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**Te Kāwanatanga o Aotearoa**  
New Zealand Government



MINISTRY OF HEALTH

# **Summary – Aotearoa New Zealand Rare Disorders Strategy 2024 – Context and evidence**

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# **Summary – Aotearoa New Zealand Rare Disorders Strategy 2024 – Context and evidence**

## **Context for developing the strategy**

### **Purpose**

The Aotearoa New Zealand Rare Disorders Strategy sets out the direction for the health system to better support people and their whānau with rare disorders over the next 10 years. It provides a framework and long-term priorities that will guide health entities in improving health and wellbeing outcomes. The strategy responds to longstanding calls from the rare disorders community and the Government's response to the 2022 independent review. It aims to improve lives, provide timely services, and ensure equitable support. The strategy will be successful if people and their whānau have better quality of life, the health system responds and relates equitably to people whose needs or situation are uncommon, unusual or unclear and those involved in designing 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.

## **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:

- A New Zealand Health Strategy
- Provisional Health of Disabled People Strategy.
- Pae Tū: Hauora Māori Strategy
- Te Mana Ola: The Pacific Health Strategy

- Women's Health Strategy
- Rural Health Strategy

It is underpinned by the United Nations Convention on the Rights of Persons with Disabilities.

## **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 the Pae Ora Strategies, New Zealand Disability Strategy and 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.

# **Evidence that informed the strategy**

## **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. About half of these individuals are children and some 80% of rare disorders are genetic. Over 7,000 different rare disorders have been identified. A person may be the only one in the country with their disorder, or there is often two or more that have the same rare disorder, but they tend not to be in the same age group or live in the same region.

We lack specific information on how many people are living with rare disorders and which 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 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 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.

People and their whānau living with rare disorders want to live well and to have choices. People and their whānau have provided the key input to the strategy, along with contributions from the wider rare disorders community and supporters from across the health and disability sector.

## **Health and wellbeing of people with rare disorders**

Rare disorders can significantly affect the lives of people and their whānau, making daily activities challenging. Experiences vary widely; some rare disorders have mild impacts on everyday life, while others can be severe and life-limiting. Diagnoses can occur prenatally or 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. Access to help and support for all needs is important, especially for high or complex needs.

### **Voice of rare disorders survey**

Rare Disorders New Zealand sponsors a national survey every two years to collect information about people and their whānau living with rare disorders. The 2021 survey found that over half of the 718 respondents 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. For many, living with or caring

for someone with a rare disorder often decreases their capacity to work, creating barriers to employment and education. The 2021 survey found that the financial burden was hard to manage, with additional out-of-pocket costs.

Emotional and psychological support for people and whānau 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 in rural and ethnic communities may experience further difficulties in accessing the support they need from the health and social systems.

## **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 play a vital role in the health system due to the expertise and knowledge they often hold. Effective health care for people and whānau can positively impact their lives beyond addressing their health needs, but it can be difficult to provide effective health care due to the rarity of both the disorders and knowledgeable professionals. In some cases, people and their whānau know more about their disorder than the professionals they encounter. Many people and whānau have good care experiences, but too many struggle to get the support and recognition for their



health needs, face delays in diagnosis, and encounter barriers to treatments and support.

Delayed access to assessments or tests can contribute to worsening health conditions before preventive care can start. Following a diagnosis, finding treatments and support can be difficult due to limited expertise and availability or affordability of treatments or specialist medical equipment. These experiences are very similar to those described in other countries. 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 leads to ongoing symptom management and a strain on health 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**

In 2021, the United Nations member states adopted a resolution on 'Addressing the Challenges of Persons Living with a Rare Disease and their Families', urging countries to enhance health systems through public policies and patient-centred programmes. Many countries have developed strategies and plans to guide their health and social sector to support people living with rare disorders,

focusing on improving understanding and knowledge, making it easier to include and respond well to people and their whānau living with rare disorders, building on collaborating with rare disorder communities, and learning from and contributing to international advances.

Screening programmes for newborns are being developed to diagnose and treat rare disorders earlier. Current evidence to inform epidemiology and cost-benefit of analyses of different health interventions is limited, so there is uncertainty about their benefits and sustainability over time.

Countries differ in their approaches and areas of focus. Significant progress is often made when concentrating 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

Each priority is based on 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’. Progress on any one priority will support improvements to health for people and their whānau. People and whānau who will benefit the most from actions and improvements that flow from the strategy are those whom the impacts of a rare disorder:

- can be prevented or lessened by earlier diagnosis and quality care 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.

## **Turning the strategy into action**

The 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, Pharmac and the Health Quality & Safety Commission are accountable for implementing and monitoring the strategy. The Ministry of Disabled People, the Ministry for Children, the Health and Disability Commissioner and the New Zealand Blood

Service have a role as central government agencies serving populations whom rare disorders are important. Non-governmental organisations like Rare Disorders New Zealand and Carers New Zealand have a role, as do iwi, universities, research institutes, professional colleges and service providers. People with lived experience have a particularly important role. It is only through real-life insights that we can fully understand and act on opportunities to improve rare disorders outcomes and ultimately achieve progress.

The full version of the strategy can be found here:

[\(<https://www.health.govt.nz/publication/aotearoa-new-zealand-rare-disorders-strategy-context-and-evidence>\)](https://www.health.govt.nz/publication/aotearoa-new-zealand-rare-disorders-strategy-context-and-evidence)

**End of Summary – Aotearoa New Zealand Rare Disorders Strategy 2024 – Context and evidence**