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**Te Kāwanatanga o Aotearoa**  
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MINISTRY OF HEALTH

# **Summary – Aotearoa New Zealand Rare Disorders Strategy 2024 – Rare voices and aspirations**

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# **Summary – Aotearoa New Zealand Rare Disorders Strategy 2024 – Rare voices and aspirations**

## **Purpose**

People and whānau living with rare disorders have provided key input to the Aotearoa New Zealand Rare Disorders Strategy. Other important contributors have been the wider rare disorders community and supporters from across the health and disability sectors.

## **Who is the strategy for**

### **For people and their whānau living with rare disorders**

All people deserve respect, support, and to be heard in health care, especially those with rare disorders who often require a higher level of support with needs that are complex, unusual and often unclear. Whānau and wider support networks are vital in supporting and making real the independence and aspirations for people living with rare disorders. The strategy outlines how the health system, practitioners and support organisations can work to support people and their whānau living with rare

disorders by providing resources, reducing wait times for tests and diagnosis, and offering ongoing support. This support, regardless of identification or diagnosis, helps people and their whānau to live well and supports child development. People with rare disorders contribute significantly to society, and reducing their negative impacts enhances these contributions.

## **For whānau Māori living with rare disorders**

People with rare disorders have significantly poorer health outcomes, with whānau Māori often experiencing even greater challenges. A report on the experiences and voices of whānau Māori living with rare disorders highlights barriers to culturally competent and safe health services, including discrimination, unconscious bias, and racism, leading to feelings of isolation. Māori often face barriers accessing information about diagnosis, funding, and support, and provided examples of being given a diagnosis without adequate guidance or respect. Many whānau, particularly parents of tamariki living with rare disorders, feel lonely and isolated and as though they have become experts in their condition due to a lack of openness from health professionals. Whānau-centred services aligned with te ao Māori have a positive impact on their own and others journeys across the health system. Many whānau Māori found terms like 'rare disorder' alienating, preferring that their tamariki be acknowledged as the taonga that they are rather than a medical term such as 'rare disorder'.

## **For health practitioners and care workers**

Health practitioners and care workers want to support people and their whānau living with rare disorders to live well and be able to contribute to their communities. The strategy outlines ways of enhancing practitioners' capabilities in two areas: relational and cultural competence, and knowledge access. Relational and cultural competence involves building strong relationships, showing manaakitanga, and creating culturally safe environments. Expanding cultural representation in the workforce and improving existing members' skills is important. Additionally, equipping health professionals with the necessary knowledge, tools, and specialties will help reduce the time taken to diagnose and treatment people with a rare disorder.

## **For the health system**

Many people and their whānau living with rare disorders have complex needs and face challenges in navigating the health system, especially those with disabilities or from rural and diverse communities, including Pacific peoples. The strategy aims to achieve a better-connected health system that has the technology and infrastructure required to provide quality care. A connected system will also benefit health users more generally.

## **For Aotearoa New Zealand communities**

Most people may not have heard of rare disorders. Some may have heard of a particular disorder but do not know that the disorder is rare. Greater awareness is one step towards people being able to support and include people and their whānau living with rare disorders. All community members benefit when people and their whānau with rare disorders can live well and participate in their communities to the full.

## **How rare voices have shaped the strategy and priorities for action**

People have said the strategy needs to work towards several aspirations for people and their whānau living with rare disorders. These include ensuring the best possible health and quality of life, providing Whānau Māori with resources to exercise tino rangatiratanga, and enabling Māori, Pacific peoples, disabled individuals, and those from rural or diverse communities to share in pae ora (healthy futures), focusing on developing a skilled health workforce and ensuring the system promotes health fairly for all, including those with rare or unknown disorders. The strategy outlines five priorities that will have the greatest impact, based on feedback from both people and their whānau living with rare disorders and health professionals, where action is most needed to drive improvements in health and wellbeing for those with rare disorders.

# **Priority areas**

## **Priority 1: Gearing the system for quality care**

### **What we've heard**

People and their whānau living with rare disorders often need help from a range of services across the health system. These services need to be connected up, easy to navigate and consistent nationwide. Practitioners have highlighted the need for integrated care, as hospital-based diagnoses of rare disorders often lack follow-up support. General practitioners and primary health services may find it difficult to get help from other parts of the system. Improved coordination and clearer referral practices and criteria are essential for effective care.

