Te Kāwanatanga o Aotearoa
New Zealand Government

MINISTRY OF
HEALTH
MANATŪ HAUORA

Aotearoa New Zealand Rare Disorders Strategy

Rare voices and aspirations

Te Rautaki o Aotearoa e Pā ana ki ngā Mate Mokorea

Ngā reo mokorea me ngā wawata -

Acknowledgements

This supporting document to the *Aotearoa New Zealand Rare Disorders Strategy* outlines how we achieved the strategy with support from a broad range of expertise across the health and other sectors, as well as from lived experiences and rare voices. This document is linked with both the strategy and the supporting document *Aotearoa New Zealand Rare Disorders Strategy: Context and evidence*. We recommend reading this document along with the other two.

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Glossary

Disability – Long-term physical, mental, intellectual or sensory impairments that, in interaction with various barriers, may hinder the full and effective participation of disabled people in society on an equal basis with others (United Nations Convention on the Rights of Persons with Disabilities 2006).

Ethnic communities – People who identify their ethnicity as African, Asian, Continental European, Latin American or Middle Eastern. Ethnic communities also include former refugees, asylum seekers, new and temporary migrants, long-term settlers and multigenerational New Zealanders.

Karakia – Prayer, grace or blessing.

Manaakitanga – Hospitality and generosity.

Pae ora – A holistic concept of health and wellbeing. This means it considers all the broad factors and contexts that influence people's health and wellbeing throughout their lives. Pae ora envisages a platform on which Māori can live with good health and wellbeing in an environment that supports Māori to flourish and thrive. It includes three interconnected elements:

- **whānau ora** – a fundamental philosophy for creating strong, healthy and empowered whānau
- **mauri ora** – expressing an aspiration to shift the mauri (life force) of a person from one that is languishing to one that is flourishing
- **waiora** – acknowledging the importance of Māori connections to whenua as part of the environments where we live and belong – and the significant impact of these connections on the health and wellbeing of individuals, whānau, hapū, iwi and Māori communities.

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 2,000 people in Aotearoa New Zealand.

Rongoā Māori and mirimiri – A wide range of traditional Māori healing modalities, incorporating holistic aspects of mind, body and spirit.

Ultra-rare disorder – an ultra-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 50,000 people in Aotearoa New Zealand.

Whānau – Often translated as 'family', but its meaning is more complex. Whānau are part of an interconnected world of tribe and subtribe, of the living and the dead, based on whakapapa.

Ngā kai o roto - Contents

Kupu whakataki – Introduction

New Zealanders living with rare disorders

People and whānau living with rare disorders have provided key input to the Aotearoa New Zealand Rare Disorders Strategy. Other important contributors have been the wider rare disorders community and supporters from across the health and disability sectors.

Vision for the strategy

All people and their whānau living with rare disorders share in pae ora (healthy futures) and live fulfilling lives

Pae ora is a holistic concept of health and wellbeing. It considers all the broad factors and contexts that influence people's health and wellbeing throughout their lives.

Sharing in pae ora (healthy futures) means that people and their whānau living with rare disorders:

- are part of healthy and inclusive communities
- live in environments that enhance quality of life and promote health and wellbeing
- have the support of an equitable and accessible health system.

Achieving this vision requires working with people and their whānau living with rare disorders; iwi, hapū and Māori communities; Pacific communities; and other communities our health system serves. It requires collective efforts across health entities and different groups of workers, and the wider organisations and agencies that contribute to the health and wellbeing of people and their whānau living with rare disorders.

Who the strategy is for

The strategy is for all who identify as or provide support or care for people and their whānau living with rare disorders in Aotearoa New Zealand. It is also for those who may not see themselves as living with a rare disorder or those who do not yet have a diagnosis, but can see their lived experiences reflected in the strategy.

For people and their whānau living with rare disorders

All people deserve to be respected, listened to, heard and supported when they seek health care. People and their whānau living with a rare disorder often require a higher level of support with needs that are complex, unusual and often unclear.

Whānau and wider support networks play a vital role in supporting and making real the independence and aspirations for people living with rare disorders. We have used the term 'people and their whānau living with rare disorders' throughout the strategy and supporting documents to represent this collective approach to the health of people living with rare disorders. The strategy also recognises the overlap between rare disorders and disability.

