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Summary – Aotearoa New Zealand Rare Disorders Strategy 2024

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Summary – Aotearoa New Zealand Rare Disorders Strategy 2024

Purpose

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 living with rare disorders over the decade 2024 to 2034.

Vision

Our vision is for all people and their whānau living with rare disorders to share in pae ora (healthy futures) and live fulfilling lives.

Pae ora (healthy futures) 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.

What a ‘rare disorder’ is

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.

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

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.

Background

Globally, there are over 7,000 known rare disorders, with more being identified each year. Many are ultra-rare and may affect only one or a handful of New Zealanders. Yet, in total, some 300,000 New Zealanders may live with a rare disorder.

People and their whānau living with rare disorders often face significant barriers to getting timely, effective and equitable health care. Health practitioners may find it difficult to identify rare disorders or may not know how to respond to support their health and wellbeing. Aotearoa New Zealand needs a rare disorders strategy to improve system responsiveness and support for people and their whānau living with rare disorders, as well as for service providers who often face difficulties meeting patient needs.

This strategy is for all who identify as, or provide support to, people (many of them children) and their whānau living with rare disorders in Aotearoa New Zealand. It is also for those who may not yet have a diagnosis or may not identify as someone with a rare disorder but can see their lived experiences reflected in the strategy.

Priority areas

Priority 1: Gearing the system for quality care

People and their whānau living with rare disorders often need help from a range of interconnected health services that are consistent nationwide. Practitioners have highlighted the need for more joined-up care, especially in hospital settings where diagnosis occurs without proper links to follow-up care and support.

Building leadership and coordination

Building leadership and coordination to address responsiveness to rare disorders will enhance system-wide visibility and understanding. Currently disconnected, responses will be integrated, establishing consistent capability and support across services. Leadership mechanisms will adopt diagnostic, referral, and care standards, ensuring long-term sustainability. International approaches, like networks and information hubs, will guide the choice of mechanisms in Aotearoa New Zealand, focusing on integration and continuous improvement.

Introducing new capabilities

New capabilities will be introduced to speed up diagnosis and enable earlier preventive care, particularly through genomic testing and rapid DNA sequencing for infants and children. Infrastructure for early testing and preventive care will be developed over time. Horizon scanning will prepare for emerging technologies, assessing risks and benefits for safe adoption. People and their whānau living with rare disorders will significantly contribute to work ensuring the safety and quality of new technologies.

Considering rare disorders when investing and prioritising

The decision-making approach for system investments will improve to value the benefits for people and their whānau living with rare disorders. A programme of work across entities will enhance access to diagnostic, preventive, and treatment services, focusing on rare disorders. Early goals include clarifying access to tests, advice, and treatments. Over time, the programme will address care aspects such as impact estimation, equity, and fairness across different types of investments and needs.

Priority 2: Learning and sustaining progress

At present, we collect little information on rare disorders systematically. With limited data, we don’t know precisely how many New Zealanders live with rare disorders, what the health impacts are, and how many receive timely diagnosis and care.

Collecting information on rare disorders

National health data sets will routinely collect rare disorder information, becoming more extensive and detailed over time. People and their whānau living with rare disorders will be able to access their information as well as general information on rare disorders in Aotearoa New Zealand. Clinical classifications will become more detailed with advanced systems. Methods to capture data on suspected but unknown rare disorders will be developed, using general codes where specific categories are unavailable.

Using information to support provision of care

In the future, services will use rare disorder information to guide, monitor, and improve patient care. Increased visibility of this information will aid in planning care, tracking results, filling gaps, and making improvements, building on existing work already done for some rare disorders. Where clinical coding is unavailable, methods will link high-quality rare disorder information with national health data. Planners and researchers will use non-identifiable information to enhance care over time.

Using information to monitor and improve care

Health information reports that include rare disorders will lead to improved care and choices for particular groups of patients. Regular and issue-specific reports will support quality care provision. Connecting up data for people and whānau will enable better and more timely care and choices. Connecting up data across groups with related rare disorders will enhance care and outcomes while ensuring careful privacy standards to protect personal information. Examining de-identified data will help the system find ways to better support and enable wellbeing for people and whānau living with rare disorders.

Using information to decide on priorities

In the future, decision-makers will prioritise rare disorders when evolving the system. They will consider impacts on rare disorder outcomes before changes. Rare disorder patients and their whānau will be routinely considered in system improvements. Priority will be given to initiatives benefiting these groups, such as intergenerational equity, child services, genetic infrastructure, and supportive therapies. Decision-makers will confirm that improvements benefit rare disorders, not just assume they do.

Supporting research and evaluation

The health research infrastructure will support rare disorder research and evaluation. An expanded framework will enable researchers and participants to engage in health and health services research, with a focus on rare disorders. This includes evaluating services, systems, and outcomes. Key activities will involve building clinical trial infrastructure, developing methodologies for assessing benefits and costs, evaluating service improvements, and integrating rare disorder data with the Stats NZ Integrated Data Infrastructure.

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

Many people and whānau have expressed frustration about the lengthy process of getting a rare disorder diagnosis, involving multiple practitioners who often lack knowledge about rare disorders. People and whānau feel they need to ‘fight’ for attention or to heard. Health practitioners want to help but often lack the necessary information, resources or tools.

Rare disorders in health practitioner education

Future education programs will teach health practitioners more about rare disorders, responsiveness to unusual needs, and finding further information. Curricula for health practitioner education at all levels will increasingly emphasise rare disorders, highlighting their impacts and the benefits of early identification and care. As diagnostics, clinical guidelines, and care pathways develop, students will learn to access the necessary information to support care.

