IMPACT OF LIVING WITH A RARE DISORDER IN AOTEAROA NEW ZEALAND

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

2024

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This white paper was written by HealthiNZ

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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 a parent of a child who has cystic fibrosis and currently sits on the Board of their patient organisation Cystic Fibrosis NZ.

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Executive summary

For New Zealanders living with a rare disorder the impact continues to be significant, affecting not only themselves but extending to families/whānau, the health system and society. Previous survey white papers have called for a Rare Disorders Strategy for New Zealand, to ensure better health outcomes for people with rare disorders. Following advocacy and the Pharmac review, a strategy led by Manatū Hauora |the Ministry of Health is now nearing completion.

Rare Disorders NZ have communicated that while they welcome the Rare Disorders Strategy they will be continuing to advocate for essential implementable actions to support people with rare disorders as a strategy alone will not lead to meaningful change for the rare disorder community.

Key findings from the 2023 survey:

The 2023 New Zealand Voice of Rare Disorders Survey provides another opportunity to understand the real impact of living with, or caring for, people with rare disorders. At 1076 responses, it is the largest-ever survey of consumer reported outcomes for rare disorders in this country. In this 2023 version, with 525 different diagnoses and better participation from Māori, it provides the most representative and collective voice for people living with a rare disorder in Aotearoa.

For people living with a rare disorder today and their carers, the picture remains a difficult one. The majority reported that the rare disorder impacts a lot on their health and everyday life, is disabling and makes a number of everyday activities difficult. Approximately half of people are self-funding costs of their healthcare (e.g. healthcare professionals, treatments and medications and special diets) to some extent with the majority finding this hard to manage, or unable to afford it. 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.

It is sobering to see that, despite some achievements since the 2021 survey, including the progress of the Rare Disorders Strategy, this has not translated into better outcomes. Change is needed.

Close healthcare service and support gaps in New Zealand's health and other systems

The survey results paint a continuing picture of isolation, lack of timely and accurate diagnosis, high utilisation of healthcare services, poor treatment access, lack of coordinated care, significant carer impact, lack of access to care services including suitable mental health and wellbeing support, and for many, being lost in the system.

To address these gaps there is an opportunity to create a Rare and Undiagnosed Disorders Centre of Expertise. This Centre would be a multidisciplinary team of internationally-networked experts within New Zealand. It will identify gaps in the delivery of service and support in New Zealand's health and other systems for people living with rare disorders, similar to other global initiatives. It will include expertise in areas of testing and diagnosis, evidence-based best practice and family/whānau support.

Need for better access to rare disorder medicines

The current one-size-fits-all model under Pharmac's pharmaceutical schedule does not work for low volume, high-cost medicines for rare disorders. The survey results confirmed this, in that there were few accessible effective modern medicines available for the majority of people, and only a few had successfully gained access to medicines through the Named Patient Pharmaceutical Assessment (NPPA) process. Importantly, most were worried that promising treatments and medicines (e.g. gene therapy/ CRISPR) will not be funded by New Zealand's public health system in the future.

Given this picture, an imperative exists for a Single barrier-free pathway to rare disorder medicines, one which takes into account approaches from other countries such as Australia and the UK. An alternative pathway will be key to addressing the lack of modern medicine access that continues to fail the rare disorder community in this country.

Ensure rare disorder coding is incorporated in health system data sets

The classification system for diseases that New Zealand uses does not include most rare disorders and this contributes to challenges in accessing the necessary treatments and services, communication between healthcare professionals, as well as for research. In turn this lack of data is problematic for proper funding and resource allocation of healthcare services.

There is an opportunity to Incorporate coding of rare disorders in the classification system of diseases, for up to 6,500 rare disorders. This should be incorporated into electronic health records (EHR) and other administrative data sets.

Implement the Rare Disorders Strategy

New Zealand's first Rare Disorders Strategy is due to be completed in 2024. It acknowledges that people and whānau with rare disorders can experience inequitable health outcomes and these outcomes are often influenced by unfair barriers, such as lack of timely access to services and difficulties in navigating the health system.

Rare Disorders NZ, as the only national organisation supporting all New Zealanders living with a rare disorder and their carers, has already played an integral role in the Strategy's development. In addition, through this survey it continues to provide the most representative and collective voice for people with rare disorders. Given this there is a key opportunity for RDNZ to be valued as a key partner with Te Whatu Ora and other government agencies in the Strategy's implementation.

Full implementation of these essential actions will be important changes to improve outcomes for people living with a rare disorder in Aotearoa.

Significant challenges remain for people living with rare disorders

Rare disorders have widespread impacts on 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.

> **[There's] nowhere to go and I feel unheard and forgotten because I'm in the too hard basket.**
>
> **Person with rare disorder**

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[2].

> **My most difficult task was trying to get care and medical staff to understand this rare disease, it has nearly broken me.**
>
> **Grandparent of a child with rare disorder**

New Zealand Rare Disorders Strategy

New Zealand has long needed a Rare Disorders Strategy to ensure better health outcomes for people with rare disorders. It would prioritise actions for improved health and wellbeing, by creating a roadmap and pathways that ensure people living with rare disorders are not left behind. It was recommended both in the RDNZ 2021 Voice of Rare Disorders report and by the 2022 Pharmac Review report.

The Rare Disorders Strategy has now been developed by Manatū Hauora |the Ministry of Health and at the time of writing is awaiting the Minister of Health's formal approval[2]. This will be the first national strategy for Pae Ora | health and wellbeing for people and whānau living with rare disorders in Aotearoa New Zealand.

The strategy will provide a framework to guide health entities in improving health outcomes for people with rare disorders and their families or whānau. It will uphold the principles of Te Tiriti o Waitangi. It will also allow the health sector to provide better support for people with rare disorders, as well as making it easier for people, practitioners, and organisations to get the information and support that they require.

Despite this welcome development, Rare Disorders NZ (RDNZ) believes the Rare Disorders Strategy will need to include a set of actionable items to drive needed changes (Table 1).

1 ANSEA report, 2019.

2 https://www.health.govt.nz/our-work/diseases-and-conditions/rare-disorders-strategy

Table 1: Essential priority actions needed to drive change in rare disorders

Challenge	RDNZ Priority Actions
Gaps in healthcare service and support systems that need to be closed	**#1** Rare and Undiagnosed Disorders Centre of Expertise
Poor access to rare disorder medicines	**#2** Single barrier-free pathway to rare disorder medicines
Coding for rare disorders is not routinely incorporated in health systems data sets	**#3** Incorporating coding of rare disorders in the classification system of diseases
Effectively implementing strategy for rare disorders once completed	**#4** Rare Disorders NZ is a key valued enabler for the Strategy's implementation

Significantly, this provides a framework for understanding key results and recommendations from this report.

2023 Voice of Rare Disorders Survey

The Voice of Rare Disorders Survey was first developed in 2019 to better understand the impact for people living with a rare disorder in New Zealand and has since been repeated, both in 2021 and 2023[3]. 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, Employment, Care Services, Whānau/Family and social life, stress and wellbeing. Eligible participants were people with a rare disorder in New Zealand or were 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[4] which to date has over 10,000 survey responses and BURQOL-RD[5]. Following from the experience in previous surveys, some additional questions were added. Also, where appropriate, comparisons with the population from the 2019 and 2021 surveys will be made and reported on in the results section. Overall, this survey will further expand the evidence-base of people living with a rare disorder in New Zealand.

Rare Disorders NZ currently engages with 159 support groups representing at least 33,000 people living with different rare disorders. All groups were encouraged to send personalised emails to their members, stating the project objectives and providing a link to a web-based questionnaire. The data collection period ran from 18th September to 2nd December 2023.

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

3 2023 NZ Voice of Rare Disorders Survey.

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

5 BURQOL RD Project: We appreciate the support of the researchers of the "Social/Economic Burden and Health-Related Quality of Life in Patients with Rare Diseases in Europe" Project (BURQOL-RD), financed by the European Commission within the framework of the Health Programme (grant A101205).

