

All s~~S~~tudents of health disciplines will experience tikanga Māori practices in health. They will learn about the impacts of showing manaaki and respect for diverse consumer preferences and needs. They will learn about supportive and strengths-based inquiry to identify wellbeing needs when people are facing uncertain or difficult circumstances.

All health service workers will have learning opportunities in supporting whānau to decide on their own health goals and plans for mauri ora, whānau ora and wai ora, and on including mātauranga Māori.

Ongoing clinical education and competence will include cross-disciplinary and experiential learning options in a wide range of supportive, counselling, tikanga and social wellbeing areas.

A wide range of clinical support tools will support diagnosis and care for rare disorders

Clinical guidelines and standards and referral, diagnostic and care pathways will support care for a large and increasing number of rare disorders. These tools will be approved by New Zealand authorities and adapted for different localities or situations where necessary. They will be housed in the standard approved collections that health practitioners most often use, and visibility will be automated through integration with patient management systems and electronic health records.

Specialist help, advice and backup will be available for practitioners in all levels of the system

Practitioners working in all settings across the health system will have access to expert opinion and advice that helps them to provide quality care for people and whānau with rare disorders. This help, advice and backup will be provided in timeframes that are responsive to needs.

A wide range of expert input will be accessible for community health providers from a range of specialists, therapists, health promoters, cultural advisors, people with lived experience. Specialist practitioners will be able to receive expert input from practitioners who have experience of the relevant rare disorder, from their own and other specialties.

~~For very rare disorders or highly specialised needs,~~ international input will be available *for rare disorders if appropriate expertise is not available in Aotearoa New Zealand*.

Practitioners will inform people and whānau about Rare disorders NZ and relevant rare disorder specific support groups as an additional resource and to provide the opportunity for them to form connections with others.

What needs to change

Health practitioners will learn about rare disorders

Curricula for health practitioner education at all levels will be revised over time to ensure rare disorders are highlighted more frequently. Examples will highlight the impacts of rare disorders and the benefits of identifying and providing the right care earlier. As diagnostics, clinical guidelines and care pathways are developed, students and practicing health practitioners will learn how to access the information to support care.

Commented [I27]: Ultimately we support this but with >7,000 rare disorders it is unrealistic to achieve for all and having international tools/guidelines etc approved for some automatically makes the others not approved. We would like to see the barriers to seeking and following international best practice reduced (recognising that our health system will not always have equivalent care available as an option). One idea is that specialists can link/indicate reputable international guidelines within the electronic clinical record to make it more official/easy to access for GPs, ED, allied health etc who come into contact with the person.

If tools/guidelines are being approved and adapted these would need a timeframe for review and a system that includes horizon scanning for the disorders that do have tools approved.

Commented [I28]: This is important for all rare disorders, Rationale is that this could be used to limit access to international advice.

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Commented [CH29]: Nothing about diagnosis in this section

Commented [I30R29]: From Interim Disability Strategy "There must be clearer pathways to accessing specialist diagnoses for rare disorders and for adults accessing diagnoses for rare disorders normally diagnosed during childhood. Waiting for a diagnosis can delay access to other support services, which is detrimental to overall health and wellbeing. This is particularly the case when a health care diagnosis can strengthen an application for funding for support in a non-health related area" (page 17)

Learning modules for rare disorders will be widely accessible

Health workforce learning opportunities in rare disorders will be increasingly available and more sophisticated. Learning modules will include rare-disorder-specific offerings as well as more general offerings that include rare disorder content. Rare disorder examples will feature in

an increasing portion of health workforce learning opportunities. Rare disorder examples will feature increasingly in competency-based learning.

Learning modules for support and wellbeing will be widely accessible

Learning opportunities relevant to the care and support for people and whānau living with rare disorders will be widespread. Rare disorder examples will be included in an increasing portion of learning opportunities in tikanga practices, cultural safety and competency, care and support, counselling and social wellbeing.

Clinical guidelines and pathways for rare disorders will be adapted, adopted and assimilated

A programme will be established to identify, adapt, authorise and publish guidelines, standards and pathways for rare disorders in New Zealand. The programme may start with rare disorders for which a national service or similar well-established approach is in place and move on to continually expand sets of tools to support care quality for increasing numbers of rare disorders.

Adaption for New Zealand may include negotiating pathways for referrals, consultations and care transfers across different levels and different service lines in the health system. It may include guidance on when to refer for highly specialised care or for exceptional circumstances access to tests, treatments or care not usually available. Applicable rare patient groups and their representative organisations will be consulted in the development of such documents.

Over time, published tools will be integrated with standard reference sources and patient management systems.

Experts in care for rare disorders will be supported to provide practitioner help and advice

Expansion of help, advice and coaching functions available to practitioners will require the resourcing and backup systems to make it easy for practitioners to ask for and receive help, including online.

This will enable Practitioners to support people on their journey to diagnosis and throughout the course of their disorder, especially at times of transition.

Capacity review and re-design of specialist expert roles is likely to be required to ensure time for this backup function is built in as well as providing for an increase in people with identified rare disorders needing care.

Capability development for specialist experts will be required to support their ongoing learning for the role. Cross-specialty learning in how backup and advice functions best cultivate quality and confident care provision will be important.

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Priority 3. Gearing the system for quality care

What we've heard

"[Support Group] would encourage protocols to written by Health NZ, be audited by the NZ Endocrine Society and then applied to hospitals throughout NZ." [RD Support Group MoH Survey]

"I would really like a brochure about hypoparathyroidism to be given to anyone getting a thyroidectomy. There are things you can look for post-surgery & having a name could help you understand what to research! you cannot research something if you do not know what to look for." [RD Support Group MoH Survey]

"I waited almost a year to get access to genetic services, no specialists would take any action or make plans for me while I was waiting. Precious preventative treatment time was wasted." [RD Webinar Engagement]

"Differences in the level of specialisation and clinical resources means healthcare is experienced differently for people across the country." [RD Webinar Engagement]

"I was able to be diagnosed relatively quickly but was given no information about what the diagnosis meant and no access to specialists with knowledge about it." [RD Webinar Engagement]

Rare Disorders NZ along with the rare disorder community have collectively identified eight strategic priorities to improve the health and well-being for people living with a rare disorder.

