Te Kāwanatanga o Aotearoa
New Zealand Government

MINISTRY OF
HEALTH
MANATŪ HAUORA

Aotearoa New Zealand Rare Disorders Strategy

Context and evidence

Te Rautaki o Aotearoa e Pā ana ki ngā Mate Mokorea

Te horopaki me ngā taunakitanga

Acknowledgements

This supporting document to the *Aotearoa New Zealand Rare Disorders Strategy* outlines the context in which the strategy was developed and will operate, along with the evidence used in developing it. This information is linked with both the strategy and the supporting document *Aotearoa New Zealand Rare Disorders Strategy: Rare voices and aspirations.* 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

Te horopaki mō te whanaketanga o te rautaki - Context for developing the strategy

Purpose of the strategy

The Aotearoa New Zealand Rare Disorders Strategy sets out the direction for the health system to better support people and their whānau living with rare disorders. It provides a framework and long-term priorities that will guide health entities in improving health and wellbeing outcomes for people and their whānau with rare disorders over the decade 2024 to 2034.

The strategy responds to longstanding calls for a strategy from the rare disorders community. It also acts on the Government's response to the 2022 independent review of Pharmac to:

- improve the lives of people and their whānau living with rare disorders
- make it easier for people, practitioners and organisations to get the information and support that would help
- develop a strategy that leads to better, more timely services and more equitable support and outcomes for people and their whānau living with rare disorders.

The strategy will be successful if:

- people and their 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
- those involved in designing, commissioning or providing new policies or services routinely consider the needs of people with rare disorders.

Key terms

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.

An ultra-rare disorder is a medical condition that affects fewer than or equal to 1 in 50,000 people in Aotearoa New Zealand.

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

- A disease is a medical condition that has a clear cause and can be diagnosed by a health care provider.
- A disorder is a medical condition that has a specific pattern of clinical signs, symptoms and findings that might indicate that a specific disease is possible but there is not enough clinical evidence for diagnosis.
- A condition is an abnormal state of health that interferes with the usual activities or feeling of wellbeing.
- A syndrome is a group of signs and symptoms that occur together, which can indicate a specific disorder or a disease.

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

Rare disorders and unknown disorders

Until they have a diagnosis, people with unclear or unusual health needs may live for some time with significant effects but no knowledge of the cause. To ensure that people with rare disorders receive timely diagnosis and care, improvements are needed for a wider group of people whose health problems are presently unknown or not attributed to a cause.

Supportive care can promote wellbeing even when a diagnosis is not yet possible.

Te horopaki o Aotearoa – Aotearoa New Zealand context

The strategy builds on and draws from other work such as the New Zealand Disability Strategy 2016–2026, the Child and Youth Wellbeing Strategy 2019 and the six Pae Ora Strategies:

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

The strategy is also underpinned by the United Nations (UN) Convention on the Rights of Persons with Disabilities, which Aotearoa New Zealand ratified in 2008.

As a small country, Aotearoa New Zealand cannot hold expert knowledge of every rare disorder on its own. We rely almost entirely on international rare disorders data, evidence and prevalence estimates to determine the effects of rare disorders on people and their whānau living with rare disorders in this country.

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 with international collaborations, globally and regionally, to share knowledge and expertise.

Honouring Te Tiriti o Waitangi

Section 6 of the Pae Ora (Healthy Futures) Act 2022 (Pae Ora Act) outlines the Crown's approach to meeting its obligations under Te Tiriti. The legislation contains specific provisions intended to give effect to the Crown's obligations. In particular, the health sector principles in section 7 of the Pae Ora Act guide the Minister of Health, the Ministry of Health – Manatū Hauora and all health entities on how to carry out their functions. The health sector principles incorporate key outcomes and behaviours that come from the principles of Te Tiriti, as set out by the courts and the Waitangi Tribunal. These outcomes and behaviours include:

- tino rangatiratanga
- equity
- active protection
- options
- partnership.