The strategy outlines how the health system, health practitioners, support organisations and others can work to support people and their whānau living with rare disorders. This support may include providing information and resources, or reducing wait times for tests, diagnosis or further assistance. Whether a rare disorder is identified or not, advice and support can help people and their whānau to live well and can benefit child development.

People living with rare disorders have much to contribute to our communities, economy and society. Reducing the negative impacts of rare disorders enables and enhances these contributions.

For whānau Māori living with rare disorders

People with rare disorders have significantly poorer health outcomes than the whole population and the impact is often even greater for whānau Māori. A report on the experiences and voices of whānau Māori living with rare disorders has provided important input to the strategy and this voices document.[1]

Whānau Māori can face barriers to culturally competent and safe health services, often needing to fight against discrimination, unconscious bias, and racism, and access to any available services may be difficult. As a result, Māori often feel isolated within the health system. Whānau spoke consistently of fighting to access information relating to diagnosis, funding entitlements and support pathways 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 being treated with disrespect. They have wanted to contribute to the solutions to meet their health needs.

Many whānau, particularly parents of tamariki living with a rare disorder, feel lonely and isolated. Whānau often felt they had to become the specialist of their rare disorder while health professionals were not open to listening to the knowledge whānau held in relation to their health.

In our kōrero with whānau, we found that services that are whānau-centred and embedded within te ao Māori would have a positive impact on their own and others' journeys across the health system.

Many whānau Māori found a medical term such as 'rare disorder' to be alienating. Parents of tamariki living with a rare disorder want their tamariki to be acknowledged as the taonga that they are, rather than being identified by a medical term such as 'rare disorder'. Within te ao Māori, differences including impairments are embraced and celebrated.

For health practitioners and care workers

Health practitioners and care workers want to help people and their whānau living with rare disorders to live well and be able to contribute to their communities. The strategy outlines ways the health system can build the capability of health practitioners and care workers to better support people and their whānau living with a rare disorder. This capability building is in two main areas: relational and cultural competence; and the ability to access knowledge.

Having relational and cultural competence is necessary for people and their whānau to feel comfortable that the health workforce will hear and support them. All people working in the health system need skills in building strong relationships and showing manaakitanga, and in relating to and listening to people from all communities. Creating culturally safe and welcoming environments involves expanding the range of cultures and communities represented in the workforce, as well as building the skills of members already in the workforce.

Over the course of their career, an individual health practitioner may treat only a few people living with a rare disorder, if any. Many health professionals may not have the necessary knowledge to identify or treat people with rare disorders. Equipping health professionals and care workers with the knowledge, tools and specialties to support people with a rare disorder will reduce the time taken to diagnose and treat people with a rare disorder.

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[1] Te Aka Whai Ora. 2023. Whānau Voice 2023 Summary: Whānau Māori with a rare disorder and those who care for them. Wellington: Te Aka Whai Ora

For the health system

Many people and their whānau living with rare disorders may have more complex needs and struggle to navigate the health system to get the care they need. Disabled people living with rare disorders and those living in rural and other diverse communities (including Pacific peoples) may experience further difficulties in accessing support from the health system.

The strategy aims to achieve a better-connected health system that has the technology and infrastructure required for people and their whānau with rare disorders to receive the care they need.

A health system that is connected up and well equipped to treat people and their whānau living with rare disorders also has benefits for health users more generally.

For Aotearoa New Zealand communities

Most people may not have heard of rare disorders. Some may have heard of a particular disorder but do not know that the disorder is rare. Greater awareness is one step towards people being able to support and include people and their whānau living with rare disorders.

All community members benefit when people and their whānau with rare disorders can live well and participate in their communities to the full.

How rare voices have shaped the strategy and priorities for action

People have said the strategy needs to work towards the following aspirations.

- People and their whānau living with rare disorders can have the best possible health and live the best possible lives.
- Whānau Māori living with rare disorders receive resources and tools that enable the exercise of tino rangatiratanga.
- Māori, Pacific peoples and disabled people living with rare disorders, and those living in rural and other diverse communities share in pae ora (healthy futures).
- The health workforce is able to deliver quality care for people and their whānau living with rare disorders.
- The Aotearoa New Zealand health system works fairly to promote health and addresses health needs for all, including for those with rare or unknown disorders.

