

# Impact of Living with a Rare Disorder in Aotearoa New Zealand

Importance of improving outcomes for people living with rare disorders, their family and whānau

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HealthiNZ

Advancing life-changing solutions

This white paper was written by HealthiNZ

## Acknowledgements

We first acknowledge the support of many Rare Disorder Patient Organisations in NZ who conducted the Voice of Rare Disorders survey that informed the recommendations outlined in this white paper. Particular mention must be made of the important contribution from the CEO of Rare Disorders NZ, Lisa Foster. Whilst the views and recommendations are wholly those of HealthiNZ they have been informed by the survey findings and by the growing evidence base and overseas policy development to improve outcomes for people living with rare disorders. Without Lisa's insight and leadership this would not have been possible.

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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 this area as a parent of a child who has cystic fibrosis.

## Executive summary

For New Zealanders living with a rare disorder today the impact is significant, affecting not only themselves but extending to affect families and carers, the health system and society. During a time of major health reform and United Nations recognition of the challenges facing those with rare conditions it is time for specific inclusion in Aotearoa so we leave no-one behind.

All change begins with awareness and in the largest survey of its kind ever conducted in this country, the **2021 New Zealand Voice of Rare Disorders Survey** provides an opportunity to understand the real impact of living with, or caring for, people with rare disorders, including comparisons with a similar survey conducted in 2019. This improves our evidence base for both the economic and societal impact and clarifies the need for a comprehensive plan to deliver sustainable improvements in health outcomes for a vulnerable and discriminated population.

### **Key findings from 2021 survey:**

- The majority of people with a rare disorder did not receive a timely diagnosis.
- Their **rare disorder is disabling and makes a number of everyday activities difficult.**
- High utilisation of healthcare services including specialist and GP visits and diagnostic tests **with the majority feeling these services have been impacted by COVID-19.**
- Many have been in hospital over the last 12 months. **One in three people were in hospital for an average of 13 days and one in seventeen people were in ICU for an average of 7 days.**
- There were **few accessible effective modern medicines available** for the majority of people. Most medicines publicly funded were used to relieve pain and inflammation, which are conditions suffered as a consequence of having a rare disorder.
- Most people living with a rare disorder believe that **professionals are poorly prepared to support them** and that there is a **clear lack of communication between service providers.**
- **Full employment and education is a challenge** for people and their carers: this may require modifying work arrangements through part time contracting; or continuing absence from school for children.
- Their rare disorder has **serious effects on social and family life, increasing tension** with family members and **triggering isolation** and feelings of being **neglected.**
- This all has a significant impact on people's mental health with **one in three often unhappy and depressed and feeling they cannot overcome their problems.**

The results paint a continuing picture of isolation, lack of timely diagnosis, poor treatment access, lack of coordinated care, significant carer impact and for many, being lost in the system. There is an opportunity to create a roadmap to improve health outcomes to positively impact communities, yet for this to occur the first significant action is for rare disorders to be specifically acknowledged within the Pae Ora (Healthy Futures) Bill.

Other recommendations include:

- **Widen engagement with communities** of people living with a rare disorder to provide a unified, collective voice, in particular with Māori, Pacific and ethnic minorities.
- Create a clear national strategy for specific collaboration amongst different stakeholders to implement **person centred co-ordinated models of care** for better quality of life for people with rare disorders, including those that are yet to be diagnosed. Enabling Good Lives model could be utilised as a basis.
- Promote **alternative assessment pathways and funding** to provide fair access to modern, life changing medicines for people with rare disorders.
- Continue to advocate for development of a **Health Strategy for Rare Disorders with priority actions for improved health and wellbeing.**

## Significant challenges remain for people living with rare disorders

Rare disorders have widespread impacts on people living with them, their families and carers, the health system and society<sup>1</sup>. Many of these conditions are life-long and debilitating and may lead to death at a young age. With approximately 50% of the people affected by rare disorders being children, not only does this impact on the child themselves but on the wider family and whānau. In a country that strives to be the best place in the world to be a child there must be acknowledgment of the impact of rare conditions and a desire to understand the specific barriers and challenges faced, so that directed actions can be taken within our reformed health system.

