IMPACT OF LIVING WITH A RARE DISORDER IN AOTEAROA NEW ZEALAND IN 2025

Key priorities to deliver improved outcomes for people living with rare disorders, their family and whānau

February 2026

HealthiNZ

Advancing life-changing solutions

Acknowledgements

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Whilst the views and recommendations are wholly those of HealthiNZ they have been informed by the 2025 Voice of Rare Disorders survey findings and by the growing evidence base and overseas policy development to improve outcomes for people living with rare disorders. This would not have been possible without the contributions and insight provided by Rare Disorders NZ.

We also acknowledge the support of the many New Zealand rare disorder patient support groups and people living with a rare disorder who contributed to the Voice of Rare Disorders Survey that informed the recommendations outlined in this white paper.

About HealthiNZ

Andrew Cameron is the Principal Consultant at HealthiNZ, providing a range of strategic advisory healthcare services in NZ and overseas. Andrew has over 20 years' commercial healthcare experience, with a passion for healthcare innovation and new technologies that deliver patient outcome-led advances in healthcare and wellbeing. Believing that in order to ensure health innovations are truly people centred it is critical that we first listen to the voice of the patient.

Beyond his role at HealthiNZ Andrew has a personal interest in rare disorders as the parent of a child who has cystic fibrosis and, until recently, sat on the Board of their patient organisation Cystic Fibrosis NZ.

Executive Summary

The results from the 2025 New Zealand Voice of Rare Disorders Survey provide an opportunity to understand the real impact of living with, or caring for people with, rare disorders. The survey captures the experiences of over 1,000 New Zealanders living with rare disorders and, with over 460 different diagnoses and good participation from Māori, remains the largest reported survey of consumer reported outcomes for rare disorders in this country.

The survey findings show systemic barriers persist in diagnosis, care pathways, access to medicines, social supports, and access to appropriate expertise for people living with a rare disorder. International comparisons reveal that New Zealand continues to lag behind OECD peers in access to modern rare disorder medicines and coordinated care.

For people living with a rare disorder, and their carers, the picture continues to be a challenging one. Most report that the rare disorder impacts significantly on their health and everyday life, is disabling and makes a number of everyday activities difficult. Many are self-funding costs of their healthcare to some extent, with the majority finding this hard to manage. Full employment is a challenge for people and their carers, often requiring modified work arrangements or the need to leave their jobs. Their rare disorder has serious effects, not only on their own mental health and wellbeing, but also on their family/whānau, with one in three often unhappy and depressed and feeling they cannot overcome their problems.

The following are key findings from the 2025 survey:

- **Delayed and inaccurate diagnosis:** Over half (52%) of respondents waited more than a year for a diagnosis, and 23% waited more than 5 years. Misdiagnosis remains common, with over half (56%) misdiagnosed at least once, while access to genetic testing is limited.

- **High healthcare utilisation, yet care remains fragmented and poorly coordinated:** Most respondents (84-93%) are accessing GPs and specialists far more frequently than the general population, and hospital admissions and ICU stays are significantly elevated. However, communication between providers is often inadequate, and most people are not informed about their rights or available support.

- **Limited access to rare disorder medicines:** While almost all respondents took at least one medicine, most are for symptoms rather than the rare disorder itself. Over three in ten (39%) people believe they are missing out on needed medicines, and almost all (96%) worry that future treatments will not be funded. The Named Patient Pharmaceutical Assessment (NPPA) process is little known and inconsistently successful. The current funding model does not meet the needs of those with rare disorders, and alternative pathways are urgently needed.

- **Variable access to social and disability supports:** People often faced significant barriers in accessing appropriate social and disability supports, with most feeling uninformed about available services and reporting that professionals are not well prepared to meet their needs. Only a small proportion (15%) received respite care, and over half (58%) believe that disability support services do not adequately consider the unique challenges of rare disorders.

- **Persistent equity gaps:** These are commonly experienced by people with rare disorders, which is further exacerbated for Māori, who experienced greater impact of the rare disorder for the person, their carer, and for their whānau/family. These findings reinforce the need for targeted, culturally responsive actions.

The finalisation of the Aotearoa New Zealand Rare Disorders Strategy by the Ministry of Health in 2024 marked a pivotal step forward, articulating a vision where "people living with rare disorders and their whānau are enabled to live their best lives" and where "no one is left behind." To date, implementation of the Strategy has been stalled but there is now cross-agency agreement to develop an implementation plan. To realise the Strategy's vision, a coordinated, measurable, and equity-focused plan for implementation is essential with key actions across each of the following areas:

- **Implementation plan** addressing the full ten-year life of the strategy, ensuring a clear line of sight from early actions to the intended end-state, with funding and resource commitment and with cross-agency leadership and oversight.
- **Leadership and coordination mechanisms** established for rare disorders.
- **Prioritise equity** for people with rare disorders compared to the general population, ensuring that efforts to close this gap lift all parts of the rare disorder community, including Māori, through culturally safe care and support.
- **Rare Disorders NZ** as a key partner in strategy implementation and ongoing advocacy to ensure the voice of rare disorders is heard.

While the implementation plan will go a long way towards addressing gaps highlighted in this survey for diagnosis, planned pathways for clinical care and workforce development, the Strategy is scarce on detail about improving access to modern rare disorder medicines or access to disability and social supports, including support for carers. The 2025 survey has shown these remain two areas with large gaps for people living with rare disorders.

Furthermore, the Strategy also highlighted the importance of improved data collection about people living with a rare disorder and this will also be a key element of the implementation, in order to better understand the problem and measure progress.

Taken together, these actions will be key to build momentum to deliver better outcomes for people living with a rare disorder in Aotearoa.

Significant challenges remain for people living with rare disorders

Rare disorders are individually uncommon but collectively significant, impacting health, wellbeing, and social participation for people living with them, their families, the health system and society.[1] This can include significant impacts on mental, social and physical functions, household budget, employment and job careers, family life and well-being. Many of these conditions are life-long and debilitating and may lead to death at a young age.

People often report being lost in the health system, starting with a long pathway to diagnosis, and difficulty in accessing treatments, care services or healthcare services, including challenges surrounding coordination of care. People can experience inequitable health outcomes and these outcomes are often influenced by unfair barriers, such as the lack of timely access to services and trying to navigate a health system ill-equipped to support them.

I am always up against a brick wall. – Person with a RD

We have no information on what the future may hold. We are on a roller-coaster ride blindfolded. – Parent of a person with a rare disorder

[1] ANSEA report, 2019

New Zealand Rare Disorders Strategy Implementation

The long-awaited Rare Disorders Strategy developed by Manatū Hauora |The Ministry of Health was finally released in July 2024.[2] This was the first national strategy for Pae Ora | health and wellbeing for people and whānau living with rare disorders in Aotearoa New Zealand. It prioritised actions for improved health and wellbeing, creating a roadmap and pathways that ensure people living with rare disorders are not left behind.

The Strategy provides a framework to guide long-term priorities for health entities over the next 10 years in improving health outcomes for people with rare disorders and their families or whānau. It upholds the principles of Te Tiriti o Waitangi. Its implementation will allow the health sector to provide better support for people with rare disorders, as well as make it easier for people, practitioners, and organisations to get the information and support that they require.

Attention has now turned to the implementation plan for the Rare Disorders Strategy with commitment evidenced through recently confirmed expectations from the Minister of Health in the outcome areas of **Implementation Planning**, **Leadership and Oversight**, **Ongoing Coordination** and **Accountability and Reporting**.

The 2025 Survey also provided the opportunity to get direction from the rare disorders community on what should be prioritised in the implementation of the Strategy. In order of priority, these were for[3]:

- *Facilitating faster diagnoses* (68%)
- *Providing a central place where health professionals can easily find trusted information about rare disorders, like care standards, treatment guidelines, and referral pathways* (43%)
- *Supporting families with coordination of care* (34%)
- P*roviding information about disability, social support and services* (25%)
- L*eading workforce development on rare disorders and championing the roll-out of coding for rare disorders* (23%)

This community feedback has guided the recommendations outlined in the report.