These are:

1. DIAGNOSIS
- *Early and accurate diagnosis of rare disorders*

2. PLANNED PATHWAYS FOR CLINICAL CARE
- *Coordinated and integrated pathways for cohesive healthcare*

3. ACCESS TO DISABILITY AND SOCIAL SUPPORTS
- *Implement simple mechanisms to ensure appropriate access to disability and social supports*

4. RARE DISORDER MEDICINES
- *Equitable access to modern rare disorder medicines through a specific assessment pathway*

5. RESEARCH
- *Coordinated and funded programme of research for rare disorders*

6. NATIONAL RARE DISORDER REGISTRY
- Data collection and incorporating coding of rare disorders in the classification system of diseases

7. WORKFORCE DEVELOPMENT
- *Planned training on rare disorders for health professionals and support staff and development of a national rare disorder workforce strategy.*

8. INCREASE AWARENESS
- Awareness of rare disorders within the health system and the general public is increased.

Rare disorders NZ has proposed the creation of a Rare and Undiagnosed Disorders Centre of

Commented [I31]: We would like to see our priorities for improving the health and well-being for people living with a rare disorder in this document. It could fit here under What we've heard or why it matters. A brief outline with the details in the appendix would not interrupt the flow of the document.

Expertise in the health system as a way to work on implementation of the rare disorder strategy and progression of these priorities.

Why it matters

As well as learning, tools and back-up that equip practitioners and others to provide quality care, how the wider health system is organised and coordinated matters. Rare disorders often come with multiple and complex needs and involve many parts of the health system. High quality, timely, safe and life-affirming care needs to be accessible and navigable. People and whānau of all needs, all stages of a health journey, and even the most rare conditions need to be accommodated.

System improvement across three areas will make a big difference to rare disorders outcomes:

- connectedness across different parts of the system and navigation support
- early adoption of more effective, efficient and safe technologies
- ways to prioritise access and investment across services, products, ~~and~~ infrastructure, ~~that work for rare needs.~~new technologies, digital capabilities, medicines, devices and other therapeutic products.

What the future will look like

The system will be connected up around people with rare disorders

Health funders and planners, practitioners and providers will recognise the many common features in the types and complexity of rare disorder health care needs. Their ways of working will be based on networking, coordinating and sharing expertise across services, specialties, locations and knowledge bases. Connecting and drawing on connections will be a large and expected part of health workforce roles.

Efforts to improve system responsiveness to people with rare disorders will be known and visible. A recognised 'home' for rare disorders leadership across the system will be well-established, with strong links into many different service lines and networks. People in any part

of the health system will be able to find information about rare disorders. They will be able to find out where to go for answers to their questions. There will be established ways of ensuring timely access to different specialties and services where these are needed to contribute to diagnosis or care.

How the system works for people with rare disorders will be consistent with other sector areas so it will be easily understood and promote maximum visibility and uptake of improvement initiatives.

As the only national organisation supporting all New Zealanders living with a rare disorder and their carers, Rare Disorders NZ has played an integral role in supporting the development of the Rare Disorders Strategy, by advising, providing essential information and establishing connections between the Ministry and stakeholders, including patients and whānau. The organisation is well-placed to continue to support the implementation stage of the strategy, and will be acknowledged and compensated accordingly.

New technology will identify more disorders, sooner, making preventive care possible

Work on precision health is building. New technologies like genomics and artificial intelligence will open up opportunities to predict, prevent, diagnose and treat health needs sooner and more precisely.

New Zealand will continue to identify and do the ground-work for new technologies and digital capabilities to prepare for early adoption. Work to ensure safety and fair application will continue to be done in advance of full development or affordability assessments. This will allow early adoption of promising new capabilities once safety and value for money is established.

Fair access to diagnosis, care, treatments and supports will be constantly improving

~~Affordability will continue to be a challenge with new and intense services, treatments and technologies.~~ While some of these will be researched and developed in New Zealand, most will be developed elsewhere in the world. Up-front investment will be required for research or use in New Zealand.

As new technologies are developed into treatments, many will first be used to treat rare disorders, as we are seeing with CRISPR-based treatments now. ~~The cost per person will be very high initially.~~

As technologies become more widely used, prices may decrease somewhat – but affordability challenges will remain. Technology, infrastructure and special expertise will often be required. At any time, there will be some diagnostics, medicines, devices and other treatments not able to be provided safely or affordably in New Zealand.

However, processes to consider new~~-~~ diagnostics, technologies, digital capabilities, medicines, devices, other therapeutic products ~~diagnostics,~~and treatments, services and infrastructure will be faster and fairer. They will account for all, including rare, needs and for lifelong needs alongside emergency and acute needs. They will be more consistent across different types of investment (such as in medicines, new services, overseas treatments). The ways that people in exceptional circumstances can be supported to access expensive or unavailable tests, clinical trials, screening, medicines, devices, technologies other therapeutic products and treatments, overseas or in New Zealand, will continue to expand.

Commented [I32]: Frame treatment more positively as an investment. "New and intense services, treatments and technologies provide an opportunity for the government to invest in pae ora for people and whānau living with rare disorders".

It is essential that a broad view is taken to this, that looks not only at savings to the health sector but to the benefits in other sectors and to individual whānau. E.g. work capacity for the individual and their wider whānau, access to education for affected children and siblings and the long-term impact on their economic activity, and importantly the difficult to measure wellbeing of people and whānau.