People often report being lost in the health system, starting with a long pathway to diagnosis, and difficulty in accessing treatments or healthcare services, including challenges surrounding coordination of care. There are systemic barriers to access disability and social support for many with rare disorders due to lack of awareness of the different conditions and the use of assessment criteria which often fails to include impairments and the resulting high complexity of need. The impact of rare disorders on mental, social and physical functions, household budget, employment and job careers, family life and well-being is significant. Yet there are solutions and opportunities to change this picture and improve health and wellbeing for those with rare disorders in Aotearoa.

The growing body of evidence from overseas has highlighted the significant impacts of living with a rare disorder and provided a catalyst for change. Consequently most OECD countries have national plans for rare disorders and clarified action plans. The United Nations (UN) Resolution on 'Addressing the Challenges of Persons Living with a Rare Disease and their Families' was recently adopted by all member states including New Zealand.

This survey provides evidence and clarity to understand the impact of living with a rare disorder in New Zealand in 2021, including what had changed in the two years since the last survey. Now we need action.

### A new global context

With the adoption of the first-ever UN Resolution on "Addressing the Challenges of Persons Living with a Rare Disease and their Families" by all 193 UN Member States in December 2021 there is a need for formal recognition of this marginalised population in New Zealand as a public health priority.

**"Recognizing the fundamental importance of equity, social justice and social protection mechanisms as well as the elimination of the root causes of discrimination"**

UN Resolution on PLWRD, 2021



It is anticipated that the impact of the resolution will be felt in NZ through increasing rare disease unity and empowerment of the global rare disorder community to break down barriers to care, diagnosis and treatment. There is international support and APEC action plans in place to offer a roadmap for countries to develop national strategies and plans, to have clear and specific national responses and policies and to advance the UN commitment to "leave no one behind" with strong international collaborations.

The Resolution provides some important direction for NZ that includes:

1. Specific inclusion and participation in society of people living with a rare disorder and their whānau
2. Ensuring universal and equitable access to quality health services without financial hardship
3. Promote national strategies and action plans that respond to resolve systemic barriers

<sup>1</sup> ANSEA report, 2019

## 2021 Voice of Rare Disorders Survey

To better understand the impact for people living with a rare disorder in NZ the Voice of Rare Disorders Survey was first developed in 2019 and has now been repeated in 2021.<sup>2</sup> This survey was conducted by Rare Disorders NZ on behalf of Patient Organisations, 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: Diagnosis, Care Profile, Healthcare services and treatments, Coordination of Care, Cost, Employment, School and well-being, Care Services, Family life, stress and wellbeing. Eligible participants were people who are living with a rare disorder in NZ or are 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<sup>3</sup> which to date has over 10,000 survey responses and BURQOL-RD.<sup>4</sup> Following from the experience in 2019 some additional questions were added, in particular relating to the extent of diagnosis, impact of COVID, and disability assessment. Also, where appropriate, comparisons with the population from the 2019 survey will be made and reported on in the results section. Overall this survey will further build on the growing overall evidence base of people living with a rare disorder in New Zealand.

Rare Disorders NZ currently engages with over 125 support groups representing at least 20,000 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 2<sup>nd</sup> November to 8th December 2021.

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

## Results

### Rare Disorder Diagnosis

Overall there were 718 responses with a confirmed diagnosis included in the primary analysis<sup>2</sup>. This represents **the largest ever survey of patient reported outcomes for people with rare disorders in NZ**, surpassing the previous 2019 survey with an overall 149% increase in response.

The primary analysis reported on the overall population of people with a confirmed rare disorder diagnosis. There were 239 different diagnoses, almost double those 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 Syndromes, Rare Cancers, Dystonia, Trigeminal Neuralgia, Cystic Fibrosis, Charcot Marie Tooth Disorder, Prader-Willi Syndrome and Muscular Dystrophy, Fragile X Syndrome, Complex Regional Pain Syndrome and Poliomyelitis.