2 https://raredisorders.org.nz/about-rare-disorders/rare-disorders-strategy/

3 People were able to select more than one option

2025 Voice of Rare Disorders Survey

The Voice of Rare Disorders Survey was first developed in 2019 to better understand the impact of living with a rare disorder in New Zealand and has since been repeated in 2021, 2023 and now 2025.[4] These surveys have been conducted by Rare Disorders NZ on behalf of rare disorders support organisations and individuals, with anonymised data provided to HealthiNZ to inform this white paper.

The survey was designed to be self-completed online and covered the following areas: Health Profile, Healthcare services, Treatments (Medicine), Coordination of Care, Cost of the Disorder, Disability Supports, Employment, Care Services, Whānau/Family and social life, Stress and Wellbeing. Eligible participants were people with a rare disorder in New Zealand or a family member and carer and over 18 years of age.

The survey was designed from similar robust patient voice surveys from European countries. This included the EURORDIS Rare Barometer Programme which to date has over 22,300 people registered.[5] Following from the experience in previous surveys, some additional questions were added. Where appropriate, comparisons with the population from the 2019, 2021 and 2023 surveys were made and reported on in the results section. This section also includes comparative analysis of Māori vs non-Māori responses. Overall, this survey further expands the evidence-base of people living with a rare disorder in New Zealand.

Rare Disorders NZ currently engages with 156 support groups representing at least 33,000 people living with different rare disorders. All groups were encouraged to communicate to their members, stating the project objectives and providing a link to a web-based questionnaire. The data collection period ran from 9th September to 1st December 2025.

The survey was screened and advised it was out of scope for HDEC review and approval. Given this, there was a robust data collection and analysis plan put in place.

Summarised results were presented as a report and this forms the basis of this white paper.

4 2025 NZ Voice of Rare Disorders Survey

5 https://www.eurordis.org/rare-barometer-programme

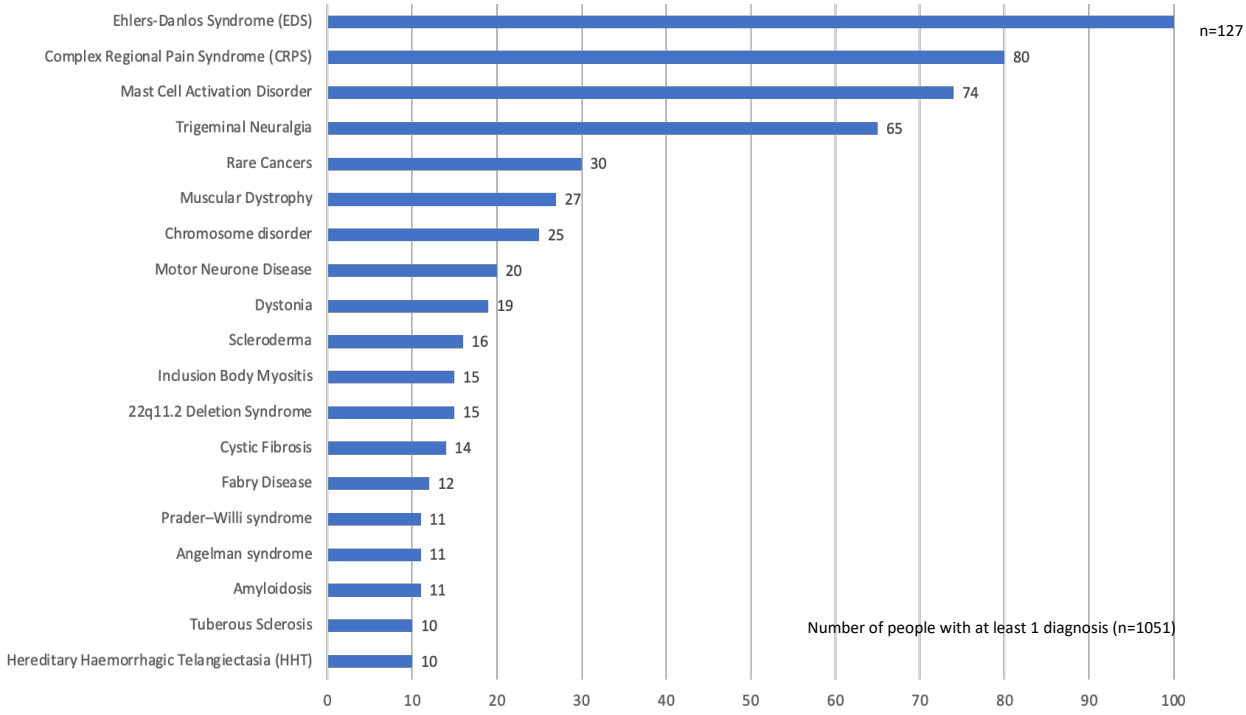
Results

Health Profile

Overall there were 1051 responses with a confirmed diagnosis included in the primary analysis.[6] This remains **the most significant survey of consumer reported outcomes for people with rare disorders in NZ**.

The primary analysis reported on the overall population of people with a confirmed rare disorder diagnosis. There were 469 different rare disorder diagnoses with a number of rare disorders having more than 10 patients diagnosed in New Zealand, including *Ehlers-Danlos Syndrome (EDS)*, *Complex Regional Pain Syndrome (CRPS)*, *Mast Cell Activation Disorder*, *Trigeminal Neuralgia*, *Muscular Dystrophy*, *Motor Neurone Disease*, *Dystonia*, *Scleroderma*, *22q11.2 Deletion Syndrome*, *Inclusion Body Myositis*, *Cystic Fibrosis*, *Fabry Disease*, *Amyloidosis*, *Angelman syndrome*, *Prader–Willi syndrome*, *Hereditary Haemorrhagic Telangiectasia (HHT)*, *Tuberous Sclerosis.* In addition 30 people were diagnosed with a rare cancer and 25 people diagnosed with a chromosome disorder.

Figure 1 Most common rare disease diagnoses in 2025 *Voice of Rare Disorders Survey*



Overall, 23% of people reported more than one diagnosis, with 7% reporting 3 or more diagnoses. The inclusion of a wide range of diagnoses in this survey reflects Rare Disorders NZ's expanding engagement reaching these communities.

6 48 responses were excluded from primary analysis for reasons that included duplication, non-consent or lack of confirmed diagnosis

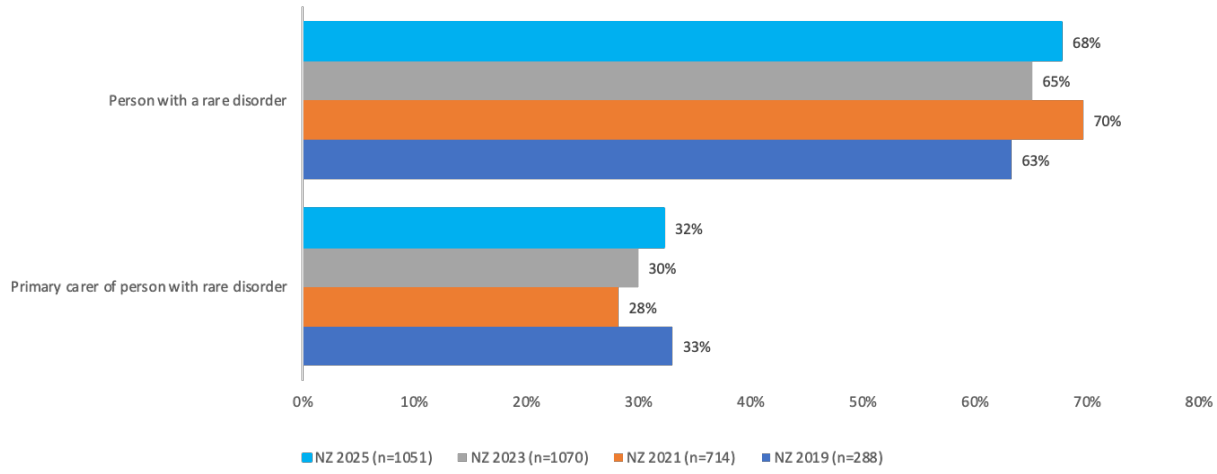
The majority of people (78%) described at least one of the ethnic groups they belonged to as New Zealand European. The proportion of people that included Māori as an ethnic group was 10%, much closer to National Census estimates[7] than earlier surveys and similar to 2023.