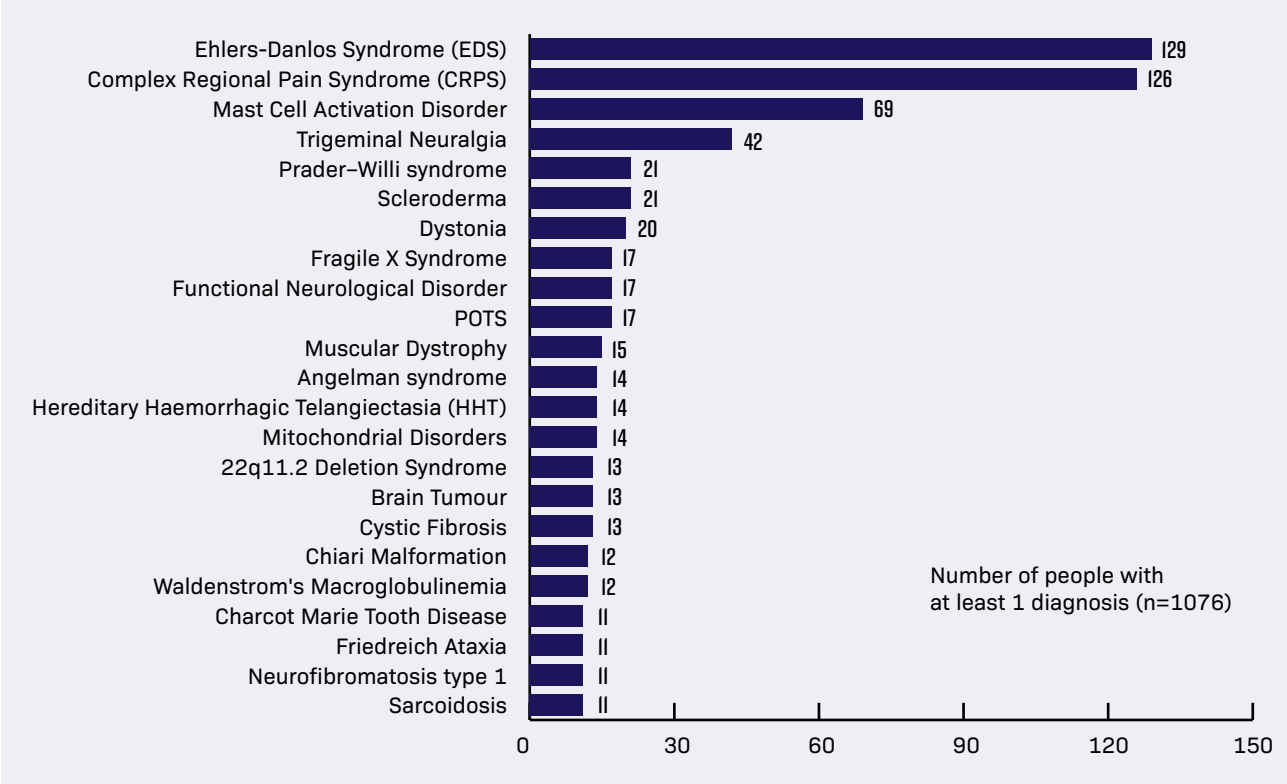
Results

Rare Disorder Diagnosis

Overall there were 1076 responses with a confirmed diagnosis included in the primary analysis[6]. This represents **the largest ever survey of consumer reported outcomes for people with rare disorders in NZ**, surpassing the previous 2021 survey with an overall 50% increase in response.

The primary analysis reported on the overall population of people with a confirmed rare disorder diagnosis. There were 525 different diagnoses; over twice as many as listed in the previous survey, with a number of rare disorders having more than 10 patients diagnosed per disorder in New Zealand, including Ehlers-Danlos Syndrome (EDS), Complex Regional Pain Syndrome (CRPS), Mast Cell Activation Disorder, Trigeminal Neuralgia, Prader–Willi syndrome, Scleroderma, Dystonia, Fragile X Syndrome, Functional Neurological Disorder, POTS, Muscular Dystrophy, Angelman syndrome, Hereditary Haemorrhagic Telangiectasia (HHT), Mitochondrial Disorders, 22q11.2 Deletion Syndrome, Brain Tumour, Cystic Fibrosis, Chiari Malformation, Waldenstrom's Macroglobulinemia, Charcot Marie Tooth Disease, Friedreich Ataxia, Neurofibromatosis type 1 and Sarcoidosis.

Figure 1: Most common rare disease diagnoses in 2023 Voice of Rare Disorders Survey



Overall, 29% of people reported more than one diagnosis, with 8% reporting 3 or more diagnoses. The inclusion of a number of new, as well as a wider range of, diagnoses in this survey reflects expanding engagement reaching these communities from Rare Disorders NZ.

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 13.3%, much closer to National Census estimates[7] than previous 2019 and 2021 surveys. This reflects improved engagement with this group that needs to be continued.

Responses from Māori are presented in a companion report to this white paper.

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.

6 17 people excluded from primary analysis because they were listed as undiagnosed.

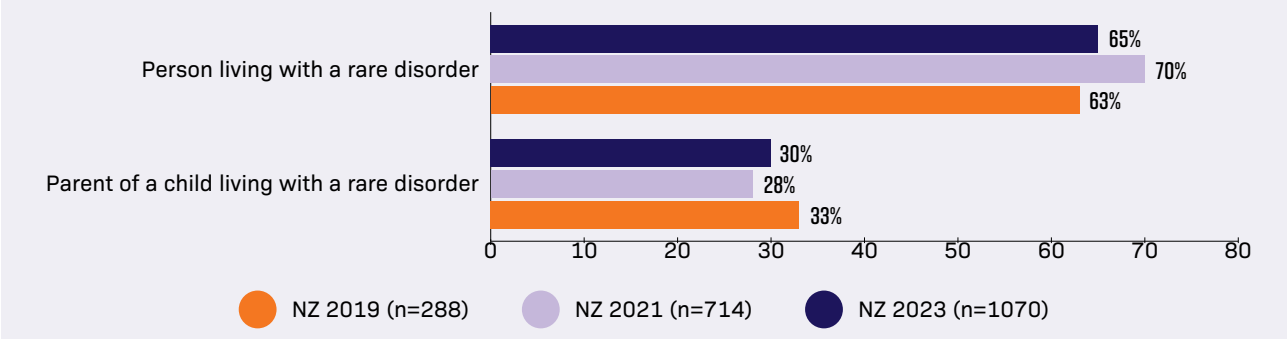
7 https://www.stats.govt.nz/information-releases/maori-population-estimates-at-30-june-2023/#:~:text=At%2030%20June%202023%3A,453%2C900%20females%20identifying%20as%20Māori

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 previous surveys.

The majority of responses (65%) were provided by people with a rare disorder, which was lower than 2021 (70%) (Figure 2). Of these, **one in eight had an additional role, either as a parent, sibling or spouse of another person living with a rare disorder**.

Overall, 30% of the New Zealand responses were from parents of a person with a rare disorder.

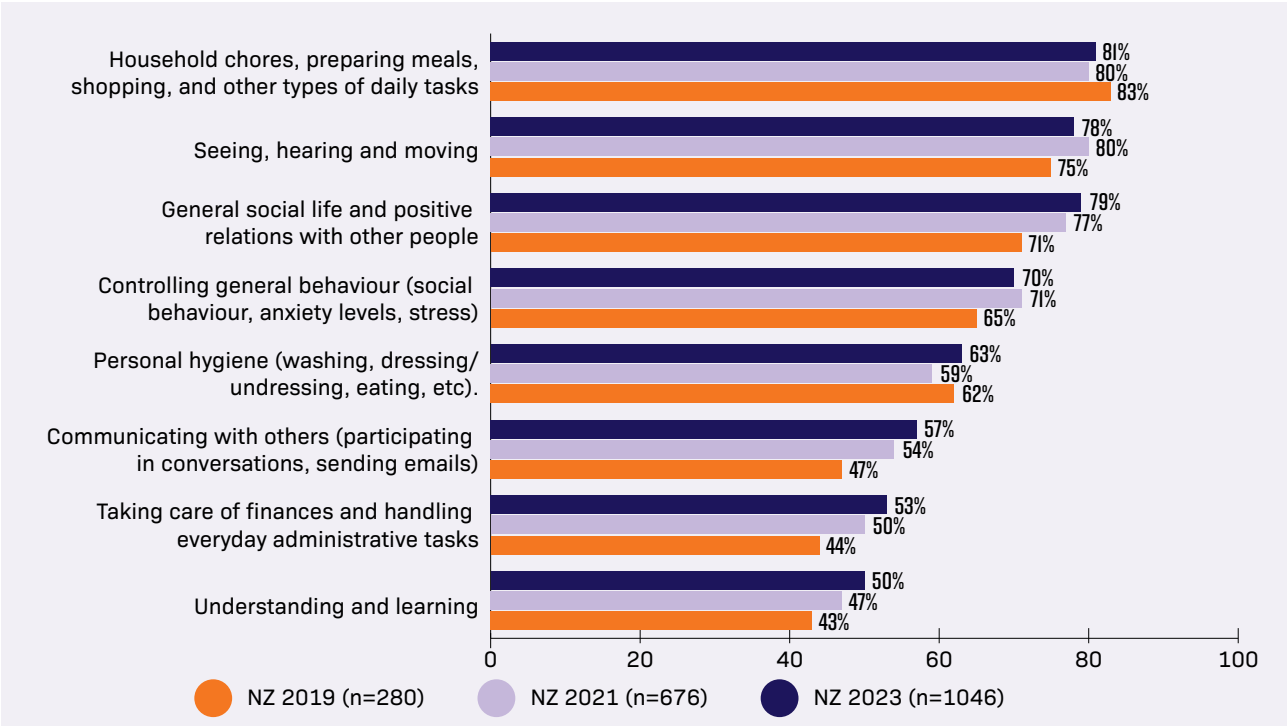
Figure 2: Relationship to person with rare disorder



Daily impact of living with rare disorder

The extent to which the person with the rare disorder could perform certain activities varied with the activity. For **50-81% of people and their families surveyed, the rare disorder makes a number of everyday activities difficult** (communicating, controlling behaviour, social life) and was similar or higher than previous surveys.