There were 16 people excluded from primary analysis because they were undiagnosed. It is recognised that **early and accurate diagnosis of a rare disorder remains important to accessing a range of treatments and healthcare services.**

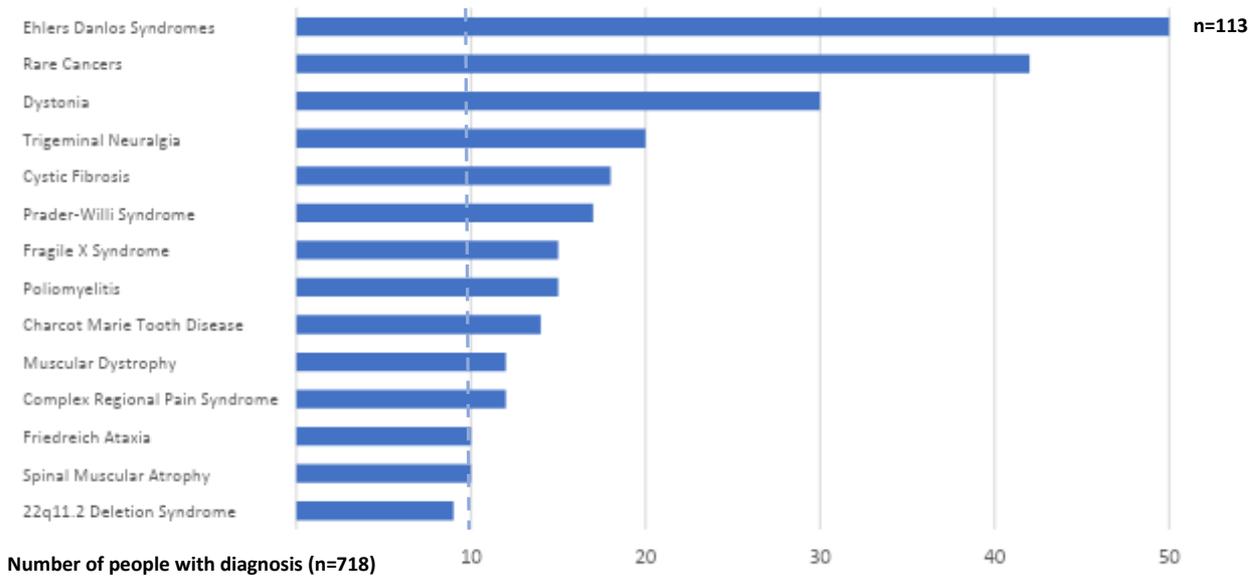
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<sup>2</sup> 2021 NZ Voice of Rare Disorders Survey

<sup>3</sup> <https://www.eurordis.org/rare-barometer-programme>

<sup>4</sup> 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)

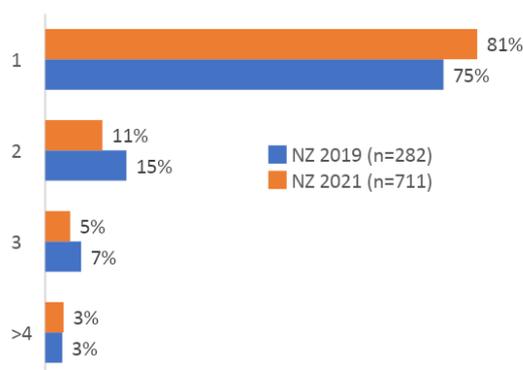
**Figure 1 Most common rare disease diagnoses in 2021 *Voice of Rare Disorders Survey***



The most common primary diagnoses in the 2021 survey expanded to include Rare Cancers, Dystonia, Complex Regional Pain Syndrome, Poliomyelitis and Cystic Fibrosis. None of these diagnoses were listed in the 2019 survey indicating better engagement in these communities.