The five priorities in the strategy describe the key changes that will have the greatest impact. The priorities outline where action is most needed to drive progress that will improve health and wellbeing for people and their whānau living with rare disorders. The key changes under each priority are based on:

- what people and their whānau living with rare disorders have said matters most
- what people working across the health system have said will achieve the most.

The next section outlines, for each priority, what we heard and why it matters to improve rare disorders outcomes. It also sets out the aspirations people have for a future health system that supports people and whānau living with rare disorders to share in pae ora (healthy futures).

Ngā aronga tōmua - Priority areas

1. Why gearing the system for quality and timely care is a priority

What we've heard

People and their whānau living with rare disorders often need help from a range of services across the health system. These services need to be connected up and easy to navigate, as well as to operate consistently throughout the country.

Practitioners have highlighted the need for more joined-up care across the health system. People working in hospital care, such as neonatal intensive care, may diagnose a rare disorder and give the news to whānau without having any way to link in to supports, counselling or other follow-up care.

General practitioners and primary health services may find it difficult to get help from other parts of the system. Accepted practices and criteria to refer people for specialist assessments or diagnostic tests may not work when practitioners have not yet established a particular suspected disorder or when they are not sure which specialty the referral is for.

Some voices from the rare disorders community that have informed priority 1

> I would like to see more Māori representation in the hospital...Have someone who can korero Māori and can also represent the tikanga of te ao Māori.
>
> **Rare Disorders Strategy engagement participant [Whānau Māori Voices Engagement]**

Why it matters

While supporting health practitioners and care workers to provide quality care is important, how the wider health system is organised and coordinated also matters if we are to improve rare disorders outcomes. Rare disorders often come with complex needs that involve many parts of the health system. Quality, timely, safe and life-affirming care needs to be accessible. The system needs to accommodate people and their whānau with all needs, at all stages of a health journey and even with the rarest conditions.

The following three areas of system improvement will make a big difference to rare disorders outcomes:

- being connected across different parts of the system and providing navigation support
- early adoption of more effective, efficient and safe technologies
- having ways to prioritise access and investment across services, products and infrastructure that work for rare needs.

> 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.
>
> **Rare Disorders Strategy engagement participant [Rare Disorders Webinar Engagement]**

Aspirations for the future

The system will be connected up around people with rare disorders

No matter where in Aotearoa New Zealand people live, expert input into assessment, diagnosis and care will be accessible. When people need to travel for this input or to visit unfamiliar services, they will have support to do so.

Health funders and planners, practitioners and providers will recognise the many common features in the types and complexity of rare disorders 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.

The system will have established ways of providing timely access to different specialties and services where these contribute to diagnosis or care. How the system works for people with rare disorders will be consistent with other areas in the sector so it will be easily understood.

A recognised 'home' for rare disorders leadership across the system will be well established, with strong links into many different service lines and networks. Efforts to improve system responsiveness for people with rare disorders will be known and visible. People in any part of the health system will be able to find information about rare disorders and will know where to go for answers to their questions.

> “The experience I had in the hospital when I was diagnosed with [condition] left me feeling helpless.
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> **Rare Disorders Strategy engagement participant [Rare Disorders Webinar Engagement**

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. As new technologies are developed into treatments, many will first be used to treat rare disorders, as we are seeing with Clustered Regularly Interspaced Short Palindromic Repeats (CRISPR) based treatments now.

Aotearoa New Zealand will continue to identify and do the groundwork for new technologies and digital capabilities so that we can prepare for early adoption. Initial work will continue to be concerned with ensuring the safety and fair application of the innovations, before conducting full development or affordability assessments. Establishing both safety and value for money will allow early adoption of promising new capabilities.

> “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 rare disorders space to have a 'suspected' it needs to have a code.
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> **Rare Disorders Strategy engagement participant [Clinical Hui]**

Note: ACC = Accident Compensation Corporation.

