

Rare disorders strategy: responsiveness to rare disorders in our future health system

Manatū Hauora x Rare Disorders NZ rare disorders strategy webinar

Summary of Input

Overview

Manatū Hauora | Ministry of Health and Rare Disorders NZ jointly hosted two webinars for people from the rare disorders community. Objectives for attendees were to:

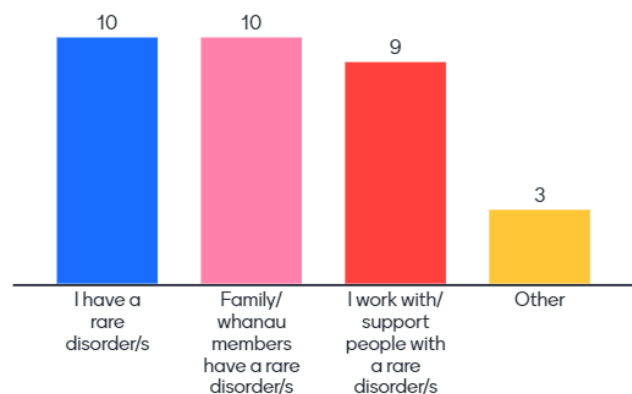
- learn about what a rare disorders strategy will provide and an update on what Manatū Hauora is doing to achieve this;
- share what they want to see changed and what would make the biggest difference to their lives and the lives of their whānau; and
- have their say as a community on the most important features needed in a rare disorders strategy for Aotearoa New Zealand.

Summary of Engagement

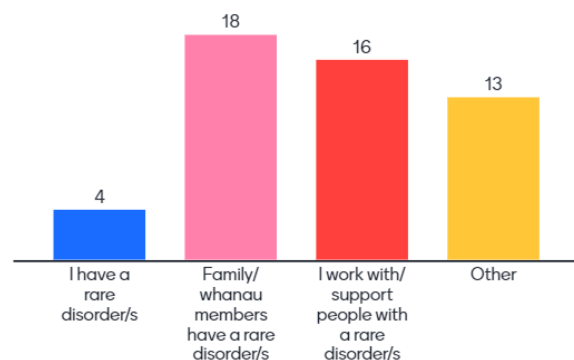
To allow participants to become comfortable with the Mentimeter mechanism used, we ran through several warm-up questions; 'I have joined this webinar because?', 'I live in a...' and 'I identify as...'. The answers below are not a full reflection of the number of active participants throughout the webinar as some joined late or chose not to answer these questions.

I have joined this webinar because?...

Webinar One:



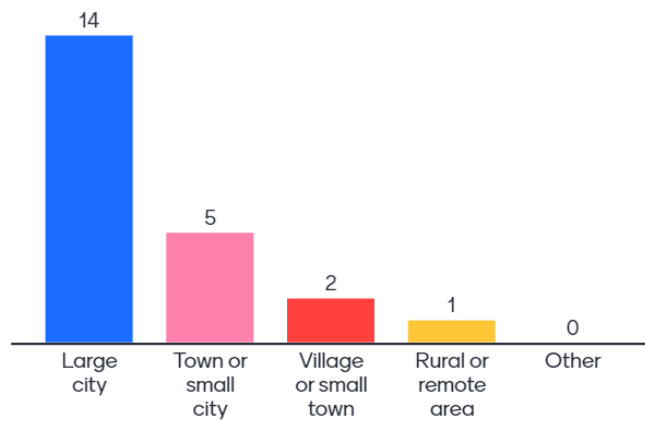
Webinar Two:



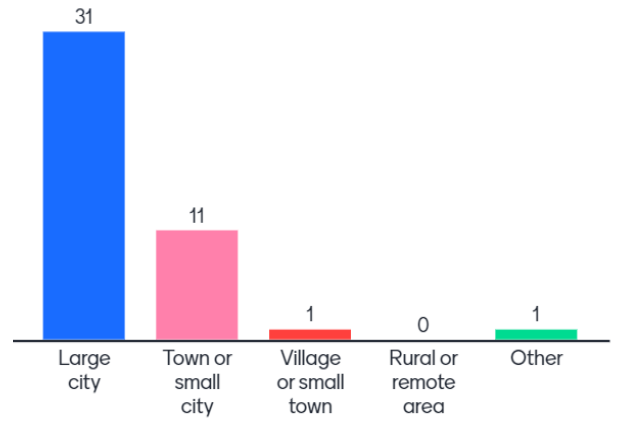
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I live in a...