Commented [I33]: if discussing costs need to also discuss savings to health system - we don't believe the info is here to provide this- nothing else is costed in this document

Commented [I34]: Please either define treatments to include medicines, devices, technologies and other therapeutic products, OR list all every time

People who are undiagnosed or who have a diagnosis of a rare disorder will be routinely given the option to be linked with Rare Disorders NZ and relevant disorder specific support groups by their health practitioners.

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What needs to change

As more rare disorders are identified and knowledge accrues globally, there will always be more to do to improve system responses and outcomes. Changes to knowledge sharing and coordination, and fair access to services and treatments will be ongoing.

Leadership and coordination will be built to spearhead responsiveness to rare disorders

Responses to rare disorders, currently in service lines and pockets across our health system, will be built on and connected up. Leadership and coordination across services and levels of the system will be established with the capability, recognition and support given to other system-wide leadership and improvement initiatives.

Leadership and coordination mechanisms will oversee the adoption of elements that together make a coordinated and responsive system. These will include:

- diagnostic, referral, navigation and care standards, guidelines and pathways
- capability development reports and initiatives
- recommendations for service and system enhancements.

Choice of leadership and coordination mechanisms will promote visibility and understanding across the system, sustainably for the long term. A range of approaches used in other countries (such as networks, centres, national services and information hubs) all have ~~adherents.~~ The choice of mechanism will be strongly influenced by how easily and sustainably it will integrate across the system and be supported with the resourcing to ensure continuous improvement.

Rare disorders NZ has advocated strongly for a Rare and Undiagnosed Centre of Expertise to be established in New Zealand. Rare Disorders NZ envisages that the Centre will identify gaps in the delivery of service and support in New Zealand's health and other systems for people living with rare disorders, similar to other initiatives globally. The Centre will comprise a multidisciplinary team of internationally-networked experts within New Zealand, who will be available to provide expertise and guidance for clinicians and professionals in how to support rare patients and clients in accordance with best practice standards. This work will be an extension of roles they hold with locality-based health and other service providers.

New capabilities will be introduced to speed diagnosis and allow earlier preventive care

New genomic testing for suspected rare disorders will speed up diagnosis,~~-~~including in infants through expansion of the newborn screening program. ~~especially for infants and children.~~ Rapid DNA sequencing will allow precise identification across several thousand genetic disorders, rather than testing for one at a time. The capability and service infrastructure to support early testing and preventive care will be built over time.

Horizon scanning will inform early work to prepare for possible introduction of developing technologies and capabilities, medicines, devices, and other therapeutic products, research clinical, diagnostic and screening advances that are of potential benefit for people with rare disorders. A work stream to clarify risks and benefits and progress the conditions for safe adoption of technologies and capabilities will be maintained for ongoing yet flexible advance preparation.

Ways to assure safety and quality of new technologies and digital capabilities, and to communicate about them, will be developed early in advance of their adoption. People and whānau living with rare disorders will provide key input to this work.

Ways to prioritise investments will improve to value and consider benefits for people and whānau living with rare disorders

Commented [I35]: Assume these will be detailed in futher depth earlier in the document? Examples of how this has been achieved internationally include (See appendix E of RDNZ's August submission, Centres of Excellence and reference networks for rare disorders around the world), would recommend at least go into detail on Centres of expertise (UK, WA Rare care centre) and Virtual clinical networks (European reference networks)- this could also be linked to the development of virtual networks here by Te Whatu Ora.

Commented [CH36]: ...and have the potential to be adapted, either singly or in combination, to address what's needed in New Zealand.

Commented [I37]: Specialist quote to be incorporated

Commented [I38]: We do not want to see adults marginalised in this document

A programme of work across health entities will focus on improving quality and timeliness of decision-making on access to diagnostic, preventive and treatment services, products and infrastructure. This work will include a particular focus on rare disorders.

An early focus of the work will be improving and clarifying how and when people with exceptional health needs or circumstances will be supported to access otherwise unfunded ~~expensive~~ or unavailable tests, advice, medicines, devices and other treatments, overseas or in New Zealand.

Over time, the work will consider how to improve such things as:

- estimating and valuing all of the impacts for people and whānau living with rare disorders of having or not having the service or product (not only direct impacts for individuals' health). This will be a holistic view that considers the whole life of the person and their whānau, including any relevant opportunity cost as well as the wider social and economic impact across systems including health, education and social services, over an entire lifetime and with consideration of intergenerational effects.

- ~~estimating and valuing lifetime impacts for the health system and government as well as people~~
- equity including inter-generational equity
- ~~fairness~~ equity across different types of investment (such as in medicines, new services, overseas treatments)
- ~~fairness~~ equity across different types of need (such as rare or common, prevention or treatment, ongoing or once-only, urgent or non-urgent, curative or ameliorative),
- Establishing an independent that which can receive and review complaints and cases where people and practioners do not believe decision making regarding access to medicines, devices, therapeurtic products, technologies etc has been equitable.

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Commented [I40]: We are not sure what this means- would equity be a better word than fair?

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Priority 4. Learning and sustaining progress

What we've heard

Commented [l41]: Incorporate quotes from clinicians and experts

"Collection of data to better inform care and supports, improve access to expertise/sharing of information for all – too many inconsistencies." [Pae Ora Webinar Engagement]

"Recording information about rare disorders so we know the true extent of the issue – we can only use overseas statistics at the moment which means we're guessing how it applies to New Zealand." [Pae Ora Webinar Engagement]

"Being able to capture those in the system that are currently being missed." [RD Webinar Engagement]

"someone with a 'sprained ankle' with the possibility of it being broken but won't be x-rayed till the next day would still get a diagnosis on the ACC form as a possible or probable and that's OK, yet when moving into the RD space to have a 'suspected' it needs to have a code." [Clinical Hui]

"We need to start measuring rare disorders data so we can understand the scope of the issue and provide better levels of care." [RD Webinar Engagement]

Why it matters

Not only does the system need to be geared towards good and timely care for people with rare disorders. It needs to learn about health and the care and outcomes people are experiencing, and to track progress over time. It needs to collect and spread information so that those planning, delivering and using health care can see where improvements are being made and what more needs to be done.