In line with population, the majority of responses were from Auckland, Canterbury and Wellington regions, similar to previous survey trends. In comparison with overall Region population estimates the Auckland region was under-represented and the Wellington region was over-represented in the survey responses.

Overall, over **80% of responses were from people living in urban areas** or areas with moderate to high urban influence. However there was an increase in responses from rural areas with low urban influence (10%) compared with 2023 findings.

The **majority of responses (68%) were provided by people with a rare disorder**, which was similar to 2023 (65%) (Figure 2). Overall, 32% of the responses were from the primary carer of a person with a rare disorder.

Figure 2 Relationship to person with rare disorder

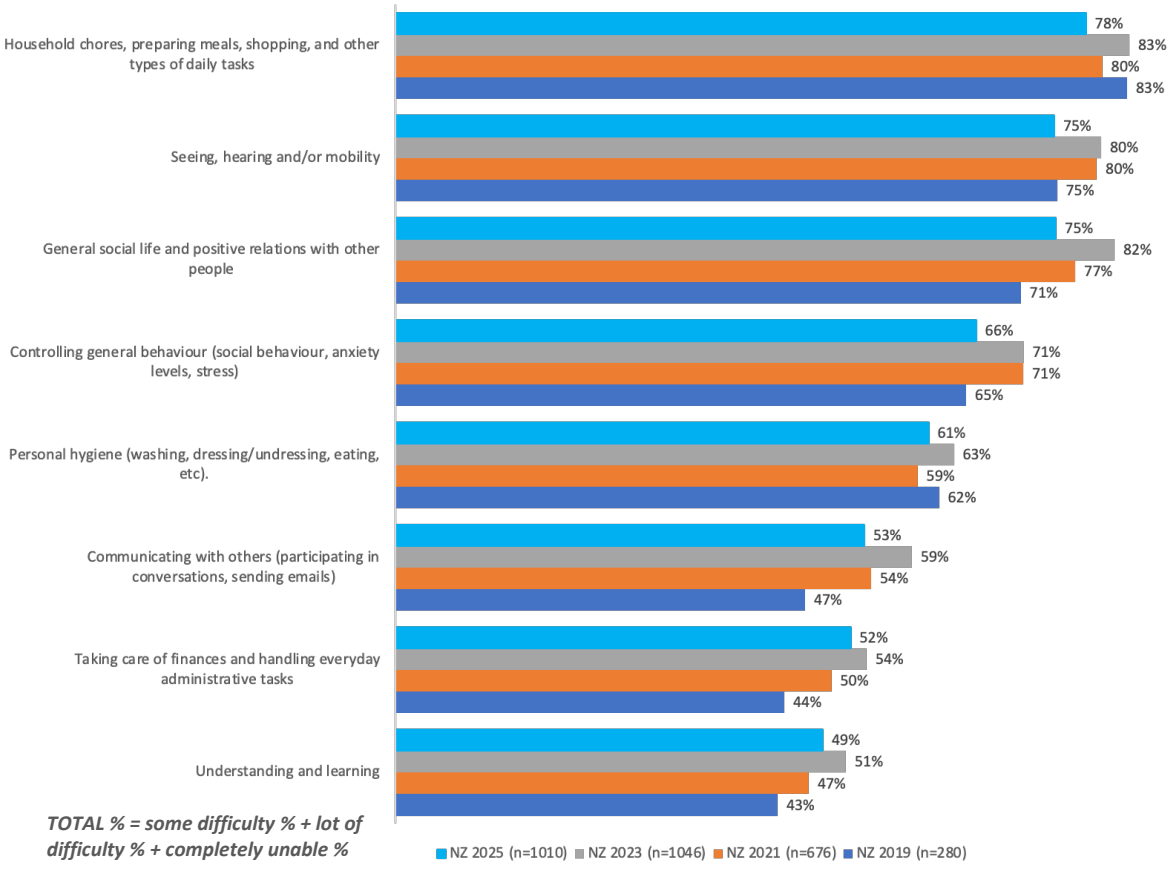


Daily impact of living with a rare disorder

The extent to which the person with the rare disorder could perform certain activities varied with the activity. For **49-78% of people and their families surveyed, the rare disorder makes a number of everyday activities difficult** (communicating, controlling behaviour, social life) and was similar to previous surveys.

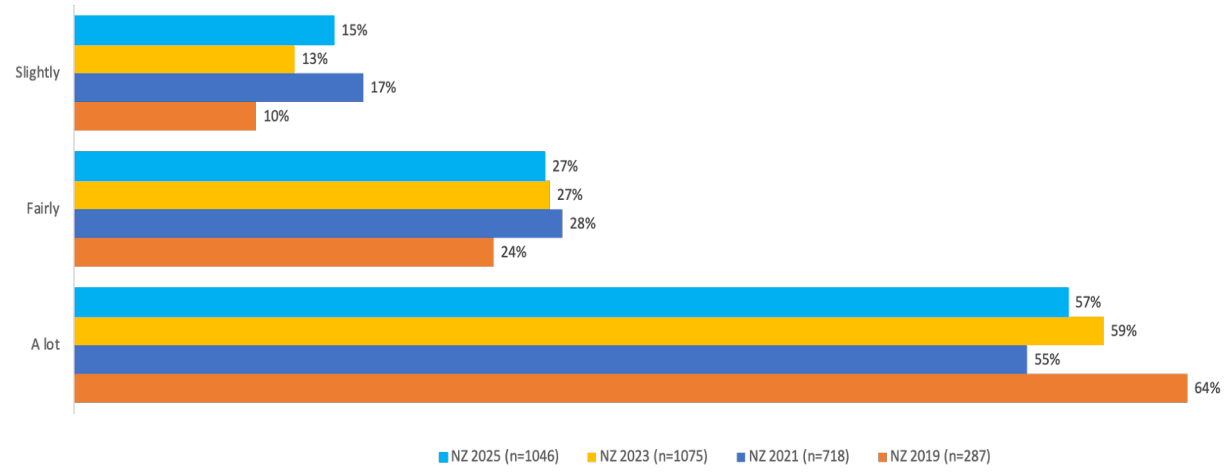
7 https://www.stats.govt.nz/information-releases/maori-population-estimates-at-30-june-2025/

Figure 3 Activities of daily living that provide greatest difficulty



The most frequently reported household carer was the person living with the rare disorder > mother> the spouse, in that order. For the 67% that needed assistance from others, an average of 2.3 whānau/family members and/or friends were involved in their care and support during an average week.

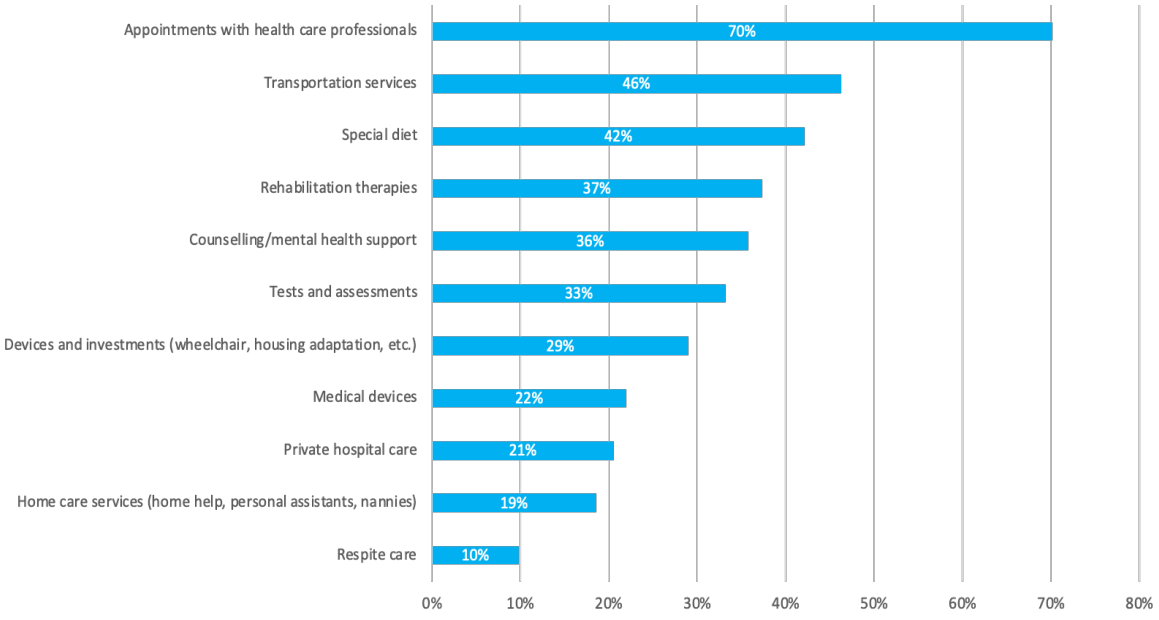
Figure 4 Impact on health and everyday life