Webinar One:

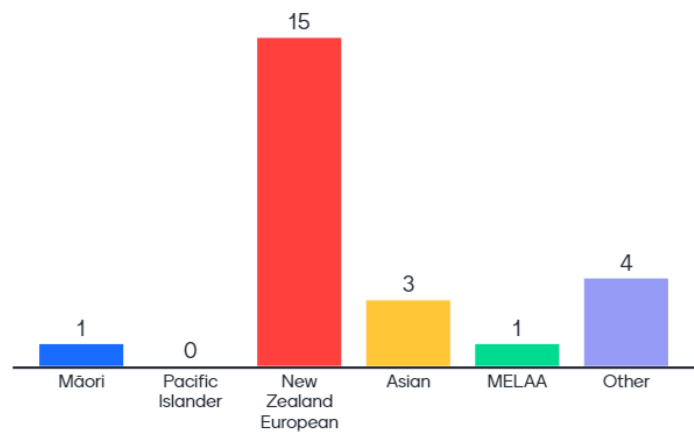


Webinar Two:

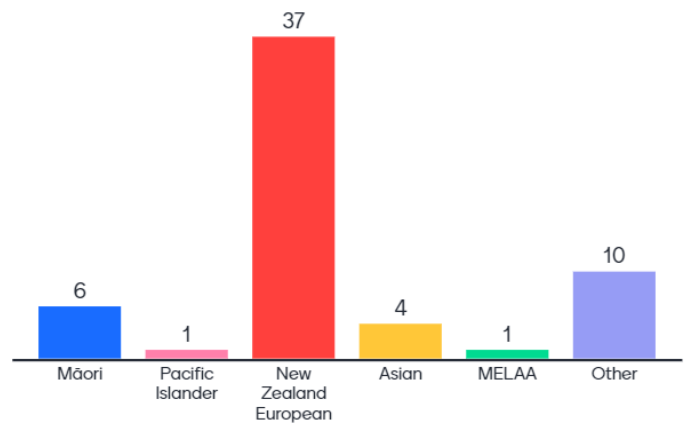


I identify as...

Webinar One:



Webinar Two:



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Free Text Questions

We received many valuable responses through the free text section of the webinars. To ensure all opinions, ideas and voices are acknowledged, these have been organised and themed, with exemplifier comments as summarised below.

What's working well in the existing systems for you and your family/ whānau in terms of your health and wellbeing, and why is it going well?

Access to Health Services

Access to health services presented as a common theme throughout the webinars. It is clear from these responses that access to relevant health services can make a significant difference to the rare disorders community:

"Genetic diagnosis as services are well-established and clear referral pathway from neurology."

"My GP took a proactive approach to making referrals that led to my diagnosis."

"Improved knowledge of local Paediatricians has helped with my quality of life."

"Haematology Nurses have been well versed with my disease and provide me with great secondary care and support."

"The existence of the Genetic Health Service."

"People providing services are great!"

"My dad had a fabulous specialist."

"Good follow-up and care from my specialist."

"What's working well is the care and support I receive from my GP and from a consultant at my local hospital who took an interest in my case and continues to monitor my heart in annual clinics. I have also been able to research and learn about my disorder."

Disability and whānau support/coordination

Disability and whānau support/coordination presented as a common theme throughout the webinars. It is clear from these responses that a wide range of community and social sector support, and coordination of care is important to the rare disorders community:

"Some great community-based support available."

"Having a support network with other families who understand."

"Support from NGO's."

"Community volunteers."

"Support from RDNZ helps people feel not so alone."

"International and national support groups."

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Funding, Assessment and Prioritisation

Funding, assessment, and prioritisation presented as a common theme throughout the webinars. It is clear from these responses that these are valued by the rare disorders community:

"Financial assistance."

"Individualised funding."

"Free prescriptions."

"Subsidised prescriptions and consultations!"

"Genetic diagnosis services are well established."

"My GP was proactive with their approach to assessment which led to my diagnosis."

Knowledge Building and Sharing

Knowledge building and sharing presented as a common theme throughout the webinars as a key area that is positive and uplifting for the rare disorders community:

"Increased exposure for rare disorders."

"Rare disorders is in the news more often, so people find out about them."

"I appreciated the opportunity to provide patient experience for the rare disorder advisory panel of PHARMAC."

"More awareness of my disease overall."

"There is often constructive curiosity among clinicians about my rare disorder. I'd love to see that fostered through further education about rare."

What's not working well in the existing systems for you and your family/ whānau in terms of your health and wellbeing and what could make things better?

Access to Health Services

"Lack of access to specialists with appropriate knowledge for my condition."

"Inconsistent levels and standards of clinical care across the country."

"The experience I had in the hospital when I was diagnosed with [condition] left me feeling helpless."

"Diagnosis, access, expertise, lack of MDT and cross services access, timeliness."

"Doctors don't talk to other doctors across the country who have clients with the same rare disorder."

"I waited almost a year to get access to genetic services, no specialists would take any action or make plans for me while I was waiting. Precious preventative treatment time was wasted."