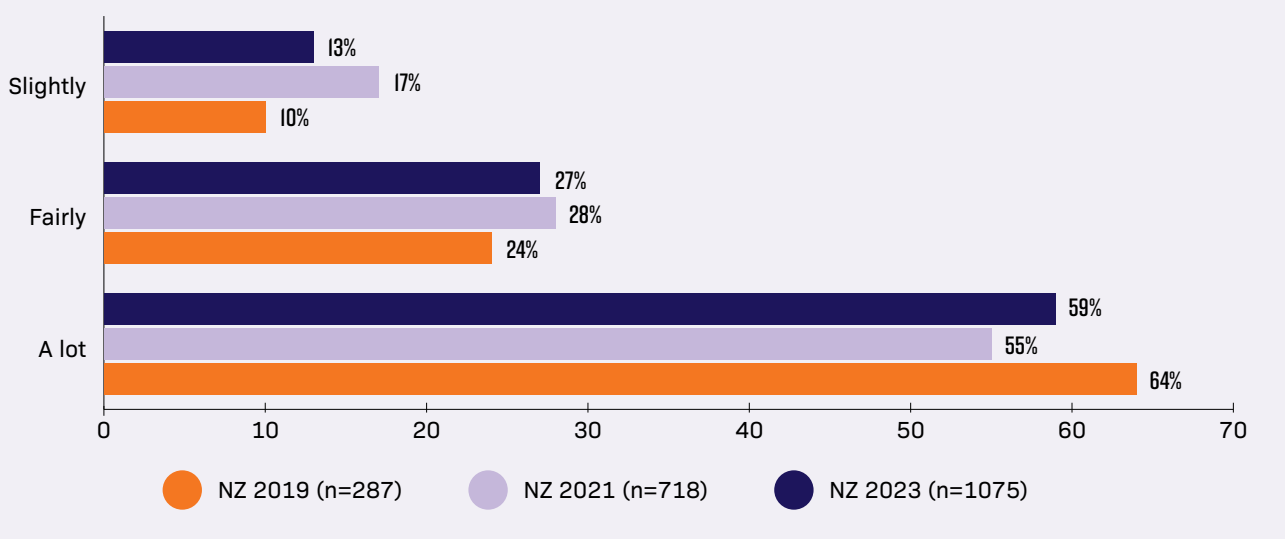
Figure 3: Activities of daily living that provide greatest difficulty



The most frequently reported household carer was the person living with the rare disorder > mother> the spouse, in that order. For the 69% that needed it, an average of 2.4 whānau/family members and/or friends were involved in their care and support during an average week.

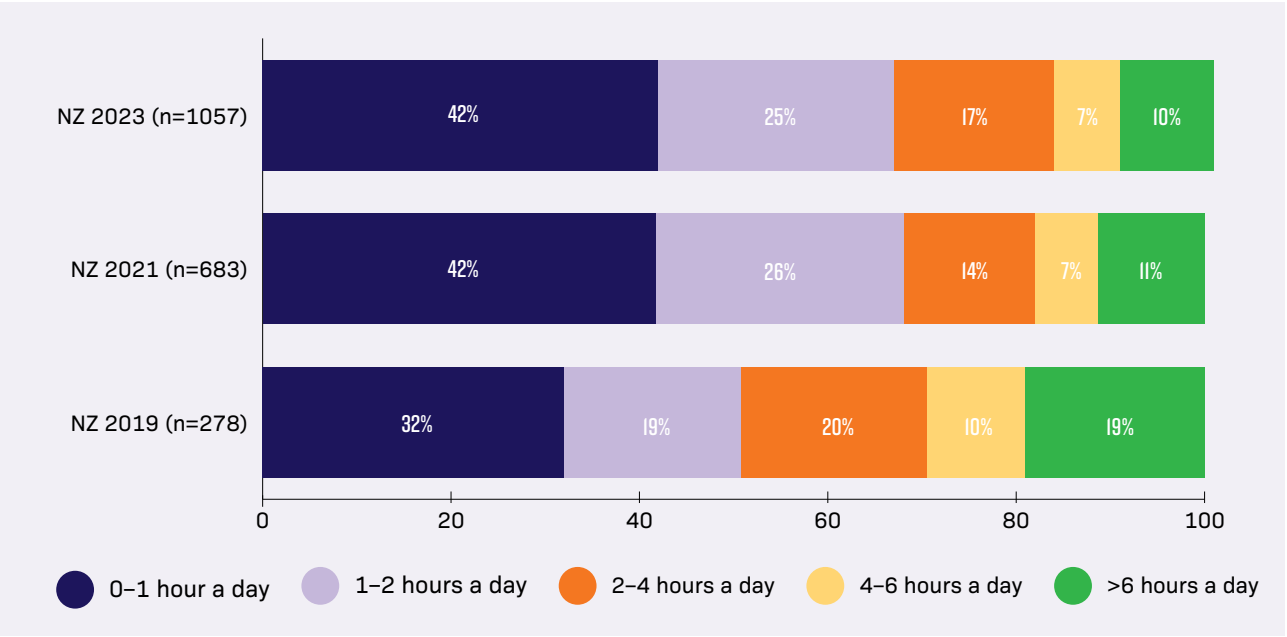
Overall, 59% of people reported that the disorder impacted a lot on their health and everyday life.

Figure 4: Impact on health and everyday life



34% required more than 2 hours per day for illness-related daily tasks including hygiene, helping with house chores, moving the person and administration of treatments.

Figure 5: Daily time investment for illness-related tasks

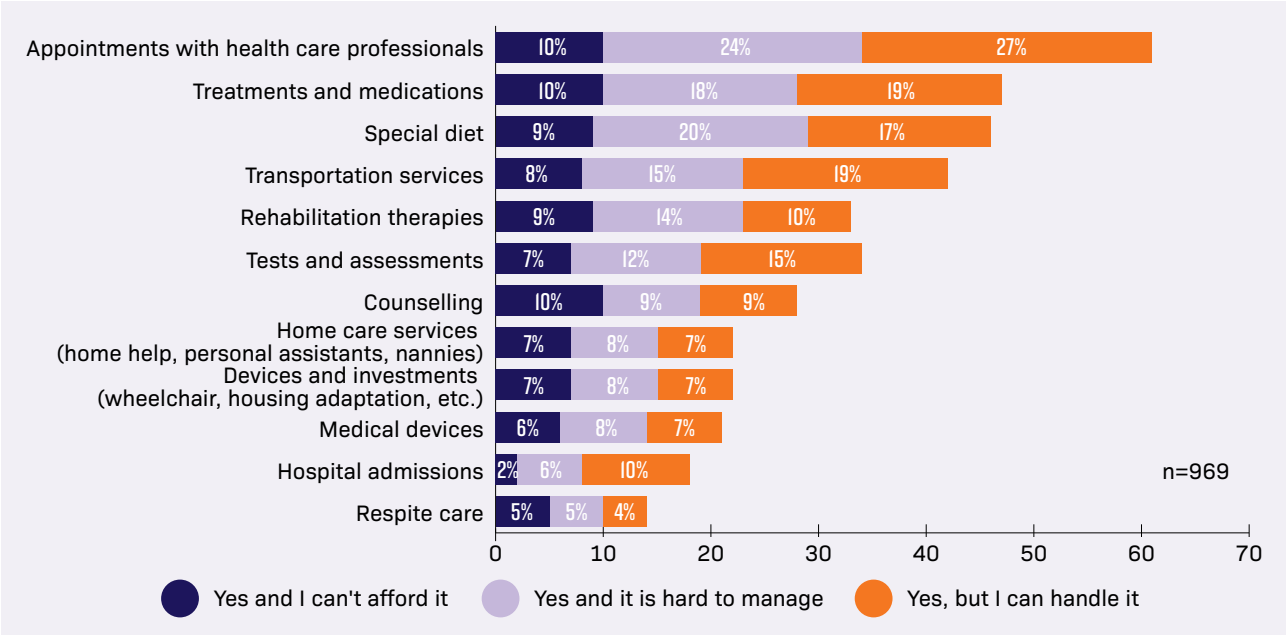


On average 2 hours per day were invested in illness related tasks.