Cost of living with a rare disorder

There were many costs covered by people living with the rare disorder or their family, with approximately half self-funding or partially self-funding healthcare costs to some extent. The most frequent self-funded costs were appointments with healthcare professionals (70%), transportation services (46%) and special diet (42%).

Figure 5 Level of self-funding for healthcare cost



53% felt the costs associated with the rare disorder for people and their whānau/family were hard to manage.

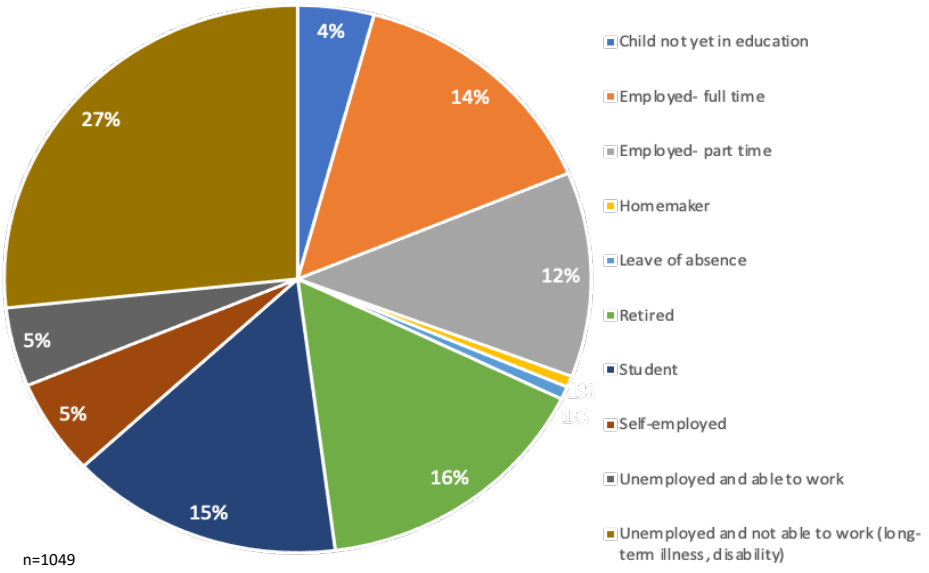
We had to go public to fundraise for equipment that would be beneficial for her development. It's humiliating. – Parent of a child with a RD

I am drowning in medical expenses and the guilt that comes with being a financial burden on my family on top of everything. – Person with a RD

Employment

In total, 40% people with a rare disorder were in some form of employment, either being full-time, part-time or self-employed (excluding children and students). **However, 39% were unemployed, with the majority citing long-term illness or disability.**

Figure 6 Employment status of person with rare disorder



At 46%, the proportion of employed **people in part-time employment was higher than the NZ average** indicating underemployment[8].

It's likely that if I'd had the correct diagnosis and treatment I would have been working for those 18 years. Yes I'm angry. – Person with a RD

I used to work full time. Then I couldn't work at all. I've now started a different job. It's part time, one that is less intense, but it comes with a much lower pay rate. – Person with a RD

A number of people had to leave previous jobs due to their rare disorder. For those, 23% were too unwell to continue working and 11% had to retire early due to their illness or disability. A further 6% resigned because their previous job was not adapted for their condition.

8 https://www.mbie.govt.nz/dmsdocument/30344-labour-market-statistics-snapshot-december-2024

There was an impact on employment for the main carers of the person with a rare disorder. In cases where there was a main carer for the person with the rare disorder, 42% of the carers were not in paid employment, with a further 18% in part-time paid employment only. Furthermore, 44% of carers reported changes in their employment circumstances due to being the carer of a person with a rare disorder, with the most common reason for change being working part-time or reduced hours due to caring responsibilities (20%), followed by the carer needing to look after the person with the rare disorder full time (16%). Overall, there were many ways the person's educational, professional or working life was impacted by the rare disorder.

Trying to keep employment and caring for two children that needed my attention because of serious medical problems has caused a lot of stress and struggle. – Parent of children with a rare disorder

Due to our son's condition my partner and I can't both work. On one wage the cost of living is crippling. We get no other financial support other than disability allowance. – Parent of a person with a RD

Family/whanāu and social life, stress and wellbeing

Since the symptoms started, the **majority of people have experienced increased tension between family members (53%) and isolation from family/whānau and friends (66%)**, amplified, or caused by, their rare disorder.

Its isolating for me as a carer...I've had to remove myself from work and step away from whānau who are unable to deal with behaviours. – Parent of a person with a RD

This has had a devastating effect on our family in every way and reduced the quality of life in particular for her younger siblings who now have very little to do with her. – Parent of an adult with a RD.

Concerningly, **one in three people often felt unhappy and depressed and felt they could not overcome their problems** (Figure 7, Figure 8). These issues were much higher than in general populations, especially given that an additional one in three people reported sometimes having these feelings[9].

9 https://www.health.govt.nz/publications/annual-update-of-key-results-202425-new-zealand-health-survey

Figure 7 Extent person felt unhappy and/or depressed in last month: Comparison over time and with general population

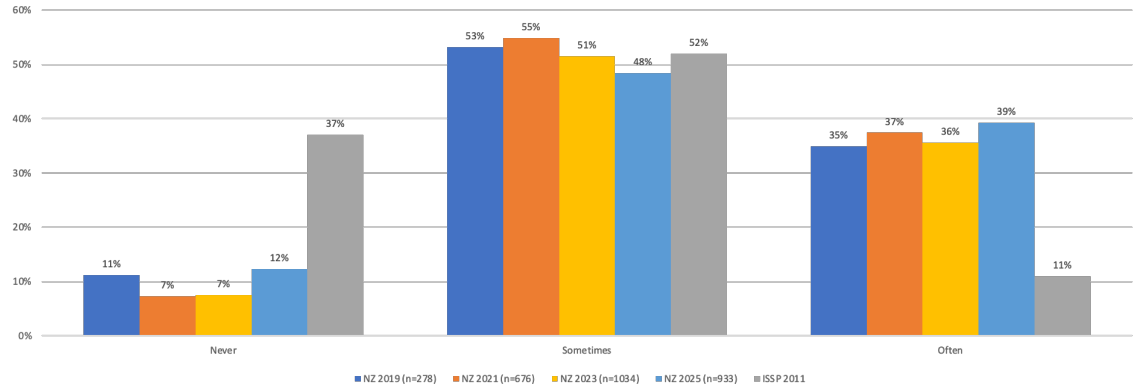
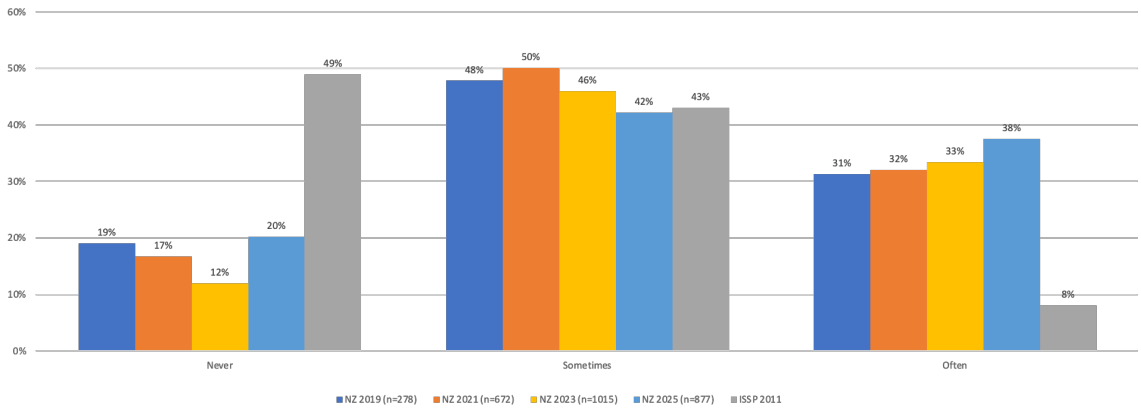