Overall 19% of respondents reported more than one diagnosis, with 6% reporting 3 or more diagnoses. This appeared more likely in those respondents with a primary diagnosis of Ehlers Danlos Syndromes, Trigeminal Neuralgia, Mast Cell Activation Disorder. In the instance of Ehlers Danlos Syndromes 55% and 23% had a 2<sup>nd</sup> and 3<sup>rd</sup> diagnosis respectively.

**Figure 2 Number of people living with a rare disorder in households**



Results from the 2021 survey (see Figure 2) highlighted that while the majority of households included one person with a rare disease, for 19% of cases there were 2 or more people. Comparatively this was lower than the 2019 survey population (25%). **For one in twelve households there were 3 or more people living with a rare disorder.** We would anticipate that the impact for carers in these households is further compounded.

The majority of New Zealand survey respondents (68%) were married or in some live-in partnership arrangements. This is similar to the general NZ population with 56% of people aged over 15 being partnered.<sup>5</sup>

<sup>5</sup> Statistics New Zealand "Partnership status in current relationship by age group and sex, for the census usually resident population count aged 15 years and over, 2001, 2006, and 2013 Censuses (RC, TA, AU)" <nzdostat.stats.govt.nz>.

The overwhelming majority of people that responded described themselves as being New Zealand European at 85%. By contrast, the percentage of people that identified as being Māori or Pacific Peoples or Asian at 3%, 0.6% and 0.8% respectively, was lower than National Census estimates and lower than in the previous survey. Moving forward, this highlights the need to improve engagement with these populations.

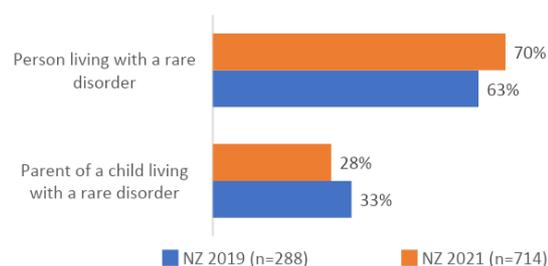
In line with population, the majority of responses were from Auckland, Canterbury and Wellington regions, similar to 2019 trends. In comparison with overall Region population estimates the Auckland region was under-represented and the Wellington region was over-represented in the survey responses.

Overall **over 80% of responses were from people living in urban areas** or areas with moderate to high urban influence.

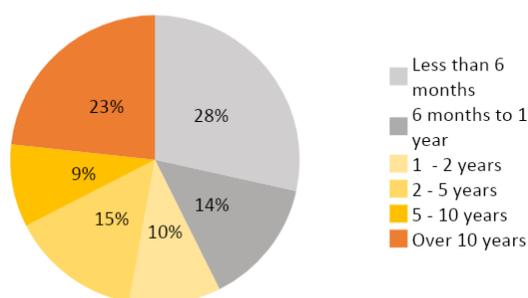
The majority of responses (70%) were provided by people living with a rare disorder, which was higher than in 2019 (63%) (Figure 3). Of these, **almost one in seven had an additional role, either as a parent, sibling or spouse of another person living with a rare disorder.**

Overall 28% of the New Zealand responses were from parents of a child living with a rare disorder.

**Figure 3 Relationship to person with rare disorder**



**Figure 4 Time taken to get diagnosis**



The majority had challenges in getting a diagnosis for their rare disorder:

Over half took longer than 1 year to get a diagnosis and **for one in five the time taken to get a diagnosis was over 10 years** (Figure 4).