Cost of living with a rare disorder

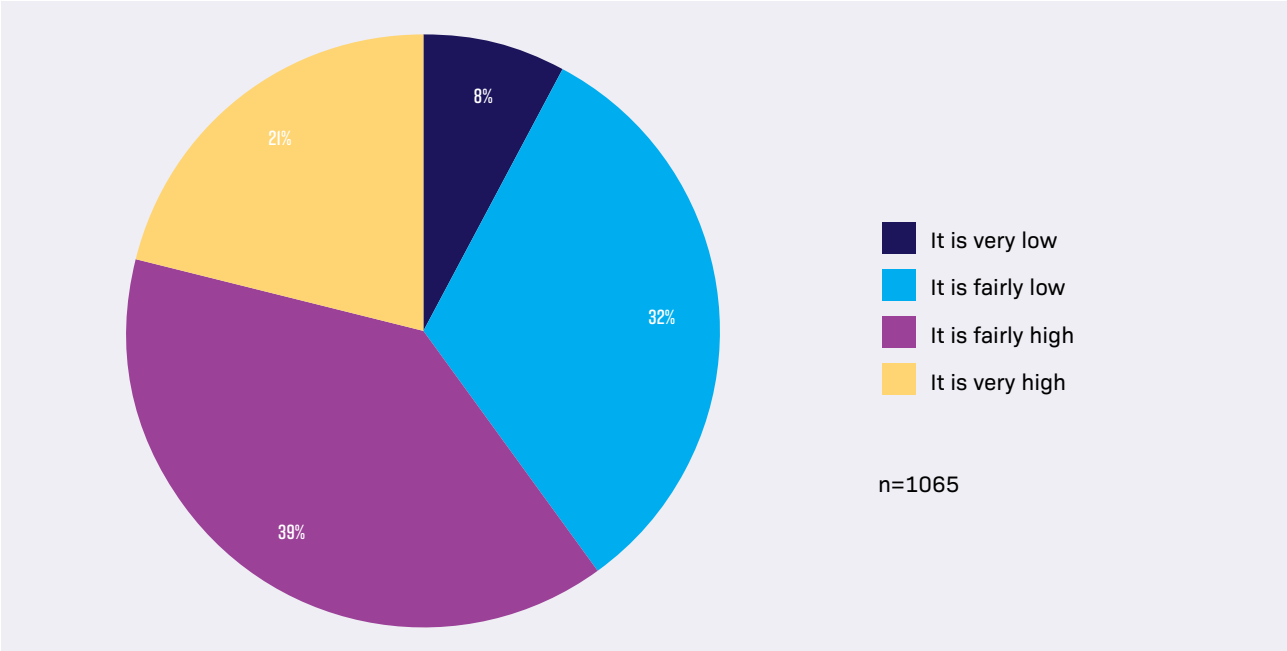
There were many costs covered by people living with the rare disorder or their family with approximately half self-funding to some extent.

Figure 6: Level of self-funding for healthcare costs



Of those that self-funded, the majority found these were hard to manage, or couldn't afford it.

Figure 7: Perception of costs associated with the rare disorder



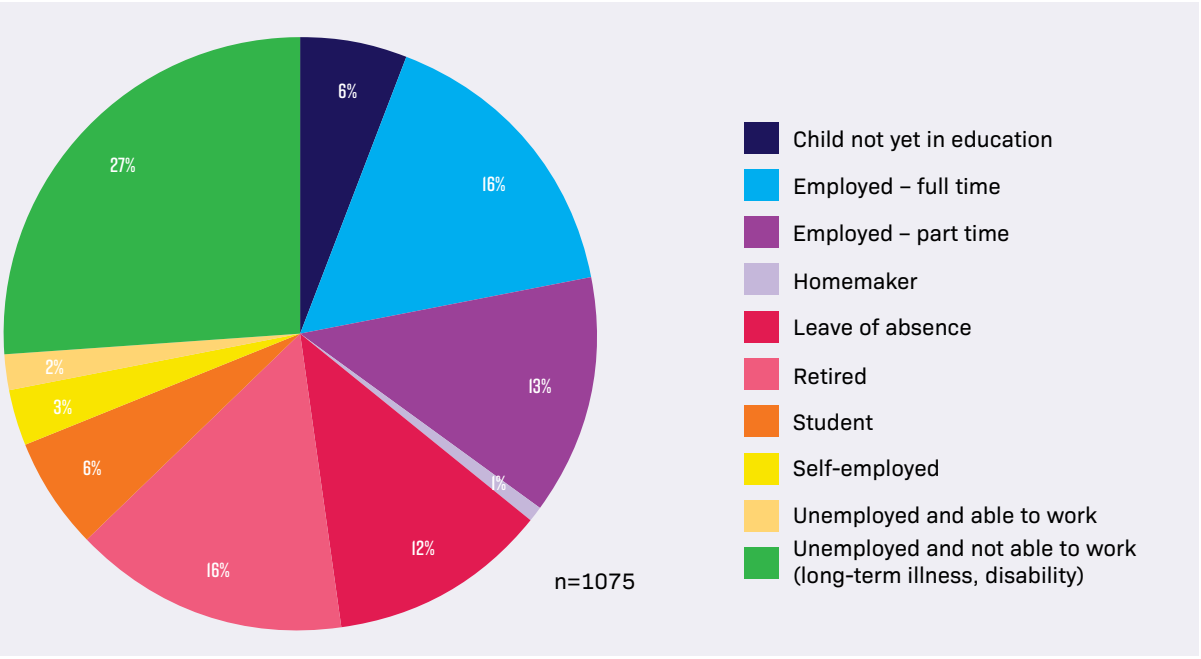
The majority (60%) of people felt that the costs associated with the rare disorder were high (Figure 7).

In addition, 54% felt the costs associated with managing their rare disorder were hard to manage.

> **Disability allowance etc doesn't anywhere near cover expenses. This results on not having the recommended treatments & living conditions required to stay well, speeding up disease progression.**
>
> **Person with a rare disorder**

Employment

Figure 8: Employment status of person with rare disorder



> **I can only work part time due to my disability. All of my wage goes into managing my disability. I want to retrain but to do that, I have to give up work. If I give up work, I can't manage my disability.**
>
> **Person with a rare disorder**

In total, 35% people with a rare disorder were in some form of employment, either being full-time, part-time or self-employed. However, 29% were unemployed, with the majority citing long-term illness or disability.

At 45%, the proportion of employed people in part-time employment was higher than the OECD average (ref), indicating underemployment. This was similar to previous surveys.

A number had to leave previous jobs due to their rare disorder. For those, 19% were declared unable to work and 12% had to retire early due to their illness or disability. A further 11% resigned because their previous job was not adapted for their condition. In addition to this, a majority would like adapted accessible work environments that took account of their rare disorder to allow for their working hours and responsibilities to remain similar or increase.

There was a slightly changed picture of employment for the main carers of the person with a rare disorder. Of these, 63% were either employed part time, full time, or self-employed. However, in many cases the carer needed to leave their previous job with 10% working in paid work part-time or on reduced hours given their responsibilities and a further 25% of carers needed to look after the person with a rare disorder full-time.

Overall, there were many ways the person's employment was affected by the rare disorder that included limiting professional choices, reducing or stopping professional activity and limiting job opportunities. It caused a decrease in income for 74% of people.

> **After my daughter's birth…and subsequent immediate long-term hospitalisation, I needed to resign from my job as it was uncertain whether my child would need full-time care forever. I have since been able to pick up freelance part-time and flexible work which I do in the hours when my child is asleep. Her health is too fragile for me to be able to go back to a full-time office-based job as I need the flexibility in case she is sick and can't go to school, or needs to go to hospital without notice. I am very tired from working odd hours and my finances are severely affected.**
>
> **Parent of a child with a rare disorder**

Family/whānau and social life, stress and wellbeing

> **Our family has been brought to our knees and our two other children are on anti anxiety medication because of a lifetime of stress and trauma because of their sisters condition and lack of support for us as a family.**
>
> **Parent of a child with a rare disorder**

Since the symptoms started, the majority of people have experienced increased tension between family members (57%) and isolation from family/whānau and friends (69%), amplified, or caused by, their rare disorder.

> **I could not get home support even when I was bed bound and it tore my family apart.**
>
> **Person with a rare disorder**

Concerningly, one in three people often felt unhappy and depressed and felt they could not overcome their problems (Figure 9, Figure 10). These issues were much higher than in general populations (based on International Social Survey Programme, 2011), especially given an additional one in three people reported sometimes having these feelings.

Figure 9: Extent person felt unhappy and/or depressed in last month: Comparison over time and with general population

