

BRIEFING

Rare Disorders in New Zealand 2025



Overview

An estimated 300,000 New Zealanders live with at least one of over 7,000 known rare disorders.

A disorder is considered rare when it affects fewer than or equal to 1 in 2,000 persons in New Zealand. Rare disorders include, but are not limited to, genetic disorders, cancers, infectious disorders, poisonings, immune-related disorders, idiopathic disorders, and undetermined conditions that are rare.

Often complex, debilitating and life threatening, these conditions require a broad range of services.

While each disorder is unique, we know from [surveys carried out by Rare Disorders NZ](#) that collectively those affected largely face many of the same barriers and challenges in the New Zealand health system – lack of timely diagnosis, limited treatment options, no support with coordinating care, isolation, significant carer impact and for many, being lost in the system.



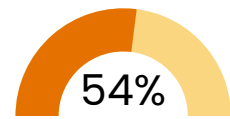
Lack of timely diagnosis

For one in five, it took over 10 years to get a diagnosis.



Poor coordination of care

Over 50% felt that communication between different service providers was poor.



Financial strain

54% felt the costs associated with managing their rare disorder were hard to manage.

Source: Impact of Living with a Rare Disorder in Aotearoa New Zealand, February 2022

The common experience of being bounced around from specialist to specialist takes a significant toll on the mental and physical health of patients and their families, and puts unnecessary pressure and additional costs onto the health system.

New Zealand's first Rare Disorders Strategy

In July 2024, the Ministry of Health released New Zealand's first [Rare Disorders Strategy](#). The Strategy outlines the Government's intentions to improve health outcomes for people living with a rare disorder. While the Strategy guides the long-term priorities for health entities over the next 10 years, the need for improvements for rare disorders remains urgent.

Many of the actions to improve health services for rare disorder patients do not involve substantive programmes of work, but rather involve tweaking and expanding existing systems and services to better respond to rare disorders. Rare Disorders New Zealand, as the only umbrella organisation for all rare

disorders in New Zealand is well-placed to provide advice and support during implementation of the Strategy.

Since the Strategy's release however, the Ministry of Health has been prevented from initiating its mandated leadership role in the Strategy's implementation due to Te Whatu Ora's 'reset' phase, halting any progress. This is causing deep discontent among the rare disorder community who have been desperately waiting for improvements to services.

Urgent actions required

There are actions Rare Disorders NZ believes can be initiated regardless of Te Whatu Ora's reset to pave the way for improvements in the health system for rare disorder patients:



Action # 1

Establishment of a Rare and Undiagnosed Disorders Centre of Expertise



Action # 2

Establishment of a single barrier-free pathway to rare disorder medicines



Action # 3

Incorporation of coding of rare disorders in the nationwide Electronic Medical Record and other data sets



Action # 4

Recognition of RDNZ as a key enabler for the Strategy's implementation

#1 Rare and Undiagnosed Disorders Centre of Expertise

A virtual Centre of Expertise would improve early and accurate diagnoses of patients and 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, such as Australia, the UK and in Europe. The centre would comprise a multidisciplinary team of experts within New Zealand, who would be available to provide expertise and guidance for clinicians and health professionals in how to support rare patients and clients in accordance with best practice standards. This work would be an extension of roles they hold with locality-based health and other service providers.

#2 Single barrier-free pathway to rare disorder medicines

While less than 10% of rare disorders have an existing treatment, ensuring access to these treatments can prevent further deterioration of patients' health and reduce their dependence on the health system and other services. The current one-size-fits-all medicine assessment model under Pharmac's pharmaceutical schedule does not work for low volume, high-cost medicines, and there are too many inconsistencies under the only other avenue – the exceptional circumstances framework. A separate assessment pathway for rare medicines with criteria separate to common conditions (as is implemented in Europe, Australia and Japan) will ensure equitable access to medicines for those with rare disorders.

#3 Incorporating coding of rare disorders in the roll-out of the nationwide Electronic Medical Record and other data sets

The classification system for diseases that New Zealand uses does not include most rare disorders. This lack of data is problematic for funding and resource allocation, as these patients tend to be high-need, high-cost patients. With the implementation of the nationwide Electronic Medical Record using SNOMED

terminology, there is an opportunity to incorporate coding of rare disorders using Orphanet Coding in this system, as 6,500 Orphacodes, have been mapped to SNOMED CT codes. Orphanet is the most comprehensive database of rare disorders.

#4 Rare Disorders NZ is a key enabler for the Rare Disorder Strategy's implementation

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 their families. The organisation is well-placed to support the implementation stage of the Strategy, to ensure actions align with the needs of patients.

These immediately implementable actions would significantly improve health system responsiveness to rare disorders and contribute overall to a more efficient health sector as a result of:



- marshalling and focusing already existing sources of expertise;
- earlier accurate diagnoses;
- earlier access to effective treatment and support services;
- more efficient use of resources;
- diagnostic and support odysseys are shortened resulting in fewer demands on over-stretched systems;
- availability of prevalence data supporting resourcing, future-proofing and budget setting.



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