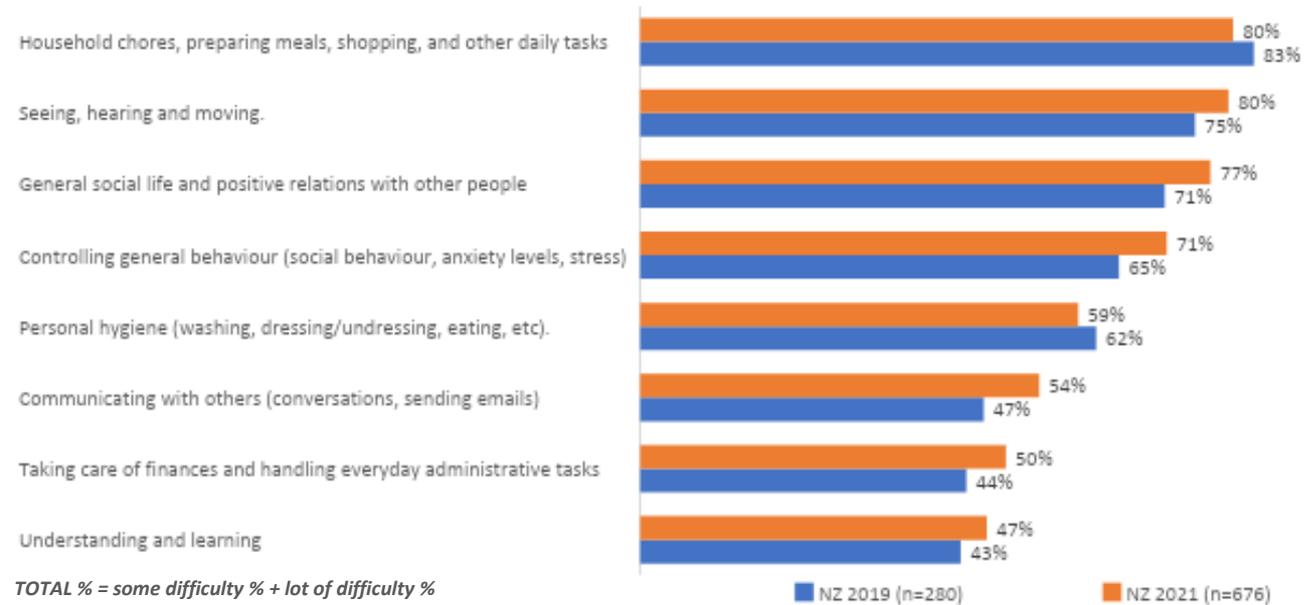
The majority (64%), had to visit 3 or more doctors to get a diagnosis and **for one in ten over 10 doctors were visited.**

62% people with a rare disorder were misdiagnosed at least once before final diagnosis was confirmed. **One in ten were misdiagnosed at least twice.**

## Care Profile

The extent to which the person living with the rare disorder could perform certain activities varied with the activity. For **60-75% of people and their families surveyed, the rare disorder makes a number of** everyday activities difficult (communicating, controlling behaviour, social life) and was generally higher in 2021.

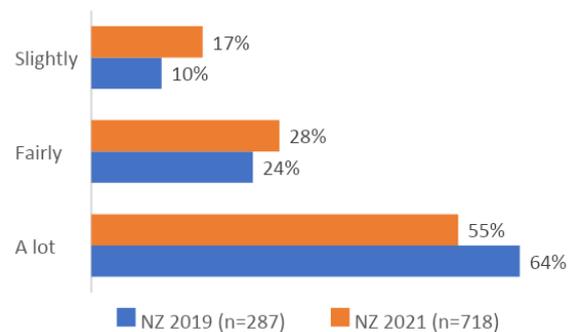
**Figure 5 Activities of daily living that provide greatest difficulty**



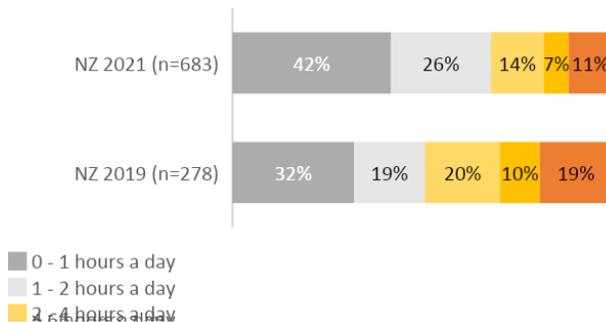
**Overall, almost 55% reported that the disorder impacted a lot on their health and everyday life.**

The most frequently reported household carer was the person living with the rare disorder > mother> the spouse, in that order.