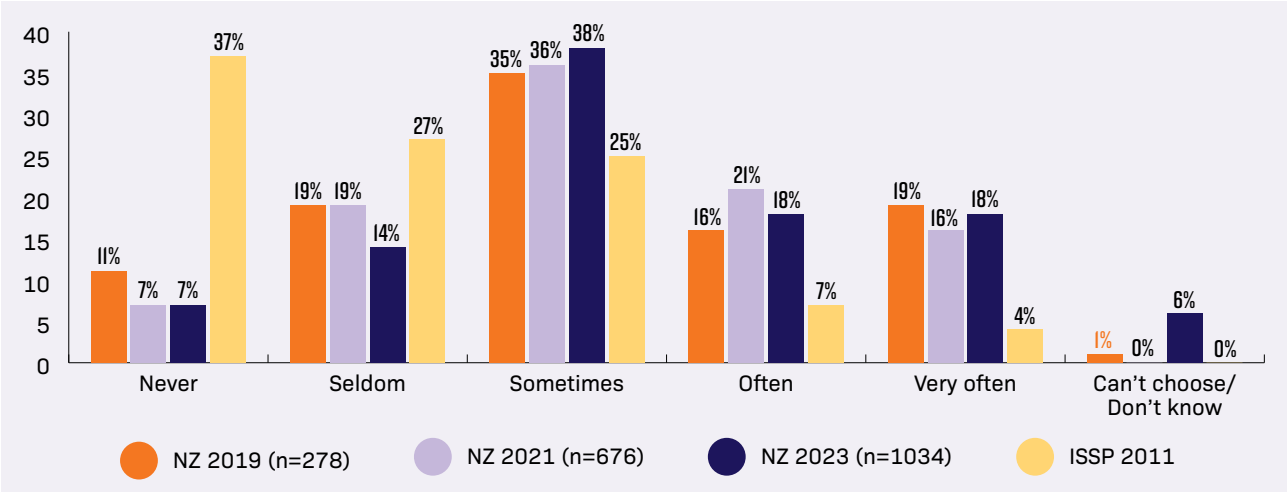
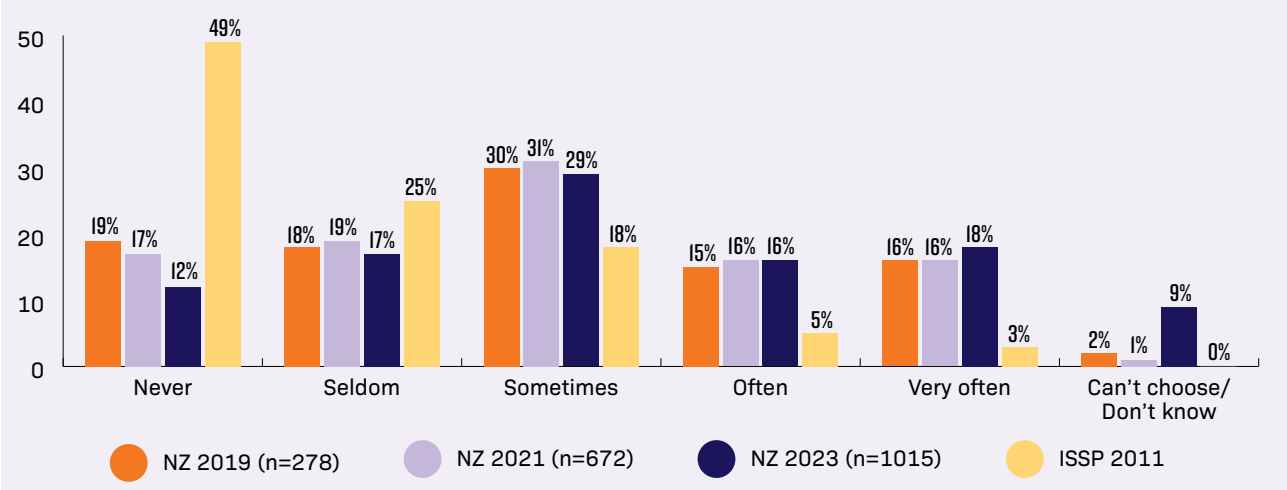


Figure 10: Extent person felt they could not overcome their problems in last month: Comparison over time and with general population



> **This year I have felt very deflated with the whole process and trying to get the help he needs.**
>
> **Parent of a child with a rare disorder**

The impact on mental health and wellbeing extended to carers with 27-30% carers often feeling unhappy and depressed and that they could not overcome their problems.

The importance of connection with others with the same condition should not be underestimated. However, while 63% of people or their carers thought that this was important, only 37% 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.

> **Having a rare condition is a lonely experience, impacts every aspect of life, and without the right kind of support, leads to depression and hopelessness as far as I can tell.**
>
> **Person with a rare disorder**

Healthcare services

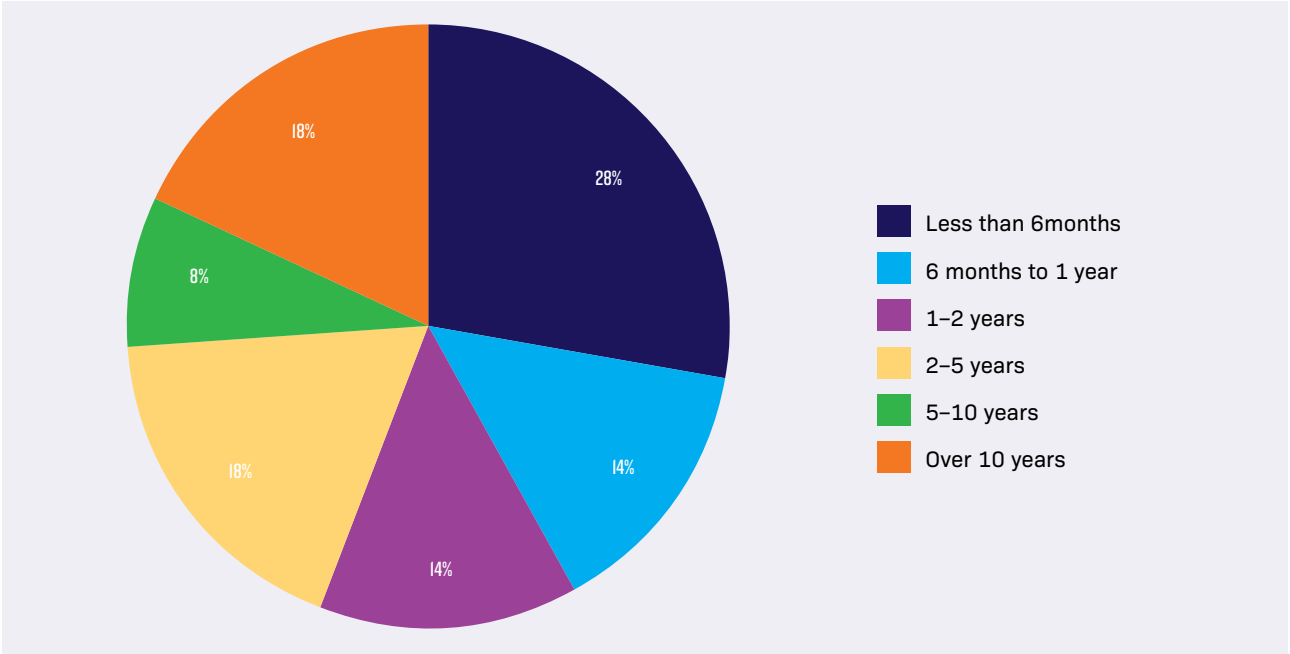
Diagnosis

> **Delayed diagnosis has been the main barrier. We were not eligible for any support in the early years and we really needed it. No diagnosis also meant issues were not treated as promptly as they might have been, resulting in more trauma for our son and for us.**
>
> **Parent of a child with a rare disorder**

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

The majority of people had challenges in getting an accurate diagnosis for their rare disorder, similar to previous surveys. Over half took longer than 1 year to get a diagnosis and for almost one in five the time taken to get a diagnosis was over 10 years (Figure 11).

Figure 11: Time taken to get diagnosis



The majority (69%), had to visit 3 or more doctors to get a diagnosis and for one in ten, over 10 doctors were visited.

61% of people with a rare disorder were misdiagnosed at least once before the final diagnosis was confirmed. Three in ten were misdiagnosed at least twice.

Almost one in two people had had access to genetic testing to confirm or investigate their diagnosis, with the overwhelming majority being through the public health system. For the remainder that needed it, a small number (5%) either had their referral declined or were still on a waiting list for genetic services.

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 2). Utilisation of healthcare services was similar or greater in 2023 than in earlier surveys.

Table 2: Healthcare service utilisation for people with rare disorders

Service	Utilisation rate (%)	Total average annual frequency	Cost per test/ visit/day[8]	Average cost per patient per year
GP visits	91%	5.8 visits	$80	$466
Specialist visits	85%	5.5 visits	$250	$1,379
Diagnostic tests	83%	5.0 tests		
ED visits	42%	1.8 visits	$370	$661
Admissions	41%	1.0 admissions		
Inpatient days	39%	11.6 days	$1,200	$13,920
ICU Inpatient days	8%	4.6 days	$5,500	$25,168

Most people had seen a specialist or GP over the last 180 days (85-91% utilisation rate) with almost 6 visits to 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).[9] 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 12 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 4.6 days in ICU. This is a significant finding and highlights the continuing impact and pressure on ICUs and other inpatient services, especially as admission rates are almost twice as high as in the general population.[10,11]

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 specialist declined** (e.g. general medicine, endocrinology, rheumatology, cardiology).

In addition, while the majority of people felt culturally safe when visiting health services, 9% considered visits to a nurse, doctor, health service, hospital service or genetic testing service to be culturally unsafe.

8 Using the Cost Resource Manual (PHARMAC): cost per specialist visits has been applied at $250 (based on physician outpatient costs, subsequent visit); cost per GP Practice visit applied at $80; cost per nurse visit applied at $40; cost per emergency department visit applied at $370; cost per day for a hospital medical ward at $1,200 (not including procedures); cost per day for intensive care unit (ICU) at $5,500.

9 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

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

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

Coordination of Care

Coordinating care is challenging for people with rare disorders, and this can be further complicated by healthcare teams.