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 research into service models and approaches for rare disorders is important to grow effective care that fits with our population and how it is spread across the country. Being able to participate in clinical trials of new diagnostics and treatments is important to people with rare disorders for whom no effective treatment may otherwise be available. It is also important to grow capacity and capability in our research and development industry and in uptake of new advances by our health services.

What the future will look like

Rare disorders will be visible in our system

Information to support care of people with rare disorders or possible rare disorders will be captured in patient records. Medical coding will keep up to date with international classification updates and allow for continual updating of world knowledge about rare disorders. Other data of equal importance, like disability information and links to specific rare disorders collections, will be built in.

Well-developed systems of governance, protections, access and ownership of system data will be in place. Māori data sovereignty will be well understood and protected. Rare voices, along with other consumer, cultural and minority voices, will be at the table when these system foundation elements are discussed and decided. Citizens will have a degree of choice in their data and will be confident in in-built protections.

Accurate health data for people with rare disorders will be reported regularly and used to inform commissioning and service design decisions.

We will know how well our system works for people with rare disorders

Information on rare disorders will be routinely collected and reported. People across the health system, including service users, planners and providers, will help guide the information collected and how it is used. A variety of data will be able to be matched to rare disorder codes so that health trends and progress are tracked without compromising personal information.

Planners and researchers will be able to investigate services and outcomes for particular rare disorder groups or clusters. They will be able to compare New Zealand results with those in other countries, or in some cases across different parts of New Zealand.

We will know how well our system works for Māori and tangata whaikaha with rare disorders

Māori will have an integral role in design and use of systems to assess and track progress in health and healthcare for whānau Māori and tangata whaikaha living with rare disorders. Reports will inform how we assess equity of outcomes and health system delivery, and programmes to continuously improve outcomes. How success and equity of outcomes is defined will be designed in consultation with whānau Māori with rare disorders.

Judging how well our system works will include how it works for people with rare disorders

Assessments of our health as a nation, and how well New Zealand's health system is performing, will include specific information on rare disorders.

- As now, these assessments will look at trends in health status over time, using averages and compounded data like life expectancy and health expectancy. They will focus, too, on the spread of these indicators across peoples and groups within our population. Trends in health status of people with rare disorders will be part of assessing how well our system works for all New Zealanders.
- Also as now, these assessments will look at trends in people's experiences of the health system, like wait times for diagnostics, assessments or treatments; how easy to access and affordable services are; and how confident people are in their practitioners and

services. They will look into differences across peoples and groups and in different parts of the country. Trends in experiences of people with rare disorders must be included and considered in evaluations and determining ~~will be important pointers to~~ where further improvements can be made.

Health research will include programmes on rare disorders and health services for rare disorders

New technologies and new research tools are being developed for clinical applications at an increasing rate. New Zealand plays a part in this research and development in genetic, metabolic and other research fields of particular relevance to rare disorders. Our part will increase over time as we build on the strong health research capability already being fostered.

New Zealand will be well connected into international clinical trial networks and have the infrastructure to support this collaboration.

Government policy, programs and funding will support a ~~A~~ coordinated approach to learning from, translating and embedding health service advances for rare disorders ~~will be visible~~. It will include New Zealand-led research as well as testing and evaluation of international research findings in our context.

What needs to change

National health datasets will routinely collect rare disorder information

The level of diagnostic information that can be readily searched and reported on will become fuller and more detailed over time.

- Clinical classifications will become increasingly detailed as more advanced classification systems are incorporated in national data collections. New rare disorders are being identified at ever faster rates. Classification systems will need to grow in specificity and to connect easily to supplementary datasets specific to rare disorders. An example of how this could work is through the implementation of the nationwide Electronic Medical Record (EMR) using SNOMED terminology, there is an opportunity to incorporate coding of rare disorders using Orphanet Coding (ORPHACODES) in this system, as 6,500 Orphacodes, have been mapped to SNOMED CT codes. Orphanet is the most comprehensive database of rare disorders.
- Ways of capturing suspected but unknown rare disorders will need to be developed. ~~General (including 'unknown' or 'unspecified') codes may be considered for interim use where more specific categories are not yet available or applicable.~~

Commented [I42]: Can this be expanded on to explain further or deleted. It currently reads that rare disorders will just get lost amoungst the NOS codes (Not otherwise specified), would it be Rare disorder suspected - unknown? We agree it is important to capture this information.

Rare disorder information will be used to inform, monitor and improve care

As rare disorder information becomes more visible in the system, it will be used to plan care, follow care provision and results, fill gaps and make improvements. This will build on work already done for some rare disorders, such as metabolic disorders.

- Ways to link high quality rare disorders information with national health information will be developed where clinical coding does not yet allow for disorders to be captured.
- Linked information will be made available, in non-identifiable form, for planners and researchers to use to improve care. Where possible, incorporation in the Statistics NZ Integrated Data Infrastructure will allow investigation of wider government services and supports in addition to health care.

Health information reports will improve care and protect personal information and choices

Both regular reporting and more specific enquiries will support quality care provision.

- Connecting up data for individuals and whānau will enable better and more timely care

and choices for them.

- Connecting up data across groups of people with rare disorders will inform improvements in care, care pathways and outcomes for the future.

- Connecting up data across the population of people living with rare disorders will inform improvements in how the system as a whole can better support and enable their wellbeing.