**Figure 6 Impact on health and everyday life**



**Figure 7 Daily time investment for illness-related tasks**



**32% 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.

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

## Healthcare services utilisation and cost estimates

**Table 1 Healthcare service utilisation for people living with rare disorders**

Service	Utilisation rate (%)	Total Annual frequency	Cost per test/visit/day <sup>6</sup>	Average cost per patient
Specialist visits	81%	8.0 visits	\$250	\$1,882
Diagnostic tests	75%	7.6 tests		
GP visits	87%	4.8 visits	\$80	\$384
ED visits	35%	1.4 visits	\$370	\$500
Admissions	38%	1.0 admissions		
Inpatient days	37%	12.7 days	\$1,200	\$15,242
ICU Inpatient days	6%	6.9 days	\$5,500	\$38,097

There was high utilisation of healthcare services with a **large number of people having seen a specialist or GPs over the last 70 days (81-87% utilisation rate by respondents)**. There were also one in three people with a rare disorder that presented at the hospital in the last year. **On average they spent almost 13 days as an inpatient in the last year and, for one in seventeen people, almost 7 days in ICU.** This is a significant finding and highlights the impact and pressure on ICUs, especially in the current context of a COVID-19 pandemic.

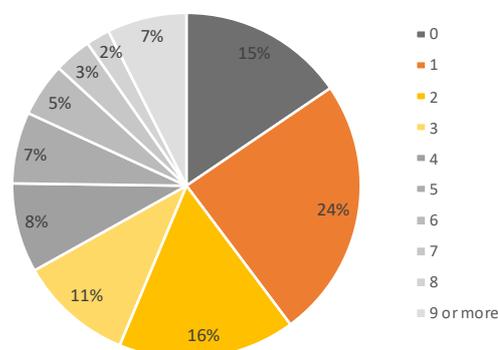
The **majority of respondents (58%) felt there was some or significant impact of COVID-19 on their healthcare services.** The main effects were through cancellation or postponement of appointments, delays in screening, tests or surgery.

## Medicine

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

- 85% people were taking medicine for their rare disorder (see Figure 8) however in the majority of cases this was not a specific modern medicine. This is not surprising given references in the literature that 95% of rare disorders do not have effective treatments available.<sup>7</sup>

**Figure 8 Number of medicines taken by people with rare disorders**

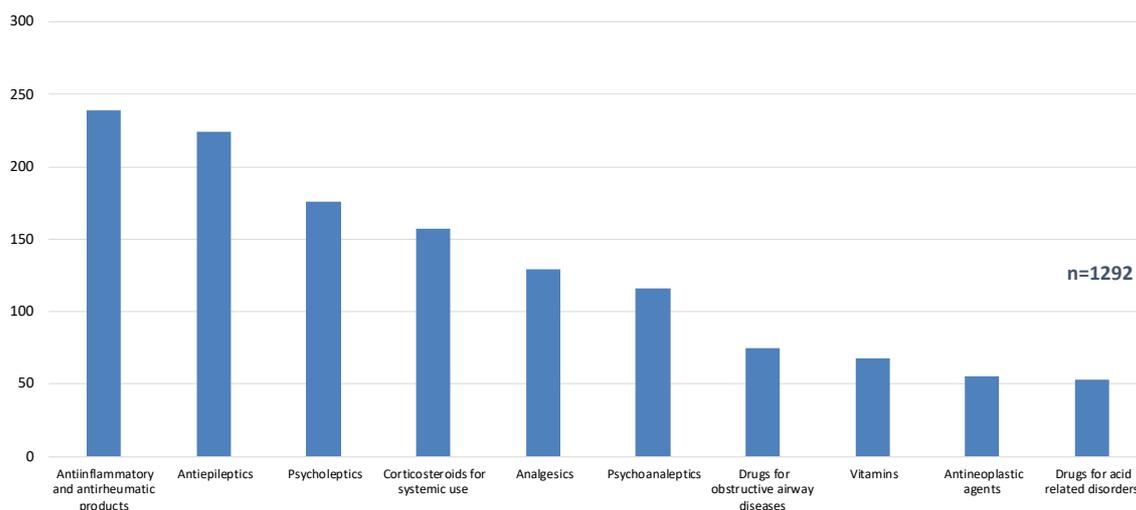


<sup>6</sup> Using the Cost Resource Manual (PHARMAC): cost per specialist visits has been applied at \$250 (based on physician outpatient costs, subsequent visit); cost per GP Practice visit applied at \$80; cost per nurse visit applied at \$40; cost per emergency department visit applied at \$370; 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