> **We receive a range of different services and supports but it's all fragmented and siloed. Occupational Therapist, Physiotherapist, Speech Language Therapist, GP, LifeLinks – but no one that can help us navigate this all successfully.**
>
> **Person with rare disorder**

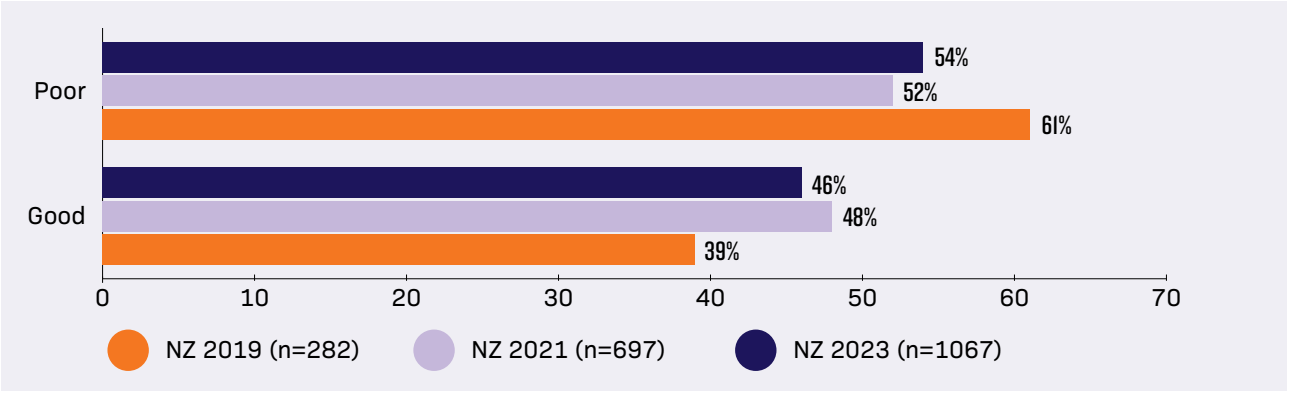
Around one in two people felt that organising care was difficult to manage. This included finding information on your disorder, your rights and administrative procedures (57%) and finding the right professionals (48%).

Overwhelmingly the internet was the main first source of information about the disorder, treatment and care, which was a somewhat surprising increase from 2021. This was then followed by various healthcare professionals as well as the 'Support Group'

Over 50% felt that communication and information exchange between different service providers was poor (Figure 12). This was sometimes linked to co-ordination or ability of healthcare professionals to access data, leading to frustration.

> **They often give you different information or perspectives that they then don't share with colleagues so when you repeat then the colleagues say "no that's not what we have here".**
>
> **Person with rare disorder**

Figure 12: Quality of communication between service providers

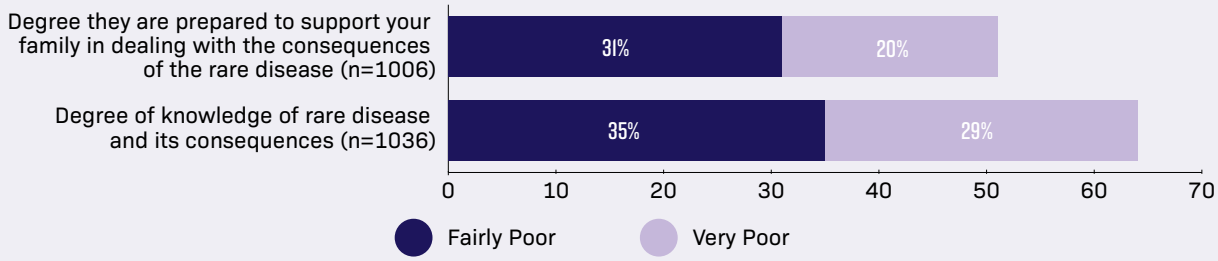


> **Even though we had a diagnosis at birth, nobody told us that a child disability allowance was available until he was 2 years old.**
>
> **Parent of a child with a rare disorder**

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).

> **We have also stumbled on support and additional funding by accident and no one person/body ever sits you down and discusses what you are eligible for which is very frustrating so we have probably missed out on some stuff.**
>
> **Parent of a child with a rare disorder**

Figure 13: Level of support and knowledge from support services



Overall, people did not feel informed at all around the rights related to the consequences of their rare disorder (79%), relevant social services that can help them (73%), financial help they could be entitled to (66%), cultural services e.g. interpreters, whānau care services (64%), nor relevant health specialists and services for the disease (54%).

A Needs Assessment will help people with a rare disorder gain an understanding of the support they need, and to learn more about goals and what help truly looks like. In this case one in two people had either had not heard of a Needs Assessment or did not want one. And even for those people that had undergone a Needs Assessment and qualified for funded services and supports, almost half believed they needed more than they were eligible for.

> **No one can give me any information on what services I qualify for or other treatment options I have available.**
>
> **Person with rare disorder**

Care Services

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

The number of hours per week received for various government-funded supports varied greatly among the 10-20% of people that qualified for it. There remains a significant unmet need for support for house chores and tasks (27%) and rehabilitation services and therapies (33%).

Where applicable and known, some type of government-funded assistance was received by 75-90% of people with a rare disorder. The most common decline was for psychological support (30%) and home and/or vehicle modifications (29%).

Only 16% people had some respite care in the last 12 months, with a range from 1 day to more than 30 days. Yet while another 7% of people qualified for respite care, they were unable to use it as it was either unavailable or did not meet their requirements.

> **I could not get anyone to help me at home following hospital discharge. I couldn't walk or move at the time and spent most days home alone. There was also no financial support when I had to leave my job. We nearly lost our house.**
>
> **Person with a rare disorder**

For three 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 they did not feel that their wairua ora/spiritual health had been considered in their healthcare.

Concluding Comment

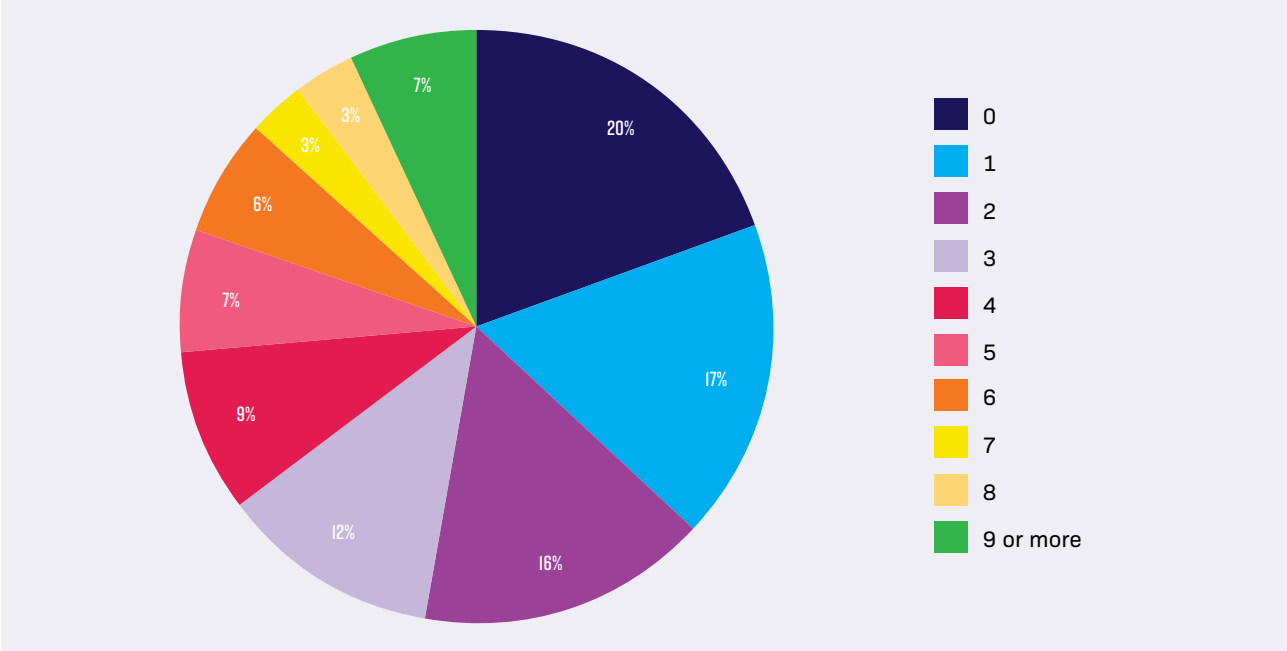
These results point to the need for improvements in how healthcare services are delivered to people who live with rare disorders. This could be achieved by implementation of RDNZ's recommendations for a rare and undiagnosed disorders centre of expertise.

Medicines and Treatments

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

80% people were taking at least one medicine (see Figure 14) however in the majority of cases this was not a modern medicine indicated specifically for treatment of their rare disorder.