"No pathways in place to transition teen patients from paediatric to adult care within the health system."

"Doctors and specialists are not always able to coordinate care in a timely way; patients are often left to connect people and keep them up to speed."

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"Adult healthcare is less collaborative between disciplines compared to paediatric healthcare."

"Differences in the level of specialisation and clinical resources means healthcare is experienced differently for people across the country."

Disability and whānau support/coordination

"Coordination of care for complex conditions with multisystem involvement."

"Respite for young adults is non-existent in our city."

"The impact on my family is not considered when dealing with my health."

"You have to be your own advocate and when you are battling and already exhausted this is hard to do."

"Not enough mental health support for parents raising a child with a rare disorder."

Funding, Assessment and Prioritisation

"Not enough funding for rare disorder medications."

"Access to newer, more effective medication not available through Pharmac."

"Lack of funding for new rare disorder treatments."

"I have had to move to Australia as the medication I need is funded there, Pharmac will not fund it."

"There seems to be a lot of confusion regarding funding and what is available."

"Inequity in prioritisation of care for rare or suspected rare."

"Genetic discrimination in health and insurance funding."

"My genetic diagnosis took over 9 months, it's too long!"

"We need more timely diagnosis for rare."

Knowledge Building and Sharing

"I was able to be diagnosed relatively quickly but given no information about what the diagnosis meant and no access to specialists with knowledge about it."

"Clinicians aren't aware of where to get help when needing specialist advice."

"Not enough training for medical students regarding rare disorders."

"It's very hard to find primary care GPs that have an understanding of my condition."

Research and Evaluation

"Rare disorders are not supported appropriately by large research funders, eg., HRC, despite the potential for significant impact."

"No investment into research for rare disorders."

"Ethical approval for research not having the appropriate expertise to recognise need vs. risk."

Data and Measurement

Data and measurement presented as a key area that needs a greater focus:

"Coding systems aren't working and in turn no data to represent our community."

"Inability to access clinical note across the country means gaps in care."

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"Not having the ability to leverage off international data including the NICE, FDA and TGA."

"No data collected for rare."

The change/s I most want to see from a rare disorders strategy is?

Access to Health Services

"Early diagnosis and clear pathways thereafter."

"Better connection/communication between medical fields."

"Rural communities to have the same access to services."

"Include rare disorders for newborn testing – early detection and early treatment."

"Better genetic testing and counselling."

"Specialists for rare disorders."

"Feeling like there is a place for us in the health system and processes that work for us, not against us."

"A centre of excellence!!"

Disability and whānau support/coordination

"Clear and readily available supports on the community that can cover physical, psychological and occupational needs."

"Paediatric support and support for caregivers as well as access to respite."

"The most impactful change would be to assign a navigator-type person to assist people with a rare disorder in managing their care across a wide range of health care providers."

Funding, Assessment and Prioritisation

"Recognition that leads to funding enabling prevention of morbidity and mortality through earlier diagnosis and appropriate treatment."

"Rare medication funding."

"The ability to apply and get funding for unfunded medications."

"Access to medicines pathway easier - without having support groups and busy clinicians having to do the hard work advocating for them."

"Earlier diagnosis and assessment/treatment pathways."

Knowledge Building and Sharing

"Greater awareness and education of rare."

"Educating healthcare professionals that may not have an understanding of rare disorders."

"Education for medical professionals about complexities and comorbidities, especially mental health for people with rare disorders."

"I'd like doctors to believe me when I tell them about my symptoms."

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"Acknowledgment for rare patients who are living with social stigma, by having better care and education opens up conversations across sectors including within the media."

Research and Evaluation

"More research of rare."

"Hoping that the strategy will trigger more research and training."

"A research framework so these disorders can be properly researched in NZ, and treatment/care can be based on evidence."

"A research strategy to bring new diagnostics and treatments to our community."

"Modern research being delivered worldwide is incorporated speedily into diagnostic ability here in NZ."

Data and Measurement

"Data collection for rare."

"That data on rare disorders is readily and rapidly available for health professionals."

"Must collect data so we know the scope of numbers in NZ."

"Being able to capture those that are currently being missed out."

"Coding system and data collection."

Digital Solutions and Enabling

Digital solutions and enabling presented as a key area that many would like to be better utilised within the system:

"Directory of physicians who have skills/expertise in certain rare disorders."

"Better digital and information systems that understand rare disorders."

"Systems that talk to each other, so information is carried through."

"A search engine or website for researchers, patients, etc to find labs and clinics focusing on different rare disorders."

"Digital centre of excellence."

The most impactful change for people with a rare disorder/s would be?

Access to Health Services

"Early diagnosis and treatment."