> “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 time was wasted.
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> **Rare Disorders Strategy engagement participant [Rare Disorders Webinar Engagement]**

> “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 research! You cannot research something if you don’t know what to look for.
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> **Rare Disorders Strategy engagement participant [Rare Disorders Support Group Ministry of Health Survey]**

> “[Support Group] would encourage protocols to be written by Health NZ, be audited by the NZ Endocrine Society and then applied to hospitals throughout NZ.
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> **Rare Disorders Strategy engagement participant [Rare Disorders Support Group Ministry of Health Survey]**

> “Differences in the level of specialisation and clinical resources means healthcare is experienced differently for people across the country.
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> **Rare Disorders Strategy engagement participant [Rare Disorders Webinar Engagement]**

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

The nature of rare disorders makes it hard to gain the evidence required to evaluate and assess medicines, diagnostics and other health services and treatments. Costs of new medicines are increasing, and 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 Aotearoa New Zealand, most will be developed and patented elsewhere in the world.

Rare disorders medicines are often significantly more expensive than other medicines. Developing and producing them often involves special technology, infrastructure and expertise. It may be affordable for producers to cover these costs for a rare disorder treatment only when they see it as a prototype for a wider range of treatments with bigger market potential.

At any given time, it will not be possible to provide certain diagnostics, medicines, devices and other treatments safely or affordably in Aotearoa New Zealand. Supporting people to access such treatments overseas will be considered, yet affordability will remain a significant issue in the future.

However, processes to consider new diagnostics, treatments, services and infrastructure will become faster and fairer over time. They will account for all needs, including rare, 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). Ways of supporting people in exceptional circumstances to access tests and treatments that are not generally available, may be highly expensive or require travel to other parts of the country or the world, will continue to expand.

2. Why learning and sustaining progress is a priority

What we've heard

People want rare disorders to be visible in the health system. They want to see improvements to services and outcomes, and for health entities and providers to have the information that tells them how well they are doing with providing fair access to safe, quality and culturally responsive diagnosis and care.

For many rare disorders, treatments or management approaches may be limited or unavailable. Participating in clinical research, including clinical trials, may offer the best hope for improving some people's health or life course. A number of people living with rare disorders have said that the choice to enrol in a clinical trial is important to them and relevant trials should automatically offer people living with rare disorders the opportunity to participate. People have also said that genetic counselling should be available to extended whānau members when an inherited rare disorder is identified. Being able to make choices about such opportunities is important.

Some voices from the rare disorders community that have informed priority 2

> "The strategy is an opportunity for impact, sustainability, scale, and equity through addressing the common challenges that crosscut all rare disorders, this includes diagnosis access to expertise, treatment and care, and mental health and wellbeing, but with great emphasis on access to registries, research, and clinical trials.
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> **Rare Disorders Strategy engagement participant [Clinical Hui]**

Why it matters

To continue to improve over time, the health system needs ways to measure and track progress. Those planning, delivering and using health care need to be able to see what is working, where improvements are being made and what more needs to be done.

We need indicators of system performance for rare disorders, and decision-makers should consider where opportunities for improvement lie. Given the close links with the disability community, rare disorders indicators are needed within disability indicators as well as within health indicators. People and their whānau living with rare disorders need to contribute to the development of these indicators.

Aotearoa New Zealand is unique in being able to gather knowledge of Māori living with rare disorders, and to contribute to Pacific knowledge. We have a key opportunity to design and monitor research and services collaboratively with Māori, Pacific peoples and other rare disorders communities.

Research into service models and approaches for rare disorders is important to develop effective care that fits with our population and where people live 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 is otherwise available. Clinical research is also important to grow the capacity and capability of our research and development industry, and to better prepare our health services to adopt new advances.

A culture of continuous learning is a priority for the future of the health system and is underpinned by the New Zealand Health Strategy 2023. The Provisional Health of Disabled People Strategy also outlines the importance of robust health system data and evidence.