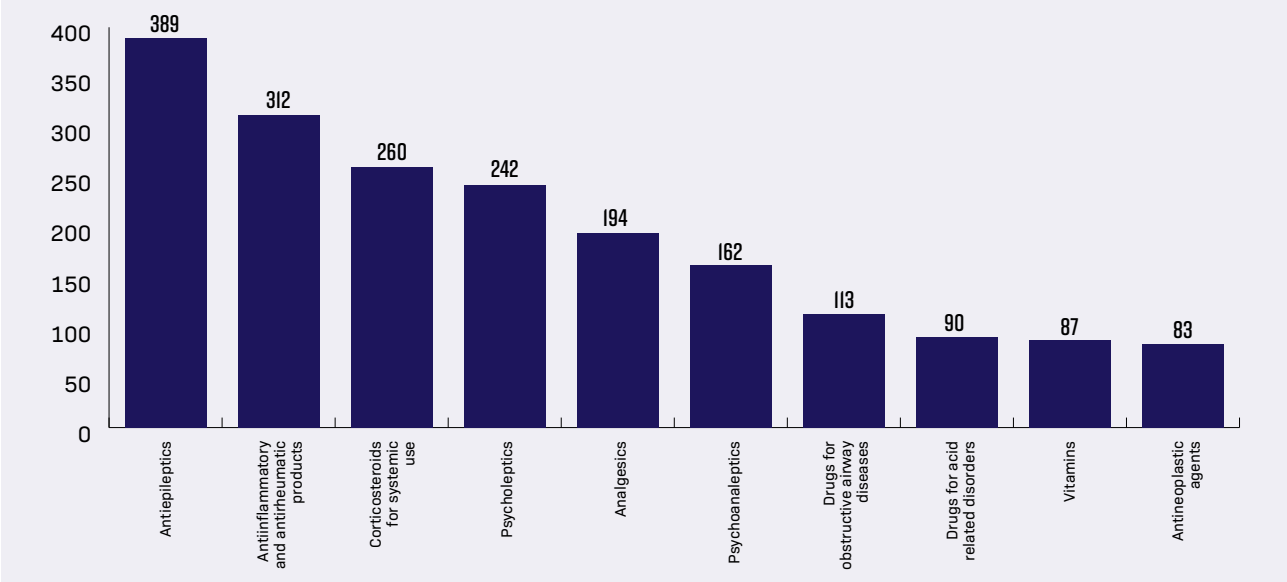
Figure 14: Number of medicines taken by people with rare disorders



There were a total of 398 different medicines listed and these have been further grouped according to their ATC code. The top 10 codes are presented in Figure 15. This equates to 3.2 medicines on average being taken per person. It was noticeable that the most common medicines prescribed and publicly funded were for conditions suffered as a consequence of having a rare disorder (e.g. pain and inflammation), rather than being a medicine for direct treatment of the rare disorder itself.

> **The meds are equivalent to using a hammer when a screwdriver is needed.**
>
> **Parent of a child with a rare disorder**

Figure 15: Number of people taking medicines, categorised by ATC code (top 10)



The most commonly reported medicines were ibuprofen for 295 people (34%), melatonin for 164 (19%), prednisolone for 146 (17%), gabapentin for 116 (13%), hydrocortisone for 104 (12%), salbutamol for 99 (11%) paracetamol for 83 people (10%). There were a number of people taking a broad range of different treatments, including antibiotics, antipsychotics, antidepressants, and vitamins (C, D, E).

Almost one in two people believed they were missing out on some medicines. Of those listed, the most frequent were for medicinal cannabis, Ketamine and Avastin which has changed since the previous survey and following public funding of some medicines (Trikafta for cystic fibrosis, Spinraza and/or Risdiplam for SMA).

> **The surveillance of my daughter's condition falls well short of the gold standard protocol. We cannot access medications that would relieve her symptoms, considered standard care throughout high-income countries.**
>
> **Parent of person with rare disorder**

Only 9% had attempted to gain access to medicines through the Named Patient Pharmaceutical Assessment (NPPA) process, funding a treatment for an individual patient whose clinical circumstances are exceptional. Where an application had been made though, for 55% it had been successful.

> **Heartbreakingly, before medicine was funded, the delay in time to get medication now has lifelong consequences for both our grandchildren.**
>
> **Grandparent of children with a rare disorder**

Almost one in three people obtain at least some of their current medicines through private/ out-of-pocket means and, for those taking this approach, almost all were self-funding, with financial impact and related consequences. It is anticipated that this could lead to inequity in medicine access.

> **Medications that would be far superior to what I'm having to take are beyond my ability to fund – as it is I'm having to self-fund as meds aren't covered by Pharmac.**
>
> **Person with a rare disorder**

In addition, one in two people reported taking or having at least one other treatment in the last 6 months, with the most commonly reported being a natural health product bought from a supermarket, health shop, chemist (29%) and other alternate or complementary medicine not prescribed by a doctor or nurse (17%).

Of those people that had thought about it, eight in ten 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.

Concluding Comment

These results point to the need for improvements in how people with rare disorders get access to the medicines they need.

Conclusions

The impact of living with a rare disorder in NZ is significant for all dimensions in the survey including; level of care required, healthcare services utilisation and access to treatments, coordination of care, overall costs, employment, education, care services, family life, stress and wellbeing.

Rare is many and represents a community of scale. There are likely to be more than 7000 rare disorders affecting around 300,000 New Zealanders.[12] Individual rare disorder prevalence can vary from as high as one in 2,000 people down to as little as only one individual in NZ being affected and it's this heterogeneity that needs to be considered in the rare disorders strategy and its implementation.[13] It is also important to look at the changing global context and international expectations, taking learnings from other countries to reduce the impact for people living with a rare disorder in New Zealand.

The overwhelming findings for the 2023 New Zealand Voice of Rare Disorders Survey show that despite increased knowledge internationally over the last two years about how to best support people with rare disorders this has not translated into better outcomes for New Zealanders.

Rare Disorders NZ (RDNZ) anticipating this and, in consultation with domestic and international experts, have identified four essential priority actions to drive needed changes.

- Close healthcare service & support gaps in New Zealand's health and other systems
- Need for better access for rare disorder medicines
- Ensure rare disorder coding is incorporated in health systems
- Implement strategy for rare disorders

Each of these essential actions, if fully implemented, will be important changes to improve outcomes for people living with a rare disorder in Aotearoa.

Close healthcare service & support gaps in New Zealand's health and other systems

The survey results paint a continuing picture of isolation, lack of timely and accurate diagnosis, poor treatment access, lack of coordinated care, significant carer impact and for many, being lost in the system.

- The majority of people with a rare disorder do not receive a timely or accurate diagnosis.
- People living with a rare disorder have high utilisation of healthcare services including specialist, GP visits, diagnostic tests and inpatient services
- Coordinating care is challenging for people with rare disorders, which can then be further complicated by healthcare teams.
- Access to appropriate care services was often variable for people living with a rare disorder, including suitable mental health and wellbeing support

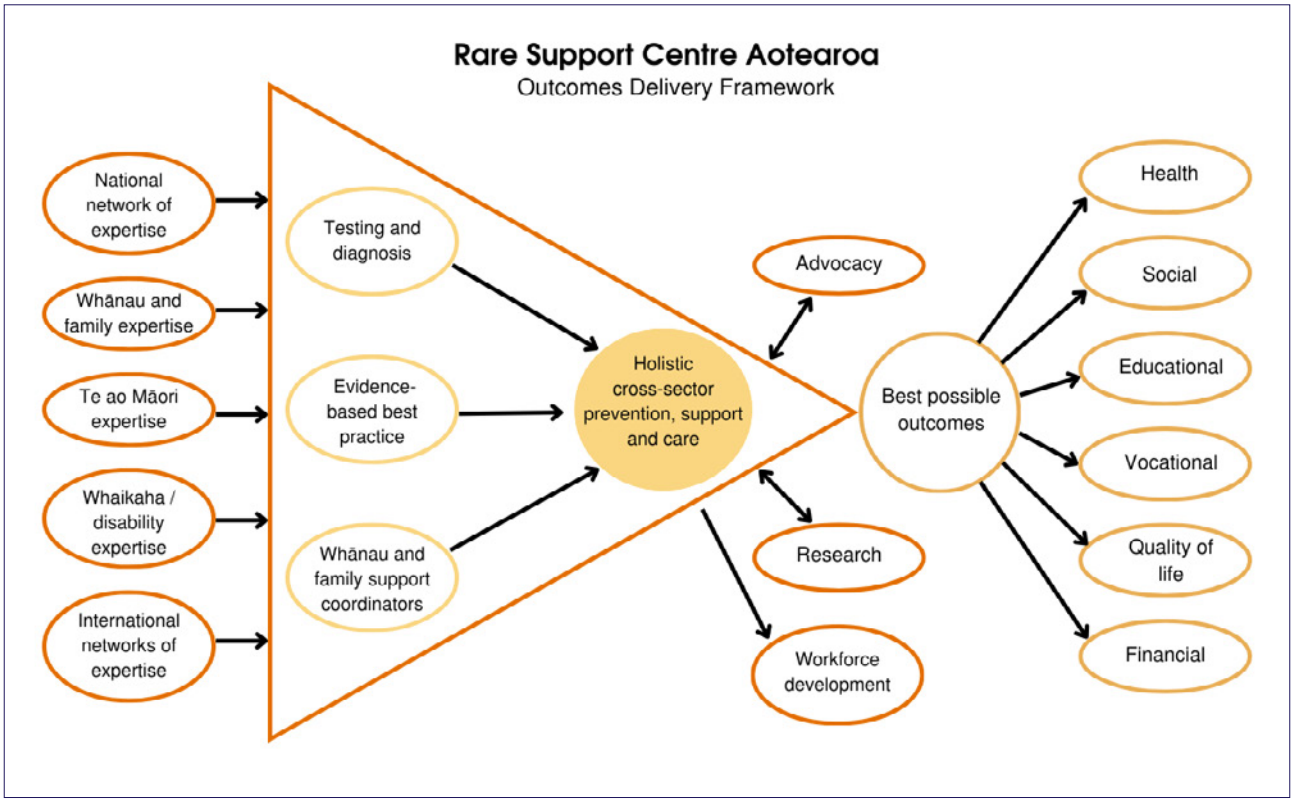
To address these specific gaps there is an opportunity to create a Rare and Undiagnosed Disorders Centre of Expertise (Action #1). This Centre would be a multidisciplinary team of internationally-networked experts within New Zealand. It will identify gaps in the delivery of service and support in New Zealand's health and other systems for people living with rare disorders. This builds on the work of other global initiatives. It will include expertise in areas of testing and diagnosis, evidence based best practice, holistic cross-sector support and care and family/whānau support.