Deciding priorities for system evolution and change will include looking at rare disorders

System planners and stewards will look at the likely impacts on outcomes for rare disorders before making system changes.

- People and whānau living with rare disorders will be one of the population groups routinely thought about when designing system improvements. Though service and system improvements for the whole population or large population groups (such as children, or Māori, or rural communities) may lead to better outcomes for rare disorders, this will not be assumed.
- Work programming for system improvements and investments, and their evaluation, will give priority to areas where benefits for people and whānau with rare disorders are likely to be higher. This will include, for example, giving weight to inter-generational equity; to improvements in services for or highly used by children; focus and improvement on services and wrap around care at times of transition (e.g. transition to adult services, transition to assisted living facilities); to genetic and genomic infrastructure and services; and to the supports and therapies (including physical and occupational) that benefit people living with rare disorders.

Health research infrastructure will support rare disorders research and evaluation

Infrastructure that supports conducting and participating in health research, and in health services research, will be increased. Rare disorders research will be a key component. This will include research and evaluation of services and systems for rare disorders, along with measuring their outcomes, effectiveness and value for money.

Important research areas for growth will include:

- clinical trials, where support infrastructure will be built and will increasingly enable participation from any part of New Zealand, in clinical trials led here or elsewhere
- methodology to measure and more accurately value a wider range of benefits and costs to people and whānau with rare disorders, and applying the methodology to support decision-making on investments and access
- evaluation of service development and improvement programmes and other initiatives to improve responses for rare disorders.

Priority 5. Joining up internationally to achieve more

What we've heard

"We need to work with and learn from other countries that are ahead of us." [RD Webinar Engagement]

"NZ should reach out international partners such as Australia to improve in knowledge, delivery & access to medication and treatment options. Much of the work has been done already overseas & we can utilise that to upskill." [RD Support Group MoH Survey]

"Incredible international network of specialists, researchers and patient groups who generously share their expertise and experience and conduct research to improve the lives of those living with …. … Australia has a nurse who provides advice on the phone or online. US, Australia and UK have … clinics in some areas." [RD Support Group MoH Survey]

"Overseas there are clinicians with a lot of experience and knowledge about … who are willing to be consulted for free by their NZ counterparts, and there are freely accessible International … Best Practice Guidelines covering many different aspects of … care and treatment, but in NZ it is very difficult to find a clinician who knows anything about … and yet we very often struggle to get the local clinicians to take any notice of the international experts or the guideline." [RD Support Group MoH Survey]

Why it matters

Due to the number of rare disorders, no country individually can~~New Zealand is a small country and, alone, cannot~~ hold expert knowledge of every rare disorder. 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.

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

What the future will look like

New Zealand will be a global partner and contributor on rare disorders

With government support, New Zealand health and research leaders and organisations will be participants in a range of international partnerships with a focus on rare disorders. The partnerships and contributions will be across a wide range of topic areas, including research, medication access, outcomes, systems, affordability and equity. People with rare disorders will be participants and inform the partnerships.

New Zealand will learn from other countries and will contribute to global knowledge on rare disorders and improving rare disorders outcomes. This contribution will include the government reporting information to global monitors as well as contributing knowledge from research, evaluation and health monitoring.

New Zealand will play a leading role in rare disorders indigenous health and equity

New Zealand indigenous health and research leaders and organisations who can speak to the experience of those with rare disorders will be participants in international indigenous health partnerships with a focus on rare disorders. Whānau Māori and tangata whaikaha will be participants and inform the partnership goals and work programmes.

International rare disorders networks will enhance care provision

Networks of experts across countries will provide backup and advice on care for rare disorders. Networks will range across healthcare disciplines and lived experience, as well as topics like medicine access, and support continuous learning.

New Zealand will receive expert input to inform care here, especially ~~for very rare disorders and~~ where there are new developments. Our experts will contribute their experience where special expertise is developed here.

New Zealand experts will provide advice and backup for neighbouring countries, especially across the Pacific.

International collaborators will work on faster uptake of new technologies and treatments

New Zealand health leaders, researchers, assessors, regulators and funders will be involved in international efforts to make it easier, faster, safer and more affordable to use new diagnostics, treatments and technologies for rare disorders. Processes, standards and regulation will be aligned across countries. Research and industry agreements will be in place to enhance global efforts towards and affordability of rare disorders advances.

What needs to change

New Zealand health leaders will actively engage in international rare disorders collaborations

We will partner with other countries in efforts to improve outcomes for people and whānau living with rare disorders. We will work towards effective contributions to global efforts, and seek out opportunities to collaborate with other countries in our region.

We will actively support international efforts to make it easier, faster, safer and more affordable to use new diagnostics, treatments, medicines, devices and other therapeutic products and technologies for rare disorders. We will support the interests of smaller and less well-resourced countries, especially our Pacific neighbours.

We will learn from other countries and adopt and adapt their tools, resources and advances

Rare disorders programmes, networks, centres and other efforts have existed for some years in a range of countries. Guidelines, decision trees, referral and care pathways and similar tools are widely used, based on or generating evidence of good outcomes for people with rare disorders.

A programme to adapt the most relevant tools for New Zealand will support good care becoming more easily and widely accessible. 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. Prior to guidelines being adapted, international best practice guidelines are sought out and utilised to the extent possible within the NZ context .

Keeping abreast of evidence on effective approaches to rare disorders will remain important. Evaluations like that currently underway of the European Union's virtual networks of healthcare

providers can generate new knowledge of ways to share and concentrate knowledge and resources in order to make highly specialised care accessible.

People with rare disorders will be more able to participate in international clinical trials

Our clinical trial infrastructure will support New Zealanders being able to participate in international rare disorders clinical trials. Clinicians involved in trials will receive backup and research support where their patients are participants in suitable funded trials, even when there are very few potential participants from New Zealand.