Figure 8 Extent person felt they could not overcome their problems in last month: Comparison over time and with general population



The impact on mental health and wellbeing extended to carers with **18-20% carers often feeling unhappy and depressed and that they could not overcome their problems.**

I struggle with my own mental health, because it was such an ordeal to even get a diagnosis. Each year, there seemed another new issue we faced medically with my daughter, with no known reason. The mental and emotional load has a huge impact. - Carer of a person with a RD

The importance of connection with others with the same condition should not be underestimated. However, while 66% of people or their carers thought that this was important, only 34% felt well-connected to others with the condition. Recognition and inclusion of mental health and wellbeing support for the person with a rare disorder and their family/whānau need to be part of integrated holistic health care.

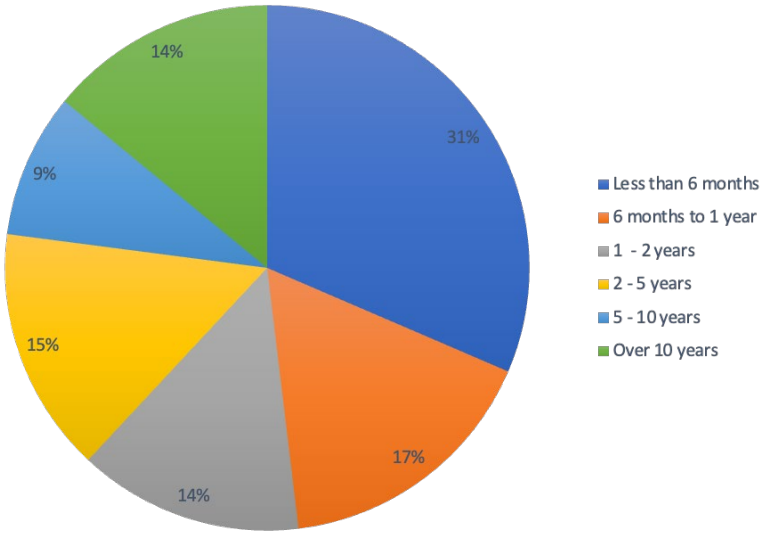
Diagnosis

Early and accurate diagnosis of a rare disorder remains important to accessing a range of treatments and healthcare services.

I feel that having an actual diagnosis gives me better access to specialist and other clinical services. Concerns are taken more seriously and there is an increased willingness to investigate issues and get referrals. Before my diagnosis my symptoms were dismissed for many years and attributed to stress. – Person with a RD

The majority of people had challenges in getting an accurate diagnosis for their rare disorder, similar to previous surveys. Over half (52%) took longer than 1 year to get a diagnosis and 23% waited more than 5 years. **For almost one in seven (14%) the time taken to get a diagnosis was over 10 years** (Figure 9).

Figure 9 Time taken to get diagnosis



The majority (67%), had to visit 3 or more doctors to get a diagnosis and **for 8%, over 10 doctors were visited.**

56% of people with a rare disorder were misdiagnosed at least once before the final diagnosis was confirmed. **30% were misdiagnosed at least twice.**

While almost **eight in ten (78%) people supported easier access to publicly funded genetic testing** to diagnose and better understand their rare disorder, a similar proportion were also worried that New Zealand's public health system will not keep up with developments in genetic testing for rare disorders.

Planned Pathways for Clinical Care

Healthcare Utilisation

There was a need to frequently access a wide range of healthcare services for people living with rare disorders, including their GP and medical teams, specialists, diagnostic testing and inpatient services (Table 1). Utilisation of healthcare services was similar to earlier surveys, albeit at an increasing cost.

Table 1 Healthcare service utilisation for people with rare disorders in the last year

Service	Utilisation rate (%)	Total average annual frequency	Cost per test/visit/day[10]	Average cost per patient per year
GP visits	93%	5.8 visits	$124	$721
Specialist visits	84%	5.2 visits	$386	$2,024
Diagnostic tests	83%	5.4 tests		
ED visits	40%	1.6 visits	$571	$964
Admissions	39%	1.0 admissions		
Inpatient days	38%	11 days	$1,200	$16,110
ICU Inpatient days	8%	4.2 days	$8,494	$35,562

Most people had reported seeing a specialist or GP (84-93% utilisation rate) with an average annual frequency of almost 6 visits for each. In terms of GP visits this is much greater than the general population, both in terms of utilisation (61%) and annual number of visits (3.1 visits).[11] There were also more than one in three people with a rare disorder that presented at the emergency department in the last year.

39% of people with a rare disorder were admitted to hospital and, for those admitted, spent an average of almost twelve days as an inpatient in the last year. In addition, **one in thirteen people with a rare disorder were admitted to ICU** and spent an average of 4.2 days in ICU**.** This is a significant finding and highlights the

[10] Based on updated Treasury estimates for the Cost Resource Manual (PHARMAC) through the CBAx Tool; https://www.treasury.govt.nz/information-and-services/public-sector-leadership/investment-management/investment-planning/treasurys-cbax-tool Cost per specialist visits has been applied at $386 (based on physician outpatient costs, subsequent visit); cost per GP Practice visit applied at $124; cost per nurse visit applied at $62; cost per emergency department visit applied at $571; cost per day for a hospital medical ward at $1,200 (not including procedures); cost per day for intensive care unit (ICU) at $8,494

[11] https://journal.nzma.org.nz/journal-articles/primary-care-doctor-and-nurse-utilisation-rates-for-billed-consultations-across-the-comprehensive-care-primary-health-organisation#:~:text=In%20New%20Zealand%2C%20reports%20of,methodological%20variability%20exists%20between%20studies

continuing impact and pressure on ICUs and other inpatient services, especially as admission rates are almost twice as high as in the general population.[12,13]

While most people were successful in getting a specialist referral from their GP or medical team, **for over one in four people, they had their referral to a medical, surgical or paediatric specialist declined**.

In addition, while the majority of people felt culturally safe when visiting health services, 7% considered visits to a nurse, doctor, health service, hospital service or genetic testing service to be culturally unsafe.

Coordination of Care

Coordinating care is challenging for people with rare disorders, and this can be further complicated by being under the care of multiple healthcare teams and limited access to information.

There is little to no understanding of the complex needs my family has, how to manage it, how to coordinate treatments, or anything. I feel completely overwhelmed and can't trust the medical system to know, treat, or otherwise most of our conditions. We fall between all the gaps and it is awful.
– Parent of a person with a RD

I am forever having to explain, be his advocate, then explain again! We keep him safe, not the health teams.
– Parent of a person with a RD

While four in ten people were confident their doctor had access to guidelines and pathways providing information on how to manage their rare disorder, for almost half it required a lot of time investment. For a further 24% they reported their doctor found it hard to know where to find information.