A proposed delivery framework for achieving improved outcomes for all New Zealanders who have a rare or undiagnosed disorder is given in Figure 16.

12 https://www.raredisease.org.uk/what-is-a-rare-disease/

13 Nguengang Wakap S, Lambert D M, Olry A, Rodwell C, Gueydan C, Lanneau V, Murphy D, Le Cam Y, Rath A. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. European Journal of Human Genetics, 2019.

Figure 16: Outcomes delivery framework to rare disorders centre of expertise[14]



A Centre of Expertise comprising world class and world leading health, disability, education, social and other support, would be well placed to deliver equitable outcomes for people and whānau living with rare and undiagnosed disorders in New Zealand/Aotearoa.

Need for better access for rare disorder medicines

While some references in the literature suggest that currently up to 95% of rare disorders do not have effective treatments available, there have been great strides in the last 40 years. More than 600 medicines have been approved for rare disorders, and the current R&D pipeline has more than 700 medicines being developed for a wide range of rare disorders.[15,16]

However there is increasing disparity in access for rare disease treatments in different countries around the world, highlighted with the variable availability of a list of 204 designated essential rare disorder medicines.[17] In fact, one report showed New Zealand sitting squarely at the bottom of the OECD, with only 5 modern medicines to treat rare disease publicly funded between 2011-2020.[18] By comparison over the same period 45 modern medicines for rare disorders had been publicly funded in 19 other OECD countries. In many of the European countries, patients with rare diseases had publicly funded access to more than five-times as many modern medicines for rare diseases as New Zealand patients.[12]

The current one-size-fits-all model under Pharmac's pharmaceutical schedule simply does not work for low volume, high-cost medicines for rare disorders. The survey results further confirmed this, in that there were few accessible effective modern medicines for the majority of people, largely taking treatments for pain and inflammation. In addition the Named Patient Pharmaceutical Assessment (NPPA) process, which presents a number of inconsistencies and potential inequities as an alternative pathway to funding for rare disorder medicines, was something only a few people in the community were aware of. Importantly though, most people were worried that any promising treatments and medicines (e.g. gene therapy/CRISPR) will not be funded by New Zealand's public health system in the future.

14 Proposal for a Rare Support Centre Aotearoa (Rare and Undiagnosed Disorders Centre of Expertise), RDNZ, November 2023.

15 https://ojrd.biomedcentral.com/articles/10.1186/s13023-018-0936-x

16 https://phrma.org/-/media/Project/PhRMA/PhRMA%20Org/Sitecore%20Send%20Assets/MID-Rare-Diseases.pdf?cmid=b127b7de-f169-c377-6812-261f2fafdd18

17 https://ojrd.biomedcentral.com/articles/10.1186/s13023-021-01923-0

18 IQVIA (2021). A Decade of Modern Medicines: An international comparison 2011-2020. IQVIA International Inc., November 2021.

There is an opportunity to implement a **Single barrier-free pathway to rare disorder medicines (Action #2)**. This would provide a separate assessment pathway for rare medicines with criteria separate to common conditions and ensure equitable access to medicines for those with rare disorders. The following countries have already implemented alternative assessment pathways and funding for rare disorder medicines (Table 3)

Table 3: Medicine assessment pathways for rare disorders by country

UK	The Innovative Medicines Fund provides routes to faster patient access for non cancer medicines, including for rare disorders, while further data can be collected. This ensured that treatment using the latest health technologies can begin without delay and that NHS clinicians can help build the evidence-base for a new treatment; utilising the world-class skills and infrastructure of the NHS.[19]
Scotland	New Medicines Fund (NMF) established to fund the cost of orphan, ultraorphan and end-of-life drugs for patients. Since its establishment, access to end-of-life, orphan and ultra-orphan medicines has increased markedly.[20]
Canada	Total investment of up to $1.5 billion over three years in support of the first-ever National Strategy for Drugs for Rare Diseases to help increase access to, and affordability of, promising and effective drugs for rare diseases to improve the health of patients across Canada.[21]
Australia	The Life Saving Drugs Program (LSDP) pays for specific essential medicines to treat patients with ultra-rare and life-threatening diseases. All applications are assessed within 30 days. Currently funds 18 different life-saving medicines for 11 diseases including high cost medicines that do not meet criteria to be funded on the PBS.[22]
Japan	The Intractable Disease Health Care Act was initiated to secure a budget for maintaining medical expense subsidies for patients specific diseases whose prevalence is only ~0.1% and to promote research.[23],[24]

These alternative models use assessment criteria which recognise the societal aspect and the unique challenges related to accessing medicines for small populations such as those with rare disorders. These challenges include, relatively higher clinical trial costs, less people to pay for the medicines once production commences and challenging processes for reimbursement.

There is an opportunity for New Zealand to implement an alternative assessment pathway which takes into account approaches taken by these countries. This will be key to addressing the lack of modern medicine access that continues to fail the rare disorder community in this country.

Ensure rare disorder coding is incorporated in health systems data sets

There are likely to be more than 7,000 rare disorders in New Zealand, however the classification system for diseases currently used does not include most rare disorders. This exacerbates issues reported in this survey including variable access to necessary treatments and services, as well as poor communication between healthcare professionals. This lack of data is also problematic for proper funding and resource allocation of healthcare services.

There is an opportunity to **Incorporate coding of rare disorders in the classification system of diseases (Action #3)**, for up to 6,500 rare disorders using the most comprehensive database of rare disorders globally (Orphanet[25]). Ideally this should be incorporated into electronic health records (EHR) and administrative data sets.

19 https://www.england.nhs.uk/medicines-2/innovative-medicines-fund/#will-non-cancer-medicines-including-medicines-in-nices-highly-specialised-technologies-programme-qualify-for-managed-access-via-the-innovative-medicines-fund

20 https://www.gov.scot/publications/foi-202200287608/#:~:text=The%20NMF%20is%20available%20to,One%20and%20PACS%20Tier%20Two

21 https://www.canada.ca/en/health-canada/news/2023/03/investments-to-support-access-to-drugs-for-rare-diseases.html

22 https://www.health.gov.au/our-work/life-saving-drugs-program/about-the-lsdp

23 https://ojrd.biomedcentral.com/articles/10.1186/s13023-023-02762-x#:~:text=In%20Japan%2C%20 patients%20with%20 specific,for%20Patients%20with%20Intractable%20Diseases

24 https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5243159/

25 https://www.orpha.net/consor/cgi-bin/index.php?lng=EN

Implement strategy for rare disorders

The rare disorders strategy is due to be completed in 2024 and reflects that people and whānau with rare disorders can experience inequitable health outcomes and these outcomes are often influenced by unfair barriers, such as timely access to services and experiences navigating the health system.

Rare Disorders NZ, as the only national organisation supporting all New Zealanders living with a rare disorder and their carers, has already played an integral role in its development. In addition, through this survey it continues to provide the most representative and collective voice for people with rare disorders. Given this there is a key opportunity to Value RDNZ as a key partner in the implementation of the rare Disorders Strategy (Action #4).

Final Remarks

In New Zealand there is still work required for rare disorders to be regarded as a significant unmet need within our public health system. Rare disorders are not often viewed as a collective, but considered in isolation, and this leads to lost opportunities and neglect. The irony is that 'Rare is many', with up to 300,000 people living with a rare disorder in New Zealand, and with a further collective impact on their family, whānau and society.

Given this, it is welcome news that completion of a rare disorders strategy – which was a key recommendation from the 2021 survey – is imminent. However its measure of success will be in how well this is implemented to deliver equitable outcomes. To this end, Rare Disorders NZ have identified four essential implementable actions to drive needed changes and these have been referred to throughout this report:

Figure 17: Essential implementable actions to support people with rare disorders[26]

Action #1

Establishment of a Rare and Undiagnosed Disorders Centre of Expertise

Action #2

Establishment of a single barrier-free pathway to rare disorder medicines

Action #3

Incorporation of coding of rare disorders in the roll-out of the nationwide Electronic Medical Record

Action #4

Recognition of RDNZ as a key enabler for the Strategy's implementation

Finally, it is disappointing to see that there have been no significant improvements in outcomes over the last two years and that the collective impact for people with rare disorders, family and whānau remains significant. This has led to re-iteration of some of the key conclusions from the previous survey, with changes still needed to ensure improved health outcomes for people with rare disorders in Aotearoa.

26 https://www.raredisorders.org.nz/about-rare-disorders/rare-disorders-in-new-zealand/rare-disorders-strategy-progress/beyond-the-strategy/

About Rare Disorders New Zealand

Rare Disorders NZ is the national peak body organisation, supporting the 300,000 New Zealanders living with a rare disorder and the people who care for them.

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This white paper was written by HealthiNZ

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Advancing life-changing solutions