European Rare Diseases
Research Alliance

Resource on the State of the Art of Rare Disease Activities

2025 Report for New Zealand

National Mirror Group

New Zealand has a National Mirror Group (NMG) in place, which was launched in December 2025. Rare Disorders New Zealand's (RDNZ) is the coordinator. The NMG utilises RDNZ working groups to reach across research, clinical and government spaces. The New Zealand NMG aims to work closely with the Australia NMG.

The collection of data from New Zealand for this report has been overseen via a data contributing committee. Contributors are listed at the end of the report.

Definition of a Rare Disease

New Zealand adopts the formal European Union (EU) definition of a rare disease (i.e. those with a prevalence of no more than 5 patients per 10,000 persons. This definition is laid down in Regulation EC no. 141/2000 on Orphan Medicinal Products, Directive 2011/24/EU on Cross Border Healthcare as well as in the Council Recommendation on an action in the field of rare diseases of 8 June 2009.). The National Strategy espouses this definition and there are no instances when a different definition is used.

Status Quo of any National Plan or Strategy for Rare Disease

What is the status quo?

New Zealand has a live, in-date, National Strategy for Rare Disease. The National Strategy is timebound, covering a period of ten years. This is New Zealand's first National Strategy for Rare Disease.

Elaboration and Adoption/How was the original strategy elaborated?

A dedicated group was in charge of overseeing the drafting and adoption of New Zealand's National Strategy for Rare Disease. The following stakeholder types were included in this group:

Co-funded by the European Union

ERDERA has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement N°101156595.
Views and opinions expressed are those of the author(s) only and do not necessarily reflect those of the European Union or any other granting authority, who cannot be held responsible for them.

- Patients/people with lived experience of a rare condition
- New Zealand's National Alliance for rare disease patient organisations
- Health Ministry/Competent National Authority in charge of Health or Care
- Research Ministry/ Competent National Authority in charge of Research
- Social or Welfare Ministry/ Competent National Authority in charge of social affairs
- Researchers/clinicians from rare disease centres

The National Strategy was led by New Zealand's Ministry of Health, with input from a reference group representing different health and medical associated entities. Rare Disorders New Zealand played a major role in partnering with the Ministry of Health to help engage with people with a lived experience of a rare disease, carer community representatives, clinicians and academics.

The same group is in charge of implementing and overseeing the National Strategy for Rare Disease. This group was not specifically set-up for overseeing/implementing the National Strategy for Rare Disease. This group oversees all strategy documents related to health in New Zealand.

This group is not currently functioning and does not meet. Since the National Strategy was published, there has been no further action to deliver the strategy.

Is there funding for the Strategy? How is it Implemented and/or Monitored or Evaluated?

No funding or resources were committed to New Zealand's National Strategy for Rare Disease, nor was there any funding to deliver any specific actions within the strategy.

No formal evaluation or monitoring of New Zealand's National Strategy for Rare Disease has taken place.

Research in National Plans or national research strategies relevant to rare disease

New Zealand's National Strategy for Rare Disease does not specifically address rare disease research. The strategy does state that rare disease should be considered in the various areas of research.

The strategy does acknowledge the need to collaborate internationally, and this is included as one of the five priorities within the strategy.

The National Strategy acknowledges the need for better diagnosis, access to clinical trials, and sociological needs, but does not explicitly mention research in these areas.

Rare Disease Research Programmes and Funding

There are no programmes or funding specifically for rare disease research but specific projects for rare disease research are funded from general research programmes. Approximately NZD$5 million from national funding has supported rare disease research. In addition, approximately NZD$1.5 million funded by charities has supported rare disease research.

Rare Disease Registration and Biobanking

Neither a national nor regional rare disease registry is in place in New Zealand. However, there are disease specific registries.

New Zealand does not have a national biobank for rare disease biosamples but there are some individual biobanks for rare disease samples.

Organisation of Rare Disease Care

Centres of Expertise

There is no policy within New Zealand for designating centres of expertise for rare disease. Rare disease expertise is identified informally e.g. online, by colleagues and via Rare Disorders New Zealand. There are currently no plans to develop or adopt a formal national policy for establishing Centres of Expertise for rare disease in New Zealand.