### **Why it matters**

Improving outcomes for rare disorders requires both supporting health practitioners and enhancing system organisation and coordination. Rare disorders often come with complex needs that involve many parts of the health system. Quality, timely, safe and life-affirming care needs to be accessible. Key areas for system improvement include ensuring connectivity and navigation support across the system, adopting effective and safe technologies early, and prioritising access and investment

across services, products, and infrastructure that work for rare needs.

## **Aspirations for the future**

### **The system will be connected up around people with rare disorders**

Expert input for assessment, diagnosis and care will be accessible throughout Aotearoa New Zealand, with support for travel if needed. Health funders and planners, practitioners and providers will focus on networking and sharing expertise across services and specialties. The system will have established ways to ensure timely access to different specialties and services. How the system works for rare will be consistent with other areas in the sector. A central leadership for rare disorders will be well established, with strong links to many service lines and networks and visible efforts to improve system responsiveness. Information and guidance on rare disorders will be readily available in any part of the health system.

### **New technology will identify more disorders, sooner, making preventative care possible**

Advances in precision health, including genomics and artificial intelligence, will enhance the prediction, prevention, diagnosis and treatment of health needs sooner and more precisely. As new technologies develop into treatments, such as CRISPR-based treatments, many

will first be applied to rare disorders. Aotearoa New Zealand will continue to focus on identifying new technologies and digital capabilities, ensuring their safety and fair application before conducting full development or affordability assessments, enabling early adoption of promising new capabilities.

### **Fair access to diagnosis, care, treatments and supports will be constantly improving**

Evaluating and assessing medicines, diagnostics and treatments for rare disorders is challenging due to the high costs and limited evidence. New medicines are often significantly more expensive and may require special technology and expertise. While some treatments will be developed in Aotearoa New Zealand, many will be developed elsewhere in the world. Supporting people to access treatments overseas will be considered, yet affordability will remain a key issue in the future. However, processes for considering new diagnostics, treatments, services and infrastructure will improve in speed and fairness, ensuring better support for rare and lifelong needs alongside emergency and acute needs.



## **Priority 2: Learning and sustaining progress**

### **What we've heard**

People want rare disorders to be visible in the health system and to see improvements in services and outcomes. They want health entities to have the information needed to provide fair access to safe, quality, and culturally responsive diagnosis and care. Participation in clinical research and trials is important, and genetic counselling should be available to extended whānau when an inherited rare disorder is identified. Being able to make choices about these opportunities is important.

### **Why it matters**

To continue to improve over time, the health system needs ways to measure and track progress. Health care planners, providers, and users need to be able to see what is working, where improvements are being made, and what more needs to be done. Indicators of system performance for rare disorders are needed within both health and disability indicators, with input from people and whānau living with rare disorders. Aotearoa New Zealand is unique in being able to gather knowledge of Māori with rare disorders and contribute to Pacific knowledge. Participating in clinical trials for new diagnostics and treatments is important to people with rare disorders for

whom no effective treatment is otherwise available. A culture of continuous learning, as emphasised by the New Zealand Health Strategy 2023 and the Provisional Health of Disabled People Strategy, is essential for future health system improvement.

## **Aspirations for the future**

### **Our system will collect information and data on rare disorders and make them visible**

Patient records will capture information to support care for people with rare disorders or possible rare disorders. Medical coding will keep up to date with changes in international classifications, allowing for continual updating of world knowledge about rare disorders. Disability data and links to specific rare disorders collections will be integrated. Governance, protections, access, and ownership of system data will be in place, with a strong emphasis on Māori data sovereignty. Rare voices and other consumer, cultural, and minority voices will be involved in discussions about these system foundation elements. Citizens will have a choice in their data and confidence in the in-built protections.

### **We will know how well our system works for people with rare disorders**

Services will routinely collect and report on rare disorders information, guided by health system service users, planners and providers. Data will be able to match a

variety of rare disorder codes to track health trends and progress without compromising personal information. Planners and researchers will be able to investigate services and outcomes for rare disorder groups and compare results nationally and internationally. National health assessments will include rare disorder data, examining health trends, wait times, service accessibility, affordability, and confidence in practitioners and services.

### **We will know how well our system works for Māori and tāngata whaikaha with rare disorders**

Māori will design and use systems to assess and track progress in health and health care for whānau Māori living with rare disorders. Reports in partnership with Māori will inform how we assess equity of outcomes and health system delivery, and how we monitor programmes to improve these. Working in partnership will help us to continuously improve outcomes.