I haven't accessed all the recommended monitoring for my condition because there isn't a care pathway. I was turned down for some recommended screening tests that my GP referred me for (and experienced a possibly-avoidable medical event subsequently). – Person with a RD

[12] https://www.health.govt.nz/publication/publicly-funded-hospital-discharges-1-july-2018-30-june-2019

[13] https://www.health.govt.nz/new-zealand-health-system/accountability-and-funding/planning-and-performance-data/reducing-acute-readmissions-hospital

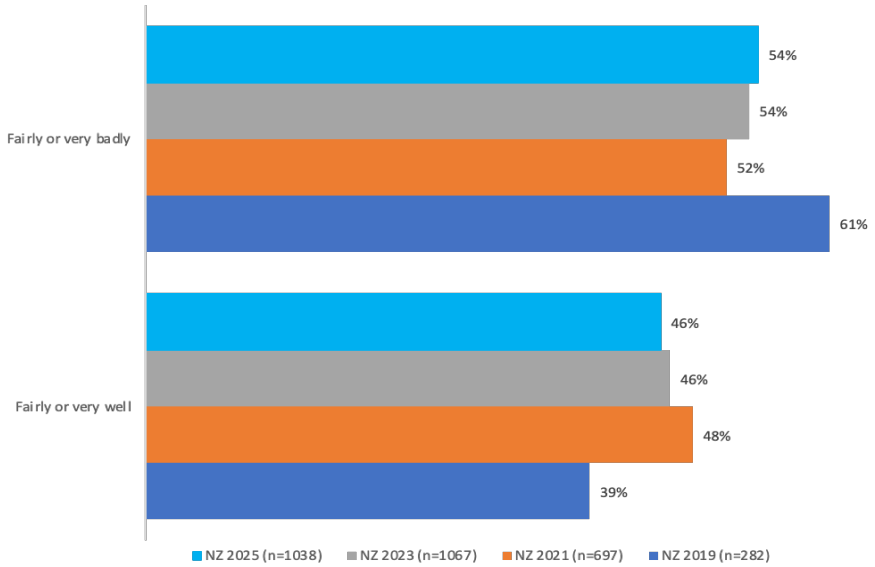
Overwhelmingly the internet was the main first source of information about the disorder, treatment and care, similar to 2023. This was then followed by various healthcare professionals as well as the 'Support Group'.

There was a lack of Information about my disease when I was diagnosed. No support system. Even my GP didn't know what it was. I had to research it myself. – Person with a RD

Facebook community groups for [the rare disorder] have been the most helpful tool. Advice from other people in the same situation, who have been dealing with this for years, is more useful than most medical professional help (since most medical professionals don't know what [the rare disorder] is). – Person with a RD

Over 50% felt that **communication and information exchange between different service providers was poor** (Figure 10).

Figure 10 Quality of communication between service providers

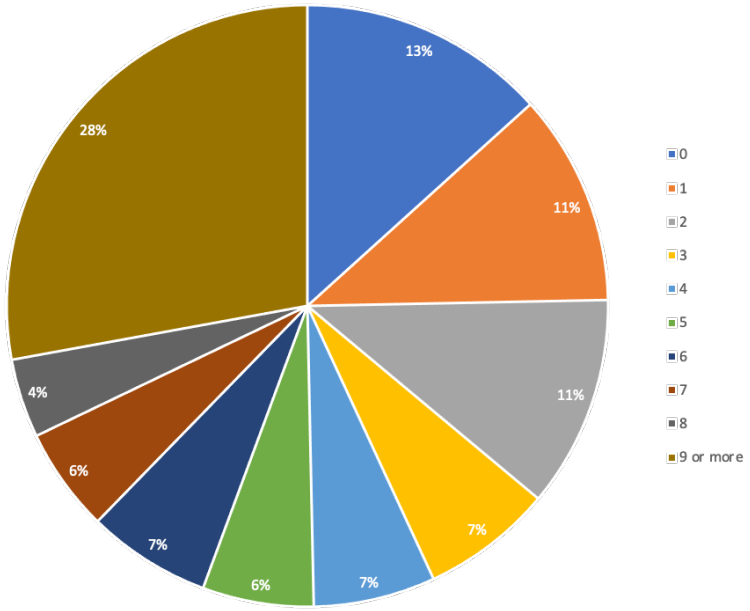


Medicines

The range of treatments people reported taking were mainly focused on reducing pain or inflammation, not treatment of the rare disorder itself.

87% people were taking at least one medicine (see Figure 11) and 28% were taking 9 or more medicines related to their disorder. However in the majority of cases this was not a modern medicine indicated specifically for treatment of their rare disorder.

Figure 11 Number of medicines taken by people with rare disorders



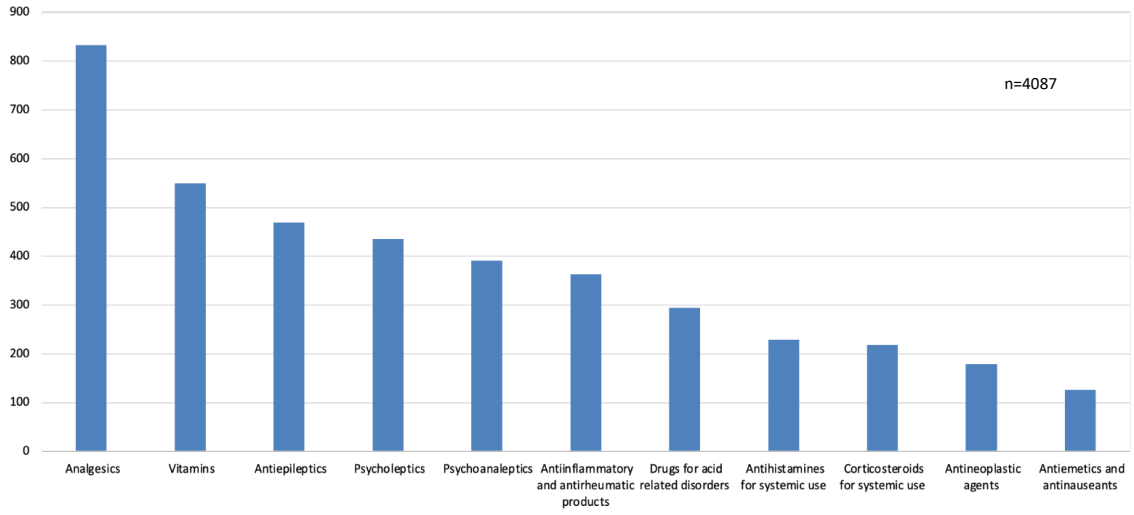
There were a total of 548 different medicines listed. On average there were 6.2 medicines being taken per person, which almost doubles previously reported. This is anticipated to be due to the increased number of medicines that were coded for in the 2025 survey, evidenced through increased utilisation of paracetamol and vitamins compared with previous surveys.

There were a number of people taking a broad range of different treatments, including analgesics, antipsychotics, anti-inflammatories, and vitamins. The most commonly reported medicines were *paracetamol* for 454 people (50%), *ibuprofen* for 255 (28%), *omeprazole* for 244 (27%), *Vitamin D* for 240 (27%), *codeine* for 172 (19%), *melatonin* for 139 (15%), *celecoxib* for 128 (14%), *gabapentin* for 128 (14%), *ondansetron* for 126 (14%), *loratidine* for 123 (14%), *tramadol* for 115 (13%), *prednisolone* for 113 (13%), *pregabalin* for 109 (12%), *cetirizine* for 106 (12%), *vitamin C* for 102 (11%), *zopiclone* for 102 (11%), *amitriptyline* for 100 (11%), *vitamin B* for 98 (11%), *iron* for 94 (10%), *morphine* for 92 people (10%).

The most frequently reported medicines were grouped according to ATC code, with the top 10 codes for these medicines presented in Figure 12. Despite the overall increase in medicines reporting, the most common medicines prescribed and publicly funded were for symptoms suffered as a consequence of having a rare disorder (e.g. pain and inflammation), rather than for direct treatment of the rare disorder itself.