Ngā mātāpono o te rautaki – Principles that underpin the strategy

To ensure the strategy supports pae ora (healthy futures) for all New Zealanders, we adopted a set of principles early in its development. To uphold these principles, action will:

- build on and draw from the Pae Ora Strategies, the New Zealand Disability Strategy and the Child and Youth Wellbeing Strategy
- honour Te Tiriti o Waitangi and work towards achieving equity for Māori
- give voice to people and their whānau living with rare disorders
- support health practitioners and providers to deliver quality care
- be informed by and seek out evidence
- be collaborative and build on partnerships
- support pae ora for all while focusing on the health system challenges that come with rarity.

Ngā taunakitanga a te rautaki – Evidence that informed the strategy

This section summarises evidence we drew together while developing the strategy. A full evidence review is also available.[1]

Rare disorders in Aotearoa New Zealand

Around 300,000 New Zealanders, and their whānau, are thought to live with a rare disorder. This estimate is based on overseas studies, as is most of our understanding of rare disorders.[2]

Also based on overseas studies:

- around half of people with rare disorders are children
- some 80% of rare disorders are genetic
- rare disorders can occur in any family or population group in our country.

Over 7,000 different rare disorders have been identified. A person may be the only one in the country with their rare disorder. Alternatively, often two or more New Zealanders have the same rare disorder, but they tend not to be in the same age group or living in the same region.

We lack specific information on how many people are living with rare disorders and which disorders are most prevalent in Aotearoa New Zealand. Very little information on rare disorders is easy to retrieve from health or statistical data. The information we do have (often about particular rare disorders) indicates that, overall, Aotearoa New Zealand is likely to have proportions of people affected by rare disorders that are similar to comparable countries.

Many of the estimated 300,000 New Zealanders living with a rare disorder may not know about it. It is also possible that its impacts may be contained to one aspect of their life or health, or they may experience limited or no health impacts. However, some rare disorders have profound and pervasive impacts and come with the highest health needs of anyone in Aotearoa New Zealand.

Like all New Zealanders, people and their whānau living with rare disorders want to live well and to have choices. People and their whānau living with rare disorders have provided the key input to the strategy. Other important contributors have been the wider rare disorders community and supporters from across the health and disability sector.

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[1] Office of the Chief Science Advisor, Ministry of Health. 2023. Evidence Brief: Healthcare and rare disorders.

[2] This number is nuanced as prevalence estimates vary widely. The independent review of Pharmac compared the wide-ranging estimates of prevalence and of the number of people who have a treatable rare disorder. The estimate reached depends on the assumed rate in the population and the percentage of disorders that are treatable. For example, an estimate that uses 1.5% prevalence of rare disorders in the population equates to 76,839 people in Aotearoa New Zealand with a rare disorder. In contrast, based on a prevalence rate of 6.2%, the number rises sharply to 317,601 people with a rare disorder. Pharmac Review Panel. 2022. ***Pharmac Review: Final report***. URL: health.govt.nz/publication/pharmac-review-final-report

Health and wellbeing of people with rare disorders

Rare disorders can have a widespread effect on the lives of people and their whānau living with rare disorders, including by making daily activities difficult. However, individuals vary widely in their experiences. Some rare disorders have relatively mild impacts on everyday life, while the impacts of others can be severe and life-limiting. Some rare disorders can be diagnosed prenatally, while others are diagnosed much later in life. From the time of diagnosis, a person's lifespan may be very short or they may live with the disorder for a long time.

It is important that people and their whānau are able to access help and support with all of their needs, especially where those needs are high or complex. People living with a disorder may have a range of health needs, including needs not related to that disorder. Their whānau may likewise have various health needs, including needs related to the disorder of their whānau member.

Voice of rare disorders survey

Rare Disorders New Zealand sponsors a national 'voice of rare disorders survey' every two years to collect information about people and their whānau living with rare disorders. The findings give insights into the impacts of rare disorders for people and their whānau.