"Being able to live a normal life; access to good and timely healthcare."

"Patient-centred healthcare."

"Mobile specialist clinic."

"Improved access to everyday healthcare."

"Medical professional having adequate time to see patients."

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Disability and whānau support/coordination

"Coordinated care."

"Pathways and coordination to ensure best practice is followed and best supports are accessible without the person/whānau having to spend their lives chasing and advocating."

"Better support for families when multiple generations are affected. It's hard to care for others when you are affected yourself."

"We feel like there is no support and things are getting worse not better."

Funding, Assessment and Prioritisation

"Increased funding for..."

- support,
- respite,
- medications and treatments, and
- mental health support."

"Earlier diagnosis and equitable care."

Knowledge Building and Sharing

"Simply having rare disorders officially recognised as a vulnerable community within the health system."

"Recognition of complexity and mental health aspects of living with a rare disorder."

"Better awareness and understanding from healthcare teams about rare disorder."

"Visibility and recognition."

Research and Evaluation

"More precision-based research to help small groups of individuals."

Data and Measurement

"Start measuring rare disorder data, so that we understand the scope of the issue and can provide appropriate levels of support."

"Coding systems fixed, then we can have visibility and funding can be assigned appropriately."

Digital Solutions and Enabling

"A rare disorders database."

"A site where rare disease resources from clinics, research labs, online group therapy, etc can be accessed."

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The most impactful change for family/ whānau & carers would be?

Access to Health Services

"Genetic testing."

"Access to IF [individualised funding]."

"Screening (prenatal, preconception, newborn), earlier diagnosis allowing prevention of morbidity and mortality."

Disability and whānau support/coordination

"Support, support, support."

"Mental health support."

"Financial and psychological support."

"Better communication and coordinated support, rather than multiple different teams being in contact."

"For genetic disorders multi-generational support is required - which includes mental health, a plan for care in advance."

Funding, Assessment and Prioritisation

"Better access to home aids e.g., wheelchairs."

"Improved funding to access respite."

"Support for financial help due to not being able to work full time."

"Access to travel funding when accessing treatment appointments."

Knowledge Building and Sharing

"Access to education and awareness tools for public spaces like schools."

"Access to support and education for families."

"Sharing of tips and tricks from people with similar rare disorders."

"Early diagnosis, earlier intervention and access to individualised funding."

Digital Solutions and Enabling

"Joined up services - not needing to fill out a million forms with the same information."

"Less paperwork!!!"

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Is there anything else you would like us to consider?

Access to Health Services

"Rural areas don't have half the access to health and support as bigger areas. Need more willingness from clinicians to travel."

"Waiting a year for OT is crazy. I haven't been able to go to a grocery store for 3 years because of lack of support."

"Education of Nurse Specialists in rare disorders."

"Navigating the system alone as a younger person and not having any doctors with knowledge about my (life limiting) condition was incredibly frightening."

"My most recent Specialist advised "that's rare, I learnt about that 30 years ago at med school and haven't seen or heard of it since, so it won't be that". That clearly sums up my ongoing experience of seeking a diagnosis."

"Access to paediatric services early can be helpful and add extra support. Paediatric palliative care can be beside you on your journey over years if required."

"Investment in a national functional and cohesive diagnostic pathology sector would be a great start."

Disability and whānau support/coordination

"Support should be offered during diagnosis process. Meet the needs of patient not the diagnosis."

"We are losing people due to mental health, there is not enough support in this area!!"

Funding, Assessment and Prioritisation

"Need funding for specialist services whilst ensuring these are equitable for all."

"It is very expensive looking out for a disease that needs real funding provided. First accurate diagnosis, and real treatment options and good guidance. there is a lot to go before this can happen!"

"Funding to identify unmet needs."

Knowledge Building and Sharing

"We are respectful that doctors cannot know all there is to know about all rare disorders. However, doctors should be given time/resources to learn when they are working with a new patient with a RD."

"International collaboration and building on work of other countries that are doing better than us, don't reinvent the wheel."

"We would like to work with the community to help learn more about rare disorders and develop new solutions to diagnose and treat, give lived experience a seat at the table."

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Research and Evaluation

"Work with other countries who are ahead of us, take action based on their research, rather than spending years doing our own."

"Connect with international research, work with Australian counterparts - where much of this work is underway."

"There is a knitting together of rare disorders. research overseas i.e., 60% of X patients also have this and this and this rare disorders....."

"Greater innovation, science and research is moving so fast, we need to keep up."

Data and Measurement

"The importance of registries for rare disorders and not being lost in system."

"Coding for rare disorders."

"Utilising data and information from other international bodies, including TGA, FDA & NICE. We're living with rare diseases and data is limited."

"Good data collection is needed for research, funding and policy development."