> 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 rare disorders space to have a ‘suspected’ it needs to have a code.
>
> **Rare Disorders Strategy engagement participant [Clinical Hui]**

> 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.
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> **Rare Disorders Strategy engagement participant [Pae Ora Webinar Engagement]**

> Being able to capture those in the system that are currently being missed.
>
> **Rare Disorders Strategy engagement participant [Rare Disorders Webinar Engagement]**

> We need to start measuring rare disorders data so we can understand the scope of the issue and provide better levels of care.
>
> **Rare Disorders Strategy engagement participant [Rare Disorders Webinar Engagement]**

> [There needs to be] collection of data to better inform care and supports, improve access to expertise/sharing of information for all – [there are] too many inconsistencies.
>
> **Rare Disorders Strategy engagement participant [Pae Ora Webinar Engagement]**

> There is a paralysis in the absence [of having] good information – registries are a great way to address this, not just in terms of the potential for clinical trials or knowing data, but to provide insights into good decision-making. This was done in my previous role in [job position and organisation], the use of pregnancy registries and evidence would determine the level of significance of the data to then advise clinicians to guide them towards informed decision-making.
>
> **Rare Disorders Strategy engagement participant [Clinical Hui]**

> [Research institute] has worked with the Integrated Data Infrastructure (IDI) through Statistics NZ looking for comorbidities (as well as other conditions). But due to the lack of coding infrastructure for rare disorders the IDI can’t be utilised as people are often unidentifiable – better infrastructure would enable the study of the rare disorders population.
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> **Rare Disorders Strategy engagement participant [Clinical Hui]v**

Aspirations for the future

Our system will collect information and data on rare disorders and make them visible

Patient records will capture information to support care of people with rare disorders or possible rare disorders. Medical coding will keep up to date with changes in international classifications 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. Discussions and decisions about these system foundation elements will occur with rare voices, as well as other consumer, cultural and minority voices, at the table. Citizens will have a degree of choice in their data and will have confidence in the in-built protections.

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

Services will routinely collect and report information on rare disorders. People across the health system, including service users, planners and providers, will help guide what information is collected and how it is used. It will be possible to match a variety of data 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 Aotearoa New Zealand results with those in other countries, or in some cases results across different parts of the country.

Assessments of our health as a nation, and how well the Aotearoa New Zealand health system is performing, will include specific information on rare disorders and the people living with them. They will look at trends in health status over time, using averages and compounded data, such as life expectancy and health expectancy. These assessments will also explore trends in experiences of the health system in terms of, for example, wait times for diagnostics, assessments or treatments; how accessible and affordable services are; and how confident people are in their practitioners and services.

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

Māori will design and use systems to assess and track progress in health and health care for whānau Māori living with rare disorders. Reports in partnership with Māori will inform how we assess equity of outcomes and health system delivery, and how we monitor programmes to improve these. Working in partnership will help us to continuously improve outcomes.

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

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

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

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

3. Why equipping the health workforce for quality rare disorders care is a priority

What we've heard

Many people and whānau have expressed frustration with how long it takes to get a rare disorder diagnosis. Multiple practitioners and services may be involved, with few or none of them knowing much about rare disorders in general or about a particular rare disorder. People and their whānau feel they need to 'fight' for attention or to be heard.

When a practitioner suspects a particular disorder, support groups may have better access to information than the practitioner does through the usual guidance channels. People and their whānau living with rare disorders shared how it is important for health and social services students to learn about rare disorders as part of their education.

Whānau Māori want to see greater Māori representation in the health and social workforces that whānau interact with across their rare disorders journey. They want their health care journey to recognise and value a worldview from te ao Māori, and would like practitioners and health workers to inform them of the options for meeting their cultural as well as health needs. In particular, whānau Māori have often found hospital care to be alienating. They place high value on having a kuia or health worker available for support through such experiences.

“ Some voices from the rare disorders community that have informed priority 3

Why it matters

Health practitioners and care workers are highly 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 can make a sizeable difference to the experiences of people and their whānau living with rare or unknown disorders.

This priority aligns with the wider health system focus on supporting the development of sustainable, diverse, skilled and confident workers for the future of the health system. The Pae Ora Strategies underpin this focus.