Over half of the 718 respondents to the most recent survey (2021)[3] reported that an accurate diagnosis took over 5–10 years and that delays and misdiagnoses prevented access to treatment or caused further harm.

Rare disorders have both direct and indirect costs for people and their whānau. Beyond the medical and treatment costs, losses due to disability, morbidity and quality of life impact the everyday wellbeing of individuals and their whānau living with rare disorders. For many, living with or caring for someone with a rare disorder often decreases their capacity to work, creating barriers to employment and education.

Individuals with rare disorders can have complex needs that also may be unique. In the 2021 'voice of rare disorders survey', people and their whānau responded that the financial burden of having rare disorders was hard to manage. On top of the costs for treatment, they faced additional out-of-pocket costs associated with having a rare disorder. These wide-ranging costs show that individuals with rare disorders require integrative care management to positively influence their wellbeing and financial situation.

A wide range of social support is often required to manage rare disorders. Emotional and psychological support for people and their whānau living with rare disorders can be vital for positive long-term outcomes, due to hereditary factors or the progressive nature of some disorders.

Disabled people living with rare disorders and those living in rural and ethnic communities (including Pacific peoples) may experience further difficulties in accessing the support they need from the health and social systems because of the barriers to health care that some members of these communities already experience.

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[3] Voice of Rare Disorders, Rare Disorders New Zealand. 2022. White Paper: *Impact of Living with a Rare Disorder in Aotearoa New Zealand*. URL: https://www.raredisorders.org.nz/about-rare-disorders/rare-disorders-in-new-zealand/nz-voice-of-rare-disorders-white-paper-2022/

Health system experiences of people living with rare disorders

People and their whānau living with rare disorders are often experts on their individual disorder. Voices of people and whānau with rare disorders play a vital role in the health system due to the expertise and knowledge they often hold.

Effective health care for people and their whānau living with rare disorders can make a positive difference to life beyond addressing their health needs. However, it can be difficult to provide effective health care for people living with rare disorders.

> ‘We had some people come and going, but nobody knew, it was all up in the air. We could try this, or we could do this, or you could see this person and then they didn't know … once seeing someone, we were still left without any information. Basically, just here, give this a go … and nobody listened to me … Look at [tamaiti] … someone needs to do tests or something. It’s just not right, you know? So, we fought. We fought for a good year and that … was the stressful time.’
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> **Whānau Māori on accessing services for tamariki living with a rare disorder, community engagement for the strategy.**

People living with rare disorders can struggle to get the support they need from the health system. Not only are the disorders rare, professionals who can provide effective care may be rare as well. No health professional, or even group of health professionals, can be experts across the great breadth of rare disorders. In some cases, people and their whānau living with rare disorders may know a lot more about their particular disorder than the health professionals they encounter.

Many people and their whānau living with rare disorders have good experiences with receiving care and treatment from our health system. However, we have heard that too many people have struggled to gain recognition of their own or their children’s health or developmental needs.

We’ve heard that without timely access to assessments or tests, expert diagnosis of a rare disorder can be delayed and can take many years. This can contribute to worsening health conditions before preventive care can start.

Following a diagnosis, people and their whānau can face barriers to finding treatments and support. Experience or expertise for a condition may be limited in specific regions or across the country. Where a treatment or specialist medical equipment exists, it may be hard to source, unavailable where they live or unaffordable. These experiences are very similar to those described in other countries around the world.

Supporting people with rare disorders to attain the highest level of health has clear benefits for the individuals and across the health system. Lack of effective care not only has a negative impact for people with rare disorders, but also results in the need for ongoing symptom management, mismanagement of the disorder, dissatisfaction for those working in the system and a strain on health (especially hospital) resources.

People living with rare disorders can experience poorer health and wellbeing outcomes. Setting up a health system that is responsive to everyone living with rare disorders takes a coordinated effort.