Regulation and regulatory processes and standards will support rare disorders outcomes

New Zealand's regulatory stewardship programme will review relevant legislation and regulatory frameworks to ensure they support rare disorders outcomes. Regulation of new technologies, digital capabilities, medicines, devices and other therapeutic products can support safe and timely access to diagnosis and effective treatments. Flexibility will be required for the appropriate level of scrutiny and safety precautions for a range of different rare disorder scenarios. Alignment of New Zealand's regulatory approaches with those of like-minded countries will reduce timeframes for assessment. New Zealand will have regulations to facilitate orphan drug access to the New Zealand marketplace and help offset orphan drug development costs (Orphan drug regulation).

Comments

Some areas are very specific and detailed and are aspirational about outcomes that clinicians, researchers and universities will have to drive. We are happy to see this, however the Strategy currently lacks the same aspiration and detailed outcomes in relation to work that Te Whatu Ora, Pharmac, and Manatū Hauora would be responsible for. We would expect to see at least the same level of detail and aspiration for government controlled and driven outcomes in the 'what needs to change' sections.

We cannot give feedback on content that we haven't seen and expect to see a final version incorporating the current 'yet to come' sections.

Missing from this document

- **Medicine access**

  Health entities will provide equitable access to modern rare disorder medicines through a specific assessment pathway including:

  - Future-proofing a pathway for new and innovative modern medicine for those with a rare disorder, for example gene therapy.
  - Updating the factors considered in cost-benefit analyses to include the wider health system, social support system, and society costs and benefits.
  - Funding PHARMAC so that it is able to fund new and innovative modern medicines in a way that is consistent with best international practice
  - Developing and implementing a medicines strategy to include rare disorders, gene therapy and innovative modern medicine.

Commented [I43]: Is the strategy separate to what Min Little was talking about here? Is there another workstream or was this meant to be in the Strategy? Minister Little 1 June 2022: "What the Pharmac review actually said is that the Ministry of Health should take responsibility for some medicines strategies, particularly in relation to rare disorders, because what they were saying was that Pharmac, as a procurement agency, should not be the sole arbiter of what a medicines strategy should be. And that is the recommendation we've accepted, as I've announced today, and we will do that." https://www.parliament.nz/en/pb/hansard-debates/rhr/document/HansS_20220601_052740000/little-andrew/

- Developing and implementing policy that supports timely and equitable access to new, emerging and best available health technologies.
- Ensure people with a rare disorder have equitable access to medicines with demonstrated clinical benefit for a rare disorder,

- **Highlighting what is not included in this strategy but needs work- education, social services, disability services**
  - throughout this Strategy and for it to be successful it should acknowledge the connection required to other non-health agencies such as Te Tāhuhu o te Mātauranga | the Ministry of Education; Ministry for Pacific Peoples, Ministry of Social Development, Ministry of Youth Development, ACC etc when they are required or impacted throughout the document.

    An example of this would be on where there is reference to the lack of effective care resulting in a strain on health resources but makes no comment on the strain this causes on education services and social development services which are also impacted by lack of effective care.
  - We would like to see recognition that rare disorders are a cross agency issue and that there is significant work required in other areas to achieve equity for people with rare disorders and their whānau. Health cannot be viewed in isolation.
- **Section on increasing and maintaining and future proofing a health workforce needs strengthening.**

  Health, disability, education and other government entities will plan and provide training on rare disorders for health and other professionals and support staff which will include:
  - Identification of existing gaps in the rare disorder workforce and development and implementation of a national rare disorder workforce strategy.
  - Creation of effective rare disorder clinical networks that connect to international research and best practice.
  - Developing and implementing pathways to work with international partners.
  - Equipping and encouraging frontline health and other professionals to consider, investigate and refer for a potential rare disorder diagnosis.
  - Embedding rare disorders in medical training, including diagnosis pathways and undiagnosed ones.
  - Promoting to healthcare professionals the importance of 'thinking rare' when presenting with symptoms.
  - Promoting to healthcare professionals the importance of taking the whole picture of a condition into account when seeing different specialists
  - Strengthening healthcare professionals understanding of tikanga Māori

especially pertaining to whakapapa, human tissues and genetic material.

- **A review period/accountability**

  We expect that this strategy will be revisited and reviewed at some point,

  In addition to monitoring the intended outcomes of the strategy to check on the success of their delivery, it is also important to ensure ongoing evaluation of the strategy direction itself to make sure it remains appropriate. We would expect the strategy to include a plan for the Ministry of Health to do this.

- **As already communicated, quotes from clinicians/experts and more use of the community input that was provided.**
- **There are no references**

  We have previously provided information that could be referenced to back up statements made etc and would expect to see a reference list.

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Questions

- What is the Ministry of Health doing to ensure that the Strategy continues to implementation?
  - When will/has MoH made recommendations to progress with associated budget bids/ incorporate implementation with the funding pot for the pae ora strategies?
  - What engagement has the Ministry of health had with Te Whatu Ora regarding the next steps?
  - What is being done and what are the next steps to get RDS content into the next Government Policy Statement – Health and the NZ Health Strategy?