Newborn Screening

Over 28 rare diseases are screened for in New Zealand's Newborn Screening programme. Further information can be found here.

Diagnostics

In New Zealand there are three main testing laboratories, which are linked to three regional clinical genetics service hubs. Each laboratory has specific tests they are responsible for (e.g. cancer panel testing, karoytyping/arrays, exome/Next Generation Screening (NGS testing)). These tests are publicly funded; therefore no costs are incurred, but only genetic specialists are able to access these tests. Private testing is available but it is generally undertaken by the same providers.

Advanced tests such as exome or genome sequencing are not able to be performed in New Zealand due to a lack of infrastructure, staff capacity and financial resourcing. These tests are sent to Australia or Finland (Blueprint Genetics). Many Indigenous people view DNA to be sacred and carry their ancestry, therefore there is an effort to repatriate testing so that no DNA samples need to leave the country. This Indigenous view is not explicitly stated in the National Strategy but is known by many in the field.

There are currently no specific strategies to address undiagnosed patients or people with currently undiagnosable conditions. There is an appetite to initiate an Undiagnosed Diseases Network in New

Zealand, but this would require research funding. There is also no national approach for reanalysis, similarly to research, funding would be needed to trial this. Diagnostic laboratories report no capacity to undertake this routinely.

There is a policy in place to ensure national providers provide genetic counselling for patients with a suspected or confirmed rare disease. However, the patients/families must first be accepted to be seen by a genetic specialist.

National Alliances of Rare Disease Patient Organisations

Rare Disorders New Zealand is New Zealand's National Alliance of rare disease patient organisations. Further information can be found here.

Information Resources for Rare Disease

National Orphanet Engagement

There is no operational national Orphanet team/branch in New Zealand.

Helplines

New Zealand does have a national helpline in place dedicated to rare disease. This helpline receives a mix of public and private funding and is available for anyone to use. The helpline is run by Rare Disorders New Zealand and comprises of a help and advice telephone service available during office hours Monday to Thursday, with an option to leave a voice message if the phone is unattended. People can also request a phone conversation via Rare Disorders New Zealand 's Facebook page and website. The service is provided by paid Rare Disorders New Zealand staff, who are not clinicians. If clinical advice is sought, they are directed to appropriate health and other support systems. Where appropriate, staff can offer to connect people to one of over 150 disorder specific support groups.

Training and Education

There are no rare disease specific training activities in New Zealand. Rare disease is occasionally and briefly covered within wider clinical training courses.

Orphan Medicinal Products (OMPs)

There are 12 OMPs available within New Zealand. These OMPs have been purchased by New Zealand's medicines procurement agency (Pharmac). Further information about these OMPs can be found on page 6 of this report.

New Zealand's early access/expanded access programmes to help patients access medicinal products for rare conditions, are currently in development. Pharmac uses its Named Patient Pharmaceutical Assessment (NPPA) process to consider whether to fund a treatment for an individual patient, whose clinical circumstances are exceptional. Rare Disease New Zealand describes this programme as in development because there is a need to review the scheme to make it more accessible, equitable and fit for purpose. Full details are available here.

There are no formal efforts or initiatives with other countries to support access to medicines and therapies for rare diseases in place in New Zealand.

Disclaimer: The data collection activities which enabled this 2025 national report were supported by the ERDERA. ERDERA has received funding from the European Union's Horizon Europe research and innovation programme under grant agreement N°101156595. Views and opinions expressed are those of the author(s) only and do not necessarily reflect those of the European Union or any other granting authority, who cannot be held responsible for them, nor should this document be viewed as an official national 'position'.

Co-funded by the European Union

The Data Contributing Committee of New Zealand, which provided this 2025 data (correct as of the end of December 2025) in the context of the Resource on the State of the Art of Rare Disease Activities, is composed of the following individuals:

- Louise Bicknell, University of Otago
- Chris Higgins, Rare Disorders New Zealand