> “ We're in Ashburton and we have a rural medical school here. For the last four years we have been working with doctors and medical students at the school for my daughter's neurological rare disorder. The students are always very intrigued and want to learn. This experience will influence how they treat rare in the future.
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> **Rare Disorders Strategy engagement Participant**

> “ Clinicians need a place to go [for information and guidance], this is why so many of us have fallen between the cracks.
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> **Rare Disorders Strategy engagement participant**

> At least half of the patients I treat will have a rare disorder and the biggest barrier I face is that there are only nine clinicians including myself that cover the entire country: two in the South Island, one in Wellington and six in Auckland. These geographical barriers add to already huge waitlists.
>
> **Rare Disorders Strategy clinical engagement participant**

> Improved knowledge of local paediatricians has helped with my quality of life.
>
> **Rare Disorders Strategy engagement participant**

> My general practitioner took a proactive approach to making referrals that led to my diagnosis.
>
> **Rare Disorders Strategy engagement participant**

> The other thing that's important to me is having access to doctors that understand the disorder you know and having that knowledge at their fingertips. That's important because otherwise that's what helps to create misdiagnosis and people waiting years before they're actually being diagnosed.
>
> **Rare Disorders Strategy whānau Māori engagement participant**

> There is often constructive curiosity among clinicians about my rare disorder. I'd love to see that fostered through further education about rare.
>
> **Rare Disorders Strategy engagement participant**

> What is working well is the emergency health needs, because we are within a 15-minute drive of Starship Hospital emergency, and they are familiar with [child] condition. If there is a health emergency, we are able to have [child] assessed by people familiar with [child] records and condition quickly without having to re-explain [child] history.
>
> **Rare Disorders Strategy engagement participant**

> Now that our local general practitioner has been taken over by … a Māori organisation, I can now walk in and there's Māori nurses and doctors. I honestly feel so much more comfortable, and they make us feel like we're at home, you know, and it's a just totally different surgery and I love walking in there and them helping [tamaiti] has just been amazing.
>
> **Rare Disorders Strategy whānau Māori engagement participant**

Aspirations for the future

Health practitioners and care workers will 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 are developed.

Ongoing learning for health practitioners will include integrated examples of rare disorders, and options for more detailed supported learning in a range of rare disorders and related topics.

Care 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 opportunities to learn about rare disorders.

Health practitioners and care workers will be able to support people and their whānau who may be living with rare disorders

Students of health disciplines will engage in tikanga Māori practices in health and understand 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 opportunities to learn how to support 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, along with referral, diagnostic and care pathways, will support care for a growing number of rare disorders. After Aotearoa New Zealand authorities have approved these tools, they will be adapted for different localities or situations where necessary. Health practitioners will be able to access the tools in the standard approved collections that they most often use. Because the tools will be integrated with patient management systems and electronic health records, they will also be automatically visible.

Specialist help, advice and backup will be available for practitioners at 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 their whānau living with rare disorders. Experts will provide this backup support in timeframes that are responsive to needs.

Community health providers will benefit from a diverse range of expert input, including from specialists, therapists, health promoters, cultural advisors and people with lived experience. Specialist practitioners will be able to receive expert input from practitioners outside and within their specialities who have experience of the relevant rare disorder. Practitioners will be able to inform people and whānau about support groups – both specific to their rare disorder and with a more general focus – as an additional resource. They will also provide the opportunity for people and whānau to form connections with others. International input will be available for rare disorders if appropriate expertise is not available in Aotearoa New Zealand.

4. Why giving voice to people and their whānau living with rare disorders is a priority

What we've heard

People and their whānau living with rare disorders want to be heard and seen in the health system. People and their whānau want practitioners to relate to and respond to them as people first, rather than as bearers of health conditions, needs or problems; to feel that each of us matters, and is believed and respected. People and their whānau would also like practitioners to have time to provide information and support for them to make their own decisions and decisions for the health and care of their whānau. People want to feel welcomed and at ease when they access health care.

Whānau Māori living with rare disorders say they feel respected when health services are whānau-centred and delivered with te ao Māori in mind. They feel welcomed and at ease when the health care team allows space for introductions, making them feel comfortable and having mihi and karakia, and when the team includes Māori workers. Parents of tamariki living with a rare disorder want their tamariki to be acknowledged as the taonga that they are, rather than being identified by a medical term like 'rare disorder'.