International efforts to improve rare disorders outcomes

> ‘If governments really aim to achieve Universal Health Coverage, “Leave No One Behind” by 2030, and build inclusive societies, then the focus needs to be on equity. Actions to achieve greater equity should be responsive to diverse realities and multiple forms of inequalities, including the inequities experienced by vulnerable populations throughout a lifetime such as people living with rare diseases and their families.’
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> **Rare Diseases International, UN Universal Health Coverage Day**

In 2021, the UN member states unanimously adopted an inaugural UN resolution on ‘Addressing the Challenges of Persons Living with a Rare Disease and their Families’. The resolution encourages countries to promote public policies, patient-centred programmes and initiatives that would strengthen health systems and calls for action to address the specific challenges facing people and their whānau living with rare diseases.

Many countries have 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. These approaches include programmes to:

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

Screening programmes for newborns are being developed to diagnose and treat rare disorders earlier. These programmes use a targeted population-level approach. Their effectiveness in enabling an earlier diagnosis depends on the methods used, range of disorders tested for and the availability of the programmes.

Current evidence to inform epidemiology and cost-benefit analyses of different health interventions for rare disorders is limited, so there is often uncertainty about their benefits or sustainability over time. Because epidemiological data and cost-benefit analyses of different interventions provide important information for prioritising very limited health resources, the high costs of rare disorder treatments mean that health system decisions on funding these treatments often involve balancing the needs of individuals with rare disorders alongside the health needs of the wider population.

Countries differ in their approaches and areas of focus. Significant progress is often made when they concentrate on multiple reinforcing priorities. The Aotearoa New Zealand Rare Disorders Strategy gives priority to five areas for action.

The priority areas for action

Priority 1:
Gearing the system for quality and timely care

Priority 2:
Learning and sustaining progress

Priority 3:
Equipping the health workforce for quality rare disorders care

Priority 4:
Giving voice to people and their whānau living with rare disorders

Priority 5:
Joining up internationally to achieve more

The key changes under each priority in the strategy are based on the evidence and information available internationally and from within Aotearoa New Zealand. This information can be found in both the strategy and supporting document *Aotearoa New Zealand Rare Disorders Strategy: Rare voices and aspirations.*

Making progress on any one priority will help to improve health support for people and their whānau living with rare disorders. For significant and sustainable improvements, action is needed across all five priorities. They all work together.

For example, priority 2: Learning and sustaining progress, is closely linked with all of the other priorities. Collecting information about rare disorders is an important action under priority 2 and is also a key enabler of achievement (and of knowing what might be achieved and how) in each of the other priorities.

People and their whānau who will benefit most from actions and improvements that flow from this strategy are those for whom the impacts of a rare disorder:

- can be prevented or lessened by earlier diagnosis and high-quality care and management
- are very high or profound and require a high level of expert, specialised or extraordinary care to achieve the best quality of life attainable.

Te whakatinanatanga o te rautaki – Turning the strategy into action

This strategy outlines the future direction for the health system to ensure that people and their whānau living with rare disorders share in pae ora (healthy futures). Action across agencies, services, organisations and communities will be involved in taking the strategy forward.

The Ministry of Health, Health New Zealand – Te Whatu Ora, Pharmac and the Health Quality & Safety Commission – Te Tāhū Hauora are accountable for implementing and monitoring the strategy.

Many other people, organisations and groups were pivotal in forming the strategy and will have important roles as it is put into action.

The Ministry of Disabled People – Whaikaha, the Ministry for Children – Oranga Tamariki, the Health and Disability Commissioner and the New Zealand Blood Service have a role as central government agencies serving populations to whom rare disorders are important. Non-governmental organisations like Rare Disorders New Zealand and Carers New Zealand have a role, as do iwi, universities and research institutes, professional colleges and service providers.

People with lived experience of rare disorders have a particularly important role. It is only through their real-life insights that we can fully understand and act on opportunities to improve rare disorders outcomes and ultimately achieve progress.

The Ministry of Health will monitor implementation of the strategy as part of its stewardship role.