### **Health research will include programmes on rare disorders and health services for rare disorders**

New technologies and research tools for clinical applications are rapidly developing. Aotearoa New Zealand contributes to genetic, metabolic, and other research fields relevant to rare disorders, with increasing involvement. Aotearoa New Zealand will be well connected into international clinical trial networks and have the infrastructure to support this. A coordinated approach to learning, translating, and embedding health service

advances, including local research and international research findings, will be visible.

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

### **What we've heard**

Many people and their whānau have expressed frustration with how long it takes to get a rare disorder diagnosis, involving multiple practitioners and services who often lack knowledge about rare disorders. People and their whānau feel they need to 'fight' for attention or to be heard. Support groups may have better access to information than practitioners. People with rare disorders emphasised the importance of including rare disorders in health and social services education. Whānau Māori want to see greater Māori representation in health and social workforces and want their health care journey to recognise and value a worldview from te ao Māori. Whānau Māori have often found hospital care alienating. They place high value on having a kuia or health worker available for support through such experiences.

### **Why it matters**

Health practitioners and care workers are highly motivated to help people and to do excellent work. When they are

alert to the possibility of a rare disorder and can easily get information, guidance and help, practitioners can make a sizeable difference to the experiences of people and their whānau living with rare or unknown disorders. This priority aligns with the wider health system focus on supporting the development of sustainable, diverse, skilled and confident workers for the future of the health system. The Pae Ora Strategies underpin this focus.

## **Aspirations for the future**

### **Health practitioners and care workers will know about rare disorders and how to find out more**

Students of clinical disciplines (medicine, nursing, midwifery and allied health, such as pharmacy, physiotherapy and psychology) will learn about rare disorders through integrated examples in their coursework. This approach will make them alert to the possibility of rare disorders and teach them how to support people while a rare disorder is investigated and care plans are developed. Ongoing learning for health practitioners will also include rare disorders, with options for more detailed supported learning. Additionally, care workers, health promoters, educators and social care roles will have learning opportunities that include rare disorder support skills.

## **Health practitioners and care workers will be able to support people and their whānau who may be living with rare disorders**

Students of health disciplines will engage in tikanga Māori practices in health and understand the impacts of showing manaaki and respect for diverse consumer preferences and needs. They will learn about supportive and strengths-based inquiry to identify wellbeing needs in uncertain circumstances. All health service workers will have opportunities to learn how to support whānau to decide on their own health goals and plans for mauri ora, whānau ora and wai ora, and on including mātauranga Māori. Ongoing clinical education will include cross-disciplinary and experiential learning options in supportive, counselling, tikanga, and social wellbeing areas.

## **A wide range of clinical support tools will support diagnosis and care for rare disorders**

Clinical guidelines and standards, along with referral, diagnostic and care pathways, will support care for a growing number of rare disorders. After Aotearoa New Zealand authorities have approved these tools, they will be adapted for different localities or situations where necessary. Health practitioners will be able to access the tools in the standard approved collections that they most often use. Because the tools will be integrated with patient management systems and electronic health records, they will also be automatically visible.

## **Specialist help, advice and backup will be available for practitioners at all levels of the system**

Practitioners across the health system will have access to expert opinion and advice that helps to provide quality care for people with rare disorders. Community health providers will benefit from a diverse range of expert input including specialists, therapists, health promoters, cultural advisors and people with lived experience. Specialist practitioners will be able to receive expert input from practitioners outside and within their specialities. Practitioners will be able to inform people about support groups as an additional resource. International input will be available if appropriate expertise is not available in Aotearoa New Zealand.

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

### **What we've heard**

People and their whānau living with rare disorders want to be heard and seen in the health system. They want practitioners to relate to and respond to them as people first, rather than as bearers of health conditions, needs or problems; to feel that each of us matters, and is believed and respected. Practitioners should have time to provide information and support to allow people and whānau to

make their own decisions for their health and care. Whānau Māori feel respected when services are whānau-centred and delivered with te ao Māori in mind, including practices such as mihi and karakia. Parents of tamariki living with rare disorders want their tamariki to be acknowledged as the taonga that they are, rather than being identified by a medical term like ‘rare disorder’.