Why it matters

People and their whānau living with rare disorders are the best informants about their own experience of health and ill health. Being listened to and understood in a welcoming and respectful environment makes people feel significantly more comfortable with and able to trust services and practitioners. Building care plans together makes a big difference to health outcomes in the longer term.

Equally important is that the voices of people and their whānau living with rare disorders influence how health care is provided at all levels – policy, design, commissioning, service delivery, research, improvement programmes, and progress and outcome monitoring.

This is a wider health system priority highlighted in the Pae Ora Strategies, including the New Zealand Health Strategy, Pae Tū: Hauora Māori Strategy, Te Mana Ola: The Pacific Health Strategy and the Provisional Health of Disabled People Strategy. How health practitioners and services respond to people and their whānau is important to all New Zealanders – and doubly important to those living with rare disorders, many of whom may already feel isolated and unsupported.

Where a system considers rare disorders and integrates them into the way it is set up, the starting point for people interacting with different parts of the system will be that services meet them with a basic level of understanding.

Some voices from the rare disorders community that have informed priority 4

> I am unique and special – not rare.
>
> –
>
> **Rare Disorders Strategy whānau Māori engagement participant**

> Being heard, everything is so transactional. Needs to be more about knowing her [tamaiti].
>
> –
>
> **Rare Disorders Strategy whānau Māori engagement participant**

> “I’ve got lovely doctors… my specialist is lovely, a joker and good friend. Your stay in hospital is always better if you have good nursing staff and good doctors. Someone who is passionate about their job. There are so many doctors who are overworked and underpaid, and it shows. 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.
>
> **Rare Disorders Strategy whānau Māori engagement participant**

> “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.
>
> **Rare Disorders Strategy whānau Māori engagement participant**

> “It’s changed our lives completely. I’ve gone from someone that was really like, I don’t know, confident, you know, hardworking, making a living, you know. We really were very social people… our lives were completely different.
>
> **Rare Disorders Strategy whānau Māori engagement participant**

> “Now that our local general practitioner has been taken over by … a Māori organisation, I can now walk in and there’s Māori nurses and doctors. I honestly feel so much more comfortable, and they make us feel like we’re at home, you know, and it’s a just totally different surgery and I love walking in there and them helping [tamaiti] has just been amazing.
>
> **Rare Disorders Strategy whānau Māori engagement participant**

> “You have to be your own advocate and when you are battling and already exhausted this is hard to do.
>
> **Rare Disorders Strategy engagement participant**

> “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 … like that but then I think about [tamaiti] 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 already has delayed development, she doesn’t need delayed services. If I didn’t push for everything she would have fallen through the cracks.
>
> **Rare Disorders Strategy whānau Māori engagement participant**

Aspirations for the future

People and their whānau living with rare disorders will be the most important contributors to their health and care

Placing people and their whānau at the heart of health interactions will ensure that what is most important to them will drive the priorities of health providers and the system. Services will adapt to the cultures, circumstances, preferences and choices of people and their whānau living with rare disorders. A holistic view of wellbeing will underpin all services, which will support health and wellbeing even when no diagnosis or treatment is available. People and their whānau with rare or unknown disorders will feel welcome and supported to improve their health outcomes. This support includes clear explanations of recommended standards of clinical care, how this translates to individual situations, what to expect and how to best manage a disorder.

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

People will have access to their own health records and will be welcome to bring in other support and expertise so that they have autonomy when making decisions about their own care. Systems will be set up to accommodate rare disorders. When people begin interacting with different parts of the system, the starting point will be that services meet them with a basic level of understanding and respect. Services will also meet whānau needs so they can nurture whānau members whether or not they have a rare disorder.

> “I just want to say that it’ll be good to have a rare disorders voice.
>
> –
>
> **Rare Disorders Strategy engagement participant**

Voices of people and their whānau living with rare disorders will be heard clearly across the health system

The health system will hear voices of people and their whānau living with rare disorders at all levels and across all functions. Rare voices, including whānau Māori voices, will be present in conversations on policy, design, commissioning and service delivery. Rare voices will inform progress and outcome monitoring, iwi Māori partnerships, research and improvement programmes, and a strategic overview of New Zealanders’ health and wellbeing.