Rare disorders learning modules

Learning modules for rare disorders will become widely accessible over time. Health workforce learning opportunities in rare disorders will be increasingly available and become more sophisticated. Learning modules will include offerings on rare disorders specifically as well as more general offerings that include rare disorder content.

Rare disorder examples will feature in an increasing portion of health workforce learning opportunities and increasingly in competency-based learning.

Support and wellbeing learning modules

Learning modules for support and wellbeing will be widely accessible and will increasingly include rare disorders content. Learning opportunities relevant to caring for and supporting people and their whānau living with rare disorders will become widespread. Rare disorder examples will be included in an increasing portion of learning opportunities related to tikanga practices, cultural safety and competency, care and support, counselling and social wellbeing.

Clinical guidelines and pathways

Standard care provision will incorporate clinical guidelines and pathways for rare disorders. A program or process will be established to identify, adapt, authorise and publish guidelines, standards and pathways for rare disorders, starting with well-established approaches and expanding to improve care quality for more disorders over time. Adaptation to the Aotearoa New Zealand context may include negotiating referral pathways and guidance on specialised care access, ensuring quality care for increasing numbers of rare disorders.

Everyday backup tools and easy-access practitioner support

Over time, published tools will integrate with standard reference sources and patient management systems. The system will also support rare disorder experts to advise practitioners. Expanding help and coaching for practitioners will require resourcing and backup systems for practitioners to ask for help, including online. Specialist roles may need redesigning to provide this support, along with capability development and cross-specialty learning to ensure high-quality and confident care.

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

People want rare disorder voices heard across the health system. They seek decision-makers to prioritise the unique challenges of rare disorders when providing services and determining access. Rare disorder communities feel the system often favours more common conditions. Elevating rare voices would lead to fairer decisions and improvements reflecting rare health needs.

Listening to rare voices

Voices of people and their whānau living with rare disorders will become evident across the health system. Increasingly, people who can speak to rare disorders will join consumer forums, advisory boards and committees, locality groups and partnership boards. Their input will be integral to system reviews, service evaluations, business cases, and investment decision-making, as seen in the 2022 independent review of Pharmac and the development of the Pae Ora Strategies.

Listening to whānau Māori rare voices

Whānau Māori living with rare disorders will increasingly experience manaakitanga in their interactions with health service providers. Experts will cater to each whānau with empathy and respect, ensuring comfort. Providers will acknowledge, welcome, and listen to whānau Māori, allowing space for tikanga Māori practices. Clear communication will build trust, supporting autonomy and self-determination. Providers will offer choices where possible, including rongoā Māori or mirimiri, to restore wellness of mind, spirit, body and emotions.

Including rare voices in communities

Community wellbeing efforts will become more inclusive of people and their whānau living with rare disorders. Health promotion activities in local settings and diverse communities will increasingly focus on promoting wellbeing for all community members. As activities and events grow, the focus on inclusivity will strengthen. People who can speak to rare disorders experiences will be included in planning and implementation, receiving support to participate.

Priority 5: Joining up internationally to achieve more

Rare disorders support groups have emphasised the importance of connecting internationally to support people and their whānau living with rare disorders. People and their carers and supporters value being able to connect with other people and whānau in similar situations or with the same disorder.

Partnering with other countries

Aotearoa New Zealand will join and initiate partnerships with other countries focusing on rare disorders. Aotearoa New Zealand health leaders will engage in international collaborations to improve outcomes for those living with rare disorders. We will contribute to global efforts, seek collaborations, and support initiatives to make new diagnostics, treatments, and technologies more accessible. We will support the interests of smaller and less resourced countries, especially our Pacific neighbours.

Learning, adopting and adapting

We will learn from other countries and adopt and adapt their tools, resources, and advances. Many countries use guidelines, decision trees, and referral and care pathways that generate good outcomes for people with rare disorders. A programme to adapt the most relevant tools for Aotearoa New Zealand will support and improve access to good care. Input from rare disorders community voices, along with Māori, Pacific, rural, and other diverse voices will be needed in testing quality and suitability for our context and service users. Staying updated on effective approaches and evaluations, such as the European Union’s virtual networks, will remain important.

Joining in international clinical trials

People with rare disorders will, in time, be more able to participate in international clinical trials. As our clinical trial infrastructure further develops, it will support New Zealanders to participate in international rare disorders clinical trials. Clinicians involved in trials will receive backup and research support where their patients are participants in suitable funded trials, even when there are very few potential participants from Aotearoa New Zealand.

Better regulation and standards

Regulation and regulatory processes and standards will support rare disorder outcomes as much as possible. Aotearoa New Zealand’s regulatory stewardship programme will review relevant legislation and frameworks to ensure they support rare disorders outcomes. Regulation of new technologies, digital capabilities, medicines, devices and therapeutic products can support safe and timely access to diagnosis and effective treatments. Flexibility will be needed for various rare disorder scenarios, and aligning with like-minded countries will help reduce timeframes for assessment.

Supporting documents and evidence

Two supplementary documents provide further detail and explanation on how we developed the priorities in this strategy:

- Aotearoa New Zealand Rare Disorders Strategy: Rare voices and aspirations
- Aotearoa New Zealand Rare Disorders Strategy: Context and evidence

We recommend reading these documents along with this strategy.

The full version of the strategy can be found here:

(https://www.health.govt.nz/publication/aotearoa-new-zealand-rare-disorders-strategy)

End of Summary – Aotearoa New Zealand Rare Disorders Strategy 2024