## **Why it matters**

People and their whānau living with rare disorders are the best informants about their own experience of health and ill health. Being listened to and understood in a welcoming and respectful environment helps build trust and comfort with services and practitioners. Rare voices should influence how care is provided at all levels – policy, design, commissioning, service delivery, research, improvement programmes, and progress and outcome monitoring. This aligns with broader priorities in the Pae Ora Strategies and is important to addressing the needs of those who already feel isolated and unsupported.

## **Aspirations for the future**

**People and their whānau living with rare disorders will be the most important contributors to their health and care**

Placing people and their whānau at the heart of health interactions will ensure that what is most important to them will drive priorities of health providers and the system.



Services will adapt to the cultures, circumstances, preferences and choices of people and their whānau. A holistic view will underpin all services, which will support health and wellbeing even when no diagnosis is available.

### **People and their whānau will have choices and be empowered to make decisions about their care**

People will have access to their own health records and will be welcome to bring in other support and expertise to ensure autonomy when making decisions about their own care. Systems will be set up to accommodate rare disorders. The starting point for will be that services meet people and whānau with a basic level of understanding and respect. Services will also meet whānau needs so they can nurture whānau members whether or not they have a rare disorder.

### **Voices of people and their whānau living with rare disorders will be heard clearly across the health system**

The health system will hear voices of people and their whānau living with rare disorders at all levels and across all functions. Rare voices, including whānau Māori voices, will be present in conversations on policy, design, commissioning and service delivery. Rare voices will inform progress and outcome monitoring, iwi Māori partnerships, research and improvement programmes, and a strategic overview of New Zealanders' health and wellbeing.

## **Communities will support the wellbeing of people and their whānau living with rare disorders**

The health sector will work with diverse communities to build, maintain and enable strong foundations for health and wellbeing that include all community members. Health promoters, occupational therapists, physiotherapists and others will support communities to include people and their whānau living with rare disorders. People and their whānau will have support to participate in everyday activities and maintain connections that promote wellbeing.

## **Priority 5: Joining up internationally to achieve more**

### **What we've heard**

People and their whānau living with rare disorders often experience both isolation and mental health challenges. Being able to connect with people, groups, specialists and experts across Aotearoa New Zealand and overseas can help people and whānau to learn more and feel less isolated. Many have highlighted the difficulty for a small country to have all the information it needs about rare disorders, particularly for ultra-rare or unidentified disorders, those with rapidly evolving knowledge, and those with active research programmes under way.

## **Why it matters**

As a small country, Aotearoa New Zealand cannot hold expert knowledge of every rare disorder on its own. Considerable attention to rare disorders is building internationally. Some countries are combining their efforts by sharing their knowledge and expertise. 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 international collaborations, globally and regionally, we can share in clinical knowledge, research and technologies to improve outcomes for people and their whānau living with rare disorders.

## **Aspirations for the future**

### **Aotearoa New Zealand will be a global partner and contributor on rare disorders**

Aotearoa New Zealand health and research leaders and organisations will engage in international partnerships focusing on rare disorders, covering areas such as research, outcomes, systems, affordability, and equity. People with rare disorders will also be participants and provide input. Aotearoa New Zealand will learn from other countries and contribute to global knowledge on rare disorders, including reporting information to global monitors and sharing information from research, evaluation, and health monitoring.

## **Aotearoa New Zealand will play a leading role in rare disorders Indigenous health and equity**

Aotearoa New Zealand Indigenous health and research leaders and organisations will be participants in international Indigenous health partnerships that have a focus on rare disorders. Whānau Māori and tāngata whaikaha will be active participants and inform the partnership goals and work programmes.

## **International collaborators will work on faster uptake of new technologies and treatments to enhance care provision**

Aotearoa New Zealand will receive expert input to inform care, especially for very rare disorders and in areas with new developments. Health leaders, researchers, assessors, regulators and funders will be involved in international efforts to improve diagnostics, treatments, and technologies for rare disorders, aligning processes and standards across countries. Research and industry agreements will be in place to enhance global efforts towards the affordability of advances in rare disorders. Aotearoa New Zealand experts will provide advice and support to neighbouring countries, especially across the Pacific Islands region.

The full version of the strategy can be found here:

<https://www.health.govt.nz/publication/aotearoa-new-zealand-rare-disorders-strategy-rare-voices-and-aspirations>)

**End of Summary – Aotearoa New Zealand Rare Disorders Strategy 2024 – Rare voices**