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

The health sector will work with diverse communities to build, maintain and enable strong foundations for health and wellbeing that include all community members. Health promoters, occupational therapists, physiotherapists and others will support communities to include people and their whānau living with rare disorders. People and their whānau will have support to participate in everyday activities and maintain connections that promote wellbeing.

> “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.
>
> –
>
> **Rare Disorders Strategy engagement participant**

5. Why joining up internationally to achieve more is a priority

What we’ve heard

People and their whānau living with rare disorders experience both isolation and mental health challenges at high rates. Being able to connect and share knowledge with people, groups, specialists and experts across Aotearoa New Zealand and overseas can help New Zealanders with rare disorders to learn more and feel less isolated.

Many people have emphasised how difficult it is for one small country to have all the information it needs about rare disorders. This is especially so for disorders that are ultra-rare or still being identified, disorders about which knowledge is growing fast, and disorders with active research programmes under way.

Why it matters

As a small country, Aotearoa New Zealand cannot hold expert knowledge of every rare disorder on its own. Considerable attention to rare disorders is building internationally. Some countries are combining their efforts by sharing their knowledge and expertise.

We can learn much from, and contribute to, collective efforts to improve outcomes for people and their whānau living with rare disorders. By joining in international collaborations, globally and regionally, we can share in clinical knowledge, research and technologies to improve outcomes for people and their whānau living with rare disorders.

Some voices from the rare disorders community that have informed priority 5

> We need to work with and learn from other countries that are ahead of us.
>
> **Rare Disorders Strategy engagement participant [Rare Disorders Webinar Engagement]**

> [There is an] 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 [disorder]. Australia has a nurse that provides advice over the phone or online. [The] United States, Australia and the United Kingdom have [disorder] clinics in some areas.
>
> **Rare Disorders Strategy engagement participant [Rare Disorders Support Group Ministry of Health Survey]**

> New Zealand should reach out to international partners such as Australia to improve knowledge, delivery and access to medication and treatment options. Much of the work has been done already overseas and we utilise that to upskill here.
>
> **Rare Disorders Strategy engagement participant [Rare Disorders Support Group Ministry of Health Survey]**

> Overseas, there are clinicians with a lot of experience and knowledge about [disorder] who are willing to be consulted for free by their New Zealand counterparts, and there are freely accessible international [disorder] Best Practice Guidelines covering many different aspects of [disorder] care and treatment. In New Zealand it is very difficult to find a clinician who knows everything about [disorder] and yet we very often struggle to get the local clinicians to take any notice of the international experts or the guidelines.
>
> **Rare Disorders Strategy engagement participant [Rare Disorders Support Group Ministry of Health Survey]**

> [We need] a pathway to international expertise for rare disorders that are very rarely treated here in New Zealand.
>
> **Rare Disorders Strategy engagement participant [Pae Ora Webinar Engagement]**

> We need the ability to leverage off international data including the US Food and Drug Administration (FDA), National Institute for Health and Care Excellence (NICE), and Therapeutic Goods Administration (TGA).
>
> **Rare Disorders Strategy engagement participant [Pae Ora Webinar Engagement]**

Aspirations for the future

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

Aotearoa New Zealand health and research leaders and organisations will participate in a range of international partnerships that have a focus on rare disorders. The partnerships and contributions will be across a wide range of topic areas, including research, outcomes, systems, affordability and equity. People with rare disorders will also be participants in and will inform the partnerships.

Aotearoa 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 reporting information to global monitors, as well as contributing knowledge from research, evaluation and health monitoring.

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

Aotearoa New Zealand Indigenous health and research leaders and organisations will be participants in international Indigenous health partnerships that have a focus on rare disorders. Whānau Māori and tāngata whaikaha will be active participants and inform the partnership goals and work programmes.

International collaborators will work on faster uptake of new technologies and treatments to enhance care provision

Aotearoa New Zealand will receive expert input to inform care, especially for very rare disorders and in areas with new developments. Our experts will contribute their experience where possible.

Aotearoa 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 the affordability of advances in rare disorders.

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

> “There is more data available in Australia and our specialists should be able to access those resources and collaborate.
>
> **Rare Disorders Strategy engagement participant [Pae Ora Webinar Engagement]**