No treatment is available in NZ specifically for my problem. Except immunosuppressants like prednisone. – Person with a RD

Figure 12 Number of people taking medicines, categorised by ATC code (top 10)



Over three in ten (39%) people believed they were missing out on some medicines. Of those listed, the most frequent were for medicinal cannabis/CBD, Ketamine, Monofer (iron infusion) and Daratumumab.

[name of rare disorder medicine] is used all around the world to treat my disease, but is not funded in NZ – Person with a RD

The world of [rare disorders] has moved on while NZ has stayed in the last century…. treatments are a long way from being approved by Pharmac, and newer treatments are not even on their radar. The treatment I had is now over 40 years old – Person with a RD

Only 10% had attempted to gain access to medicines through the Named Patient Pharmaceutical Assessment (NPPA), a process whereby Pharmac considers funding a treatment for an individual patient whose clinical circumstances are exceptional. Where an application had been made, for 61% it had been successful.

Very very hard to get funded medications- the specialist has had to make many applications to Pharmac because the disease is very rare (roughly 3 per million) - this is incredibly stressful as we don't know if we will be successful or not.
– Person with a RD

Almost 30% of people are self-funding their rare disorder medicines, the majority of whom are doing so out-of-pocket with a small proportion utilising private insurance (2%) or crowd funding (0.6%). It is anticipated that this could lead to inequity in medicine access.

The "correct" best medication for [the rare disorder] is not funded in NZ. I suffered through the "fail first" approach for about a year before I gave up and self funded the correct medication. It has been life changing. However the cost is wildly prohibitive and it is getting to the stage where I cannot afford to pay any more. I'll have to go back to the meds that don't work as well and have heaps of side effects. It's really not fair that these medications are not funded in NZ. With these medications, I can work a couple days a week and have some sort of normality. However, it comes at a huge cost to my family.
– Person with a RD

Of those people that had thought about it, almost all (96%) were either worried or very worried that promising future treatments and medicines (e.g. gene therapy/CRISPR) will not be funded by New Zealand's public health system.

Disability and Social Support Access

Care Services

Access to appropriate care services and support was often variable for people living with a rare disorder.

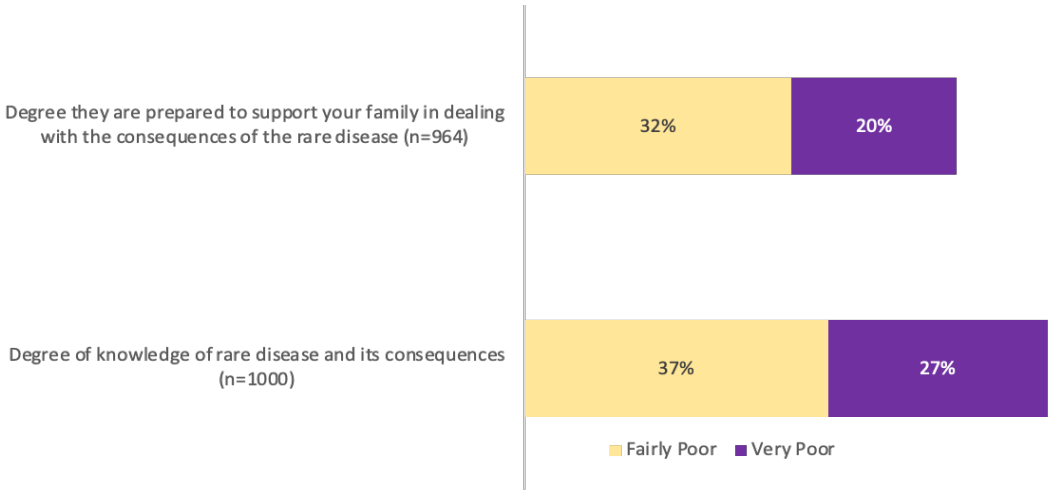
Overall, people did not feel informed at all around relevant social services that can help them (66%), financial help they could be entitled to (59%), cultural services e.g. interpreters, whānau care services (31%), nor relevant health specialists and services for the disease (50%).

A majority of people said that **professionals from social, local and support services are not well prepared to support their family**, nor informed about the disorder and its consequences (Figure 13).

Difficult to navigate funding streams. You have to find everything out about this yourself and there is so much admin to get it organised. It's exhausting. – Person with a RD

We haven't been assigned a social worker as there isn't currently one in our area. We missed out on getting the child disability allowance for months because of this, and we are probably missing out on other supports we are unaware of too. – Parent of a child with a RD

Figure 13 Level of support and knowledge from support services



We had to fight hard to get adequate carer support hours due to her being very high needs, in addition to other funding to support with paying for a carer. Her disorder had never been heard of in NZ when diagnosed, up until recently she was the only child in NZ so any additional support has been a challenge. – Parent of a child with a RD

Only 15% of people had some respite care in the last 12 months, with a range from 1 day to more than 30 days. Yet while another 8% of people qualified for respite care, they were unable to use it as it was either unavailable or did not meet their requirements.

For almost two in ten people with a rare disorder, or for their family/whānau, they reported a gap in receiving needed mental health and wellbeing support.

Finally, for almost one in two people (41%) they did not feel that their wairua ora/spiritual health had been considered in their healthcare.

Disability Support

Overall, 58% of people did not generally think that Disability Support Services took into account the unique needs of the rare disorders population.

Equity

While it is noted that a number of inequities exist in access to care for people with a rare disorder when compared with the general population, this may then be exacerbated for Māori. While the overall results were generally similar in reported measures of diagnosis, healthcare utilisation and disability and social support access, there were some differences between Māori and non-Māori reported here (Table 2).

Table 2 Key differences between Māori and non Māori

Survey response	Māori	Non-Māori	Difference
For person living with rare disorder			
Experienced difficulties in activities of daily living - Understanding and learning - Communicating with others - Personal hygiene - Taking care of finance and admin tasks - General social life and positive relations **Rare disorder impacts a lot on several aspects of their health and everyday life**	 68% 68% 74% 65% 87% 67%	 46% 51% 59% 50% 74% 56%	 +22% +15% +16% +15% +14% +11%
People (whānau/family, friends) are involved in the care and/or support in average week	81%	66%	+16%
Tensions experienced between whānau/family members since the onset of the rare disorder	70%	58%	+12%
Members of the whānau/family feel neglected since the onset of the rare disorder	62%	48%	+14%
Feel well informed about cultural services e.g. interpreters, whānau care services	37%	16%	+21%
Wairua ora/spiritual health has been considered in healthcare (where applicable)	35%	24%	+10%
Patient organisation is primary source of information about disease, treatment and care (*excluding RDNZ)*	10%	22%	-12%
Carer of person living with rare disorder			
Has taken at least 1 day of publicly funded respite care in year	27%	14%	+14%
During the past 4 weeks has sometimes felt unhappy and/or depressed?	60%	48%	+12%
During the past 4 weeks has sometimes felt they could not overcome their problems	47%	34%	+13%

Overall the majority of differences between groups related to the impact of the rare disorder for the person, their carer, and for their whānau/family. It is noted that Māori respondents were more likely to be the primary carer (+19%) rather than the person with the disorder which may explain some of these findings.

Conclusions

The 2025 Voice of Rare Disorders Survey demonstrates that people living with rare disorders in Aotearoa New Zealand continue to encounter significant barriers to timely diagnosis, coordinated care, access to medicines, and appropriate support services. The finalisation of the Rare Disorders Strategy in 2024 marked a pivotal step forward, articulating a vision where "people living with rare disorders and their whānau are enabled to live their best lives" and where "no one is left behind." To realise this vision, a coordinated, measurable, and equity-focused plan for implementation is essential, with key actions across each of the following areas:

- **Implementation plan** with funding and resource commitment, and with cross-agency leadership and oversight.
- **Leadership and coordination mechanisms** established for rare disorders.
- **Prioritise equity** for people with rare disorders compared to the general population, ensuring that efforts to close this gap lift all parts of the rare disorder community, including Māori, through culturally safe care and support.
- **Rare Disorders NZ** as a key partner in strategy implementation and ongoing advocacy to ensure the voice of rare disorders is heard.