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- How much consultation has been undertaken with medical schools regarding proposed changes to curricula vs what is already in place/feasibility?
- Can we see the covering report for the strategy going to the Minister

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RARE SUPPORT CENTRE AOTEAROA

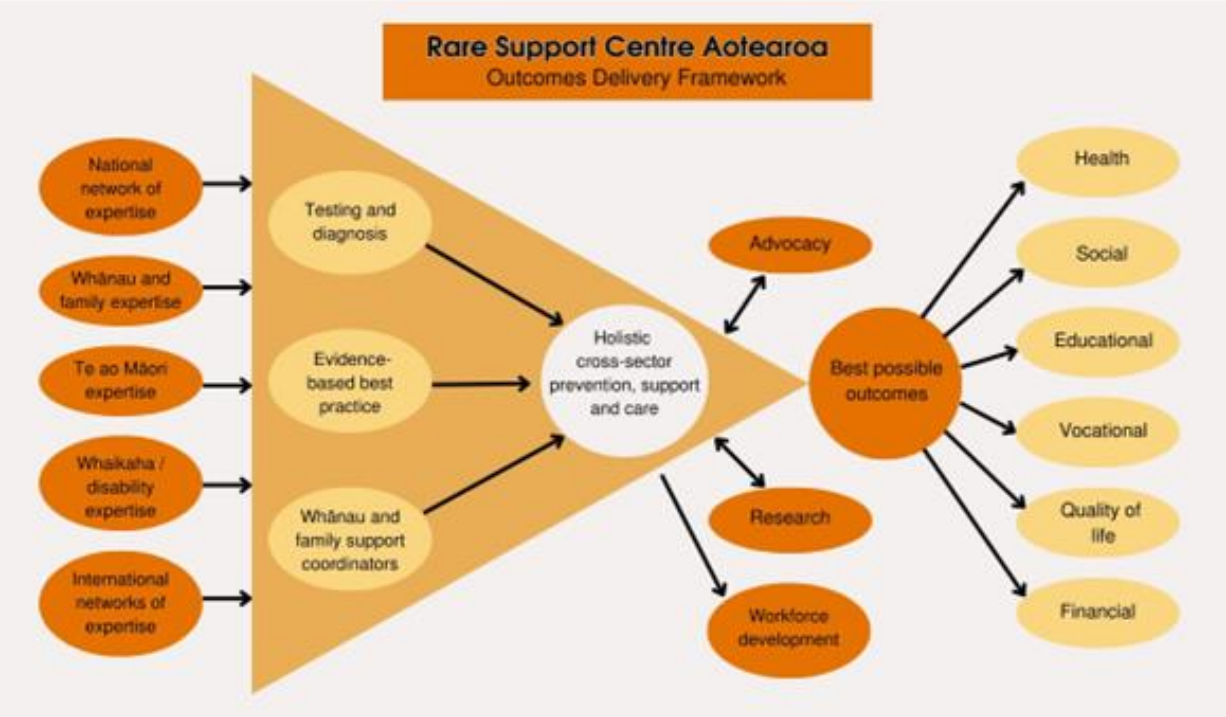
(Rare and Undiagnosed Disorders Centre of Expertise)

"Delivering equitable outcomes and best possible health and wellbeing for people and whānau living with rare and undiagnosed conditions in New Zealand/Aotearoa through world class and world leading health, disability, education, social and other support."

Inequity of outcomes for people with rare disorders is represented by the health and other outcome differences experienced by people who receive a full suite of planned and coordinated evidence-based services and therapies compared with those who don't. These outcome inequities are exacerbated for people with rare disorders who are Māori and/or disabled.

The figure below sets out a framework for achieving improved outcomes for all New Zealanders who have a rare or undiagnosed disorder.

Figure 1: Rare Support Centre Aotearoa Outcomes Delivery Framework



"The absence of a holistic approach made the whole family's devastation unnecessarily more profound and led to 10 years of feeling let down" (Jaime Christmas, Chief Executive, New Zealand Amyloidosis Patients Association)

The Rare Support Centre Aotearoa (RSCA) will comprise networked holistic[1] cross-sector multidisciplinary prevention, support and care services which seek to eliminate these inequities by addressing the service and support gaps in New Zealand's health and other systems. Health will be nested in a larger support centre which addresses a wide set of important life outcomes.

[1] Miller, N. Where is the wraparound care? Interview with Jaime Christmas, Chief Executive, New Zealand Amyloidosis Patients Association. RARE Revolution Magazine, p34. October 2023. Rare Revolution (pagesuite-professional.co.uk)

Drawing on and extending the WHO definition of health[2] the RSCA takes its inspiration from comparable initiatives globally[3] [4] [5], including the Government of Western Australia's Clinical Centre of Expertise for Rare and Undiagnosed Diseases[6] [7] [8], Undiagnosed Diseases Programs[9] and the Undiagnosed Diseaseas Network International[10].

Coordinated from [city] the Centre will be a member of the Global Network for RD[11] and comprise a diverse multidisciplinary virtual team of internationally networked experts distributed across various Aotearoa/New Zealand locations. Typically their contribution to the work of the Centre will be complementary to and an extension of roles they hold with locality based health and other service providers. Clinical experts will be affiliated with Te Whatu Ora's properly resourced Rare Disorders National Clinical Network[12], rather than be fragmented across multiple clinical networks as is the case currently.

Prior to the enactment of the Pae Ora (Healthy Futures) legislation people with rare disorders often reported that they faced obstacles to receiving support because of the "postcode lottery" effects of living in a location where the services they required were unavailable. People are now reporting that services that have since become (together with those that already were) available nationally rather than just locally or regionally are not able to meet the resulting increases in demand, resulting in long waiting times to be seen.

It will be necessary for the RSCA, the extended roles of its networked experts, and allied and complementary services to be appropriately resourced. They will also need to be technologically connected with each other and with patients. Sufficient resources supported by appropriate technology will ensure that people are assessed and supported in a timely manner, especially Māori, people with disabilities, and those who live in rural locations.

Testing and diagnostic service[13]

For those without a definitive diagnosis the RSCA's testing and diagnostic service will be the portal to the RSCA itself and available as a referral centre to any health or other professional with clients or patients presenting with unusual co-presenting symptoms, with or without a suspected diagnosis.