Taken together, these actions will be key to build momentum to deliver better outcomes for people living with a rare disorder in Aotearoa.

Implementation Plan for the Rare Disorders Strategy

The absence of an implementation plan to date represents a significant gap in delivering the Rare Disorders Strategy and should be addressed as a priority. The plan should set out clear priority actions, timelines, responsibilities, and reporting arrangements across responsible agencies. Progress against the plan should be reported through existing performance and accountability mechanisms, including annual and performance reporting.

As part of implementation planning, agencies should carry out a scan of where rare disorders intersect with their current priorities and workstreams to ensure opportunities are not lost for the rare disorder community. The implementation plan needs to set out what is achievable within the next few years in aligning with existing work streams, and what needs to be built on each year to ensure achievement of the end goal in ten years.

Ongoing engagement with the rare disorders community must be embedded, ensuring the voices of people living with rare disorders and their whānau remain central to decision-making.

Key Implementation Action:
Develop and deliver a fully funded, cross-agency implementation plan with clear priorities, responsibilities, and reporting, supported by strong governance and ongoing engagement with the rare disorders community.

Leadership and Coordination Mechanisms

Consistent with the Strategy's recognition of the need for 'leadership and coordination mechanisms' for rare disorders, a Rare Disorders National Clinical Network or similar should be established and funded as

a foundational leadership and coordination mechanism. This Network should be understood as a first step towards the sustainable, appropriately resourced leadership and coordination function envisaged by the Strategy. The Network should provide a recognised and authoritative voice for rare disorders within the health system and support improvements in diagnostic pathways, referral and navigation, care standards, capability development, and system design. The leadership and coordination mechanism should also link to workforce development and planning, including building clinical capability, identifying areas of expertise, and ensuring appropriate tools, guidance, and pathways are in place when a rare disorder is suspected or diagnosed.

Key Implementation Action:

Establish and fund a Rare Disorders National Clinical Network to provide system-wide leadership, coordination, and capability-building for improving diagnosis, referral pathways, care standards, and overall outcomes for people with rare disorders.

Prioritise Equity

The 2025 Voice of Rare Disorders Survey reconfirms that people living with rare disorders suffer from profound inequities compared to those who do not live with a rare disorder across multiple dimensions of their health and wellbeing. These inequities manifest in delayed and inaccurate diagnosis, fragmented care pathways, limited access to essential medicines, and significant barriers to social and disability support, as well as education and employment. These challenges may be greater for Māori, who report stronger impacts on themselves, their carers, and their whānau, compounded by limited culturally safe services and inadequate recognition of their needs.

Achieving equity requires sustained, intentional action throughout implementation of the Rare Disorders Strategy. All initiatives should be co-designed with people living with a rare disorder including with Māori, align with Te Tiriti o Waitangi, embed cultural safety, remove barriers to diagnosis and support, and track progress through strong data and public reporting. People living with a rare disorder including underserved groups must be genuinely represented in governance and decision-making so their voices shape every stage of implementation. Equity is essential to ensuring that "no one is left behind."

Key Implementation Action:
Develop and implement a co-designed Equity Action Plan that identifies and removes barriers to diagnosis, care, and support, sets measurable equity targets, and ensures accountability through regular audits and public reporting.

Rare Disorders NZ as a Key Partner

Rare Disorders NZ provides the most representative and collective voice for people with rare disorders in New Zealand. Their leadership and expertise are vital to ensuring that the Strategy delivers real, measurable improvements for the community. It is recommended that Rare Disorders NZ be resourced to lead community engagement, advocacy, and monitoring, and that their role be formalised in all governance and implementation structures. Ongoing funding and support will enable Rare Disorders NZ to facilitate research, education, and peer support, and to hold the system to account for progress.

Key Implementation Action:
Formalise and resource Rare Disorders NZ as a central partner across all governance and implementation structures, enabling them to lead community engagement, advocacy, research, and monitoring to ensure the collective voice of people with rare disorders informs all decision-making.

Where rare disorders are being overlooked - addressing the gaps

In 2019, Rare Disorders NZ collective identified seven strategic priorities to improve health and wellbeing for people living with a rare disorder. These priority areas were reaffirmed following the release of the Rare Disorders Strategy in 2024. While it is hoped that the implementation plan will go a long way towards addressing diagnosis, planned pathways for clinical care and workforce development priority areas, the following are at risk of continuing to be overlooked without a targeted plan. This is despite being essential to improving the health and wellbeing of people living with rare disorders:

1. Rare disorder data collection
2. Equitable access to modern rare disorder medicines through a specific assessment pathway
3. Access to disability and social supports, including support for carers

Rare disorder data collection

You can't improve what you don't measure. Currently New Zealand has no nationwide process for collecting data on rare disorders. As a result, people living with rare disorders are currently invisible in health system data. Health entities must be required to capture relevant data on rare disorders in New Zealand. Due to the complexity of care for many rare disorders, the data needs to be accessible beyond primary care. This requires that the right frameworks are in place for the transfer of patient records from the primary care level to the soon-to-be rolled out Shared Digital Health Record. This would then enable health professionals to access their patients' medical history, including medications, allergies, previous diagnostic assessments etc. when permitted and required, to recommend the best and safest health care plan.

Recommendation:
All Patient Management Systems adopted by primary care providers should be required to integrate coding systems with a significant number of rare disorder codes, such as SNOMED – CT, to begin routinely collecting rare disorder data. Furthermore, a regulatory framework for Patient Management Systems to transfer patient data to the Shared Digital Health Record should be established in conjunction with its rollout.

Equitable access to modern rare disorder medicines through a specific assessment pathway

While there have been great strides in the last 40 years in rare disorder medicine development, there remain disparities in access to rare disorder treatments in New Zealand compared to different countries around the world. This is highlighted by variable availability of 204 designated essential rare disorder medicines[14] as well as publicly funded access levels compared with European countries.

[14] https://ojrd.biomedcentral.com/articles/10.1186/s13023-021-01923-0

This is echoed in this survey which reveals that nearly half of respondents believe they are missing out on needed medicines, and the vast majority are concerned about future access. The current funding model is not fit for purpose, and the Named Patient Pharmaceutical Assessment (NPPA) process is inconsistently applied. To address these challenges, a dedicated, transparent assessment and funding pathway for rare disorder medicines should be developed, drawing on successful models from overseas, in particular the UK, Australia, and Canada.

This pathway must be responsive to the unique challenges of rare disorders, including small patient populations and high treatment costs, and should be designed and reviewed in partnership with Rare Disorders NZ and clinical experts. The goal is to ensure that all New Zealanders with rare disorders have timely and equitable access to effective therapies.

Recommendation:
Design and implement a dedicated, transparent assessment and funding pathway for rare disorder medicines, informed by international best practice and developed in partnership with Rare Disorders NZ and clinical experts. This pathway must ensure timely and equitable access to effective therapies for all New Zealanders with rare disorders.

Access to disability and social supports, including support for carers

The rare disorder population often falls through the gaps in our support systems. The survey results highlight systemic barriers to accessing disability and social support for many with rare disorders, with the majority of respondents feeling that Disability Support Services do not take into account the unique needs of the rare disorder population. Often those affected by rare disorders find they do not meet criteria to access disability support services and funding despite having the equivalent need to those that do.

People with undiagnosed and rare disorders who have disabilities are often not recognised nor receive disability support services, because the criteria are not designed with this population in mind. Carers are also missing out on support, with only 15% of respondents receiving respite care in the last 12 months.

Recommendation:
Develop pathways for people living with rare disorders to access information and support services available to them, ensuring those undiagnosed but with unusual and debilitating co-presenting symptoms are included. Recognise the important role carers play in the wellbeing of those affected by rare disorders, and improve access to respite care.

Final Remarks

The Rare Disorders Strategy provides a clear framework and vision for change. It is essential to translate this vision into actionable, time-bound priorities that will drive measurable improvements for people living with rare disorders and their whānau. Full implementation, with a relentless focus on equity, partnership, and accountability, is essential to ensure that “no one is left behind” and that Aotearoa New Zealand becomes a world leader in rare disorder care.

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