A definitive diagnosis is often an essential component of understanding how to best provide services and therapies to a person with a rare disorder. A definitive diagnosis can be obtained

[2] Health is a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity. https://www.who.int/about/accountability/governance/constitution
[3] Centres of Excellence for Rare Diseases. Rare Diseases UK. 2013. centres-of-excellence.pdf (raredisease.org.uk)
[4] European Reference Networks. https://health.ec.europa.eu/european-reference-networks/overview_en#work
[5] National Expertise Centres for Rare Disorders. Leiden University Medical Centre, Netherlands. https://www.lumc.nl/en/patient-care/polyclinics-nursing-wards-and-expertise-centers/expertisecentra/expertise-center-for-rare-disorders-ecza/
[6] https://pch.health.wa.gov.au/Our-services/Rare-Care-Centre
[7] Rare Care Centre. First Year Impact Report. Feb 2022-2023. Rare-Care-Centre-Impact-Report-Y22-23.pdf (health.wa.gov.au)
[8] Rare Care Clinical Centre of Expertise for Rare and Undiagnosed Diseases Strategic Framework 2022-2023.
[9] Baynam G et al. Initiating an undiagnosed diseases program in the Western Australian public health system. Orphanet Journal of Rare Diseases. 2017.https://ojrd.biomedcentral.com/articles/10.1186/s13023-017-0619-z
[10] Taruscio D et al. The Undiagnosed Diseases Network International: Five years and more! Molecular Genetics and Metabolism Volume 129, Issue 4 (https://pubmed.ncbi.nlm.nih.gov/32033911/)
[11] Baynam G. Rare Care Centre: global needs, local leadership. Sept 2022. Medical Forum. https://mforum.com.au/rare-care-centre-global-needs-local-leadership/
[12] National Clinical Networks – Te Whatu Ora- Health New Zealand *"Joined-up clinical leadership comprised of diverse expert voices to drive system shifts through development of national standards and models of care".*
[13] The importance and increasing ability to achieve a diagnosis is discussed in Baynam G et al. Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. M. Posada de la Paz et al. (eds.), Rare Diseases Epidemiology: Update and Overview, Advances in Experimental Medicine and Biology 1031, chapter 4. 2017. https://doi.org/10.1007/978-3-319-67144-4_4

via non-genetic testing, genetic testing, expert observation or sometimes requires a combination of the three.

The RSCA will offer a culturally appropriate and safe diagnostic counselling and coordination service which will fully inform and directly assist clients and patients to access tests which are consistent with their values, actual and potential life choices, and symptoms, having regard to the results of any previous inconclusive or negative tests. This service will be available until a definitive diagnosis is achieved or until the client or patient withdraws consent to continue, and will be supported by use of AI[14] and other emerging technologies.

Unless there are compelling clinical reasons to do otherwise, and in any continuing absence of a definitive diagnosis, clients and patients who have been accepted into the service will be presumed to have a diagnosis of a rare disorder. Suspecting or believing that a client or patient is imagining or making up their symptoms does not constitute a "compelling clinical reason".

Care and support

The RSCA's cross-sector and multidisciplinary care and support service will:

- Accept rare disorder referrals from the RSCA's testing and diagnostic service, and other credentialed diagnostic and screening services (such as the national newborn screening service)
- Develop and maintain evidence-based world class, world leading and globally connected standards of holistic best practice cross-sector care and support for **specific and identifiable** rare disorders
- Develop and maintain evidence-based world class, and world leading globally connected standards of holistic best practice cross-sector care and support for **undiagnosed** rare disorders[15] [16]
- Provide expertise and guidance for general practitioners, specialists, other clinicians and other professionals in how to support their patients and clients in accordance with best practice standards
- Develop and implement measurement of appropriate outcome measures for patients and clients focusing on patient experience measures and disease-appropriate outcome measures
- Facilitate belonging to relevant national and international rare disease support groups
- Support workforce development through:
    - provision of curriculum development and rare disorders training to clinical and other training schools and entities, continuing professional development providers, other professional development agencies and service providers
    - identification of workforce gaps including in the supply of skilled staff to meet demand and requirements for new subject matter experts to harness and implement new knowledge and technologies, and facilitate closing these gaps.

[14] Eg Cliniface. https://cliniface.org/
[15] Baynam G et al. Initiating an undiagnosed diseases program in the Western Australian public health system. Orphanet Journal of Rare Diseases. 2017. https://ojrd.biomedcentral.com/articles/10.1186/s13023-017-0619-z
[16] Taruscio D et al. The Undiagnosed Diseases Network International: Five years and more! Molecular Genetics and Metabolism Volume 129, Issue 4, April 2020, Pages 243-254 (https://pubmed.ncbi.nlm.nih.gov/32033911/)

Service coordination

The RSCA will engage a team of professionally trained service coordinators who will acknowledge whanau and family as their own best experts, and support them as personal advocates to navigate the health and other systems in a timely way, ensuring that all appropriate services are accessed, and appointments are made and kept, in accordance with best practice individual care and support plans

Research and clinical trials

The RSCA will identify gaps in knowledge and expertise and engage with health and wellbeing researchers, research funders, service professionals and international rare disorders research networks[17] to have those gaps filled, and the resulting knowledge translated into best practice standards of support. For 95% of rare disorders clinical trials are the only hope of a disorder specific prognosis altering drug therapy[18]

Advocacy and awareness

The RSCA will identify gaps in service provision and support, both internally and externally in the health and other sectors, and will partner with rare disorders support groups through the Rare Disorders New Zealand peak body to advocate both for required system changes and professional and public awareness of the RSCA's services.

[17] Eg International Rare Diseases Research Consortium (IRDiRC) (Who we are – IRDiRC) and The European Joint Programme on Rare Diseases (EJP RD) (EJP RD – European Joint Programme on Rare Diseases)
[18] Baynam G. Personal communication. 7 November 2023.