<sup>7</sup> <https://ojrd.biomedcentral.com/articles/10.1186/s13023-018-0936-x>

There were a total of 414 different medicines listed and these have been further grouped according to their Anatomical Therapeutic Chemical (ATC) code. The top 10 codes are presented in Figure 9. It was clear 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 medicines for direct treatment of the rare disorder itself.

**Figure 9 Number of people taking medicines, categorised by ATC code (top 10)**



The most commonly reported medicines were ibuprofen for 220 people (36%), melatonin for 99 (16%), prednisolone for 82 (14%), paracetamol for 63 (10%), Gabapentin for 62 (10%) salbutamol for 62 people(10%) , and hydrocortisone for 61 people(10%) . There were a number of people taking a broad range of different treatments, including antibiotics, antipsychotics, anti-depressants, and vitamins (C, D, E).

The majority of people did not know if they were missing out on any medicines. The exception was for specific conditions where medicines are publicly funded and available in other countries but not New Zealand. e.g. for people with cystic fibrosis 13/15 (87%) respondents felt they were missing out on Trikafta. For people with SMA 9/10 (90%) mentioned Spinraza and/or Risdiplam.

*“Starship hospital have been amazing. Unfortunately not having life changing treatments funded for her condition has been awful and we are moving to Australia to access treatment”*  
**Parent of person with rare disorder**

Of further note, a significant proportion of respondents ( 30% ) were having to undertake self-funding for at least some of their medicines, with financial impact and related consequences. It is anticipated that this could lead to inequity in medicine access.

## Coordination of Care

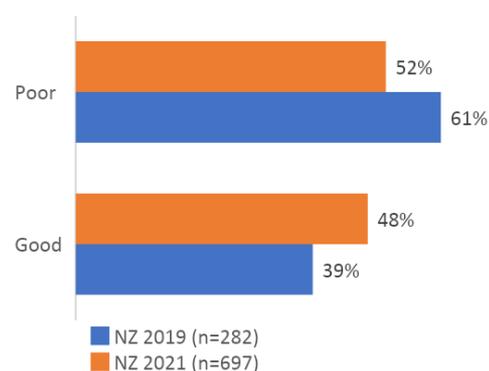
Almost half of the respondents felt that organising care was difficult to manage. This included finding information on your disorder, understanding your rights and administrative procedures (48%) and finding the right professionals (58%).

Over 50% felt that **communication and information exchange between different service providers was poor** (Figure 10). This was sometimes linked to co-ordination or ability of healthcare professionals to access data, leading to frustration.

While various healthcare professionals were a main first source of information the persons friends and family were vital. There was a reduced preference for internet accessed information compared with 2019.

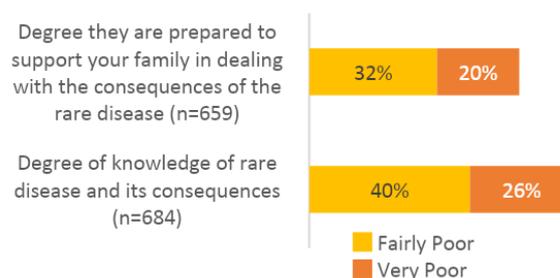
*"I have to constantly repeat myself to different doctors which is frustrating. I just wish they all had access to my latest data"*  
**Person with rare disorder**

**Figure 10 Quality of communication between service providers**



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 11).

**Figure 11 Level of support and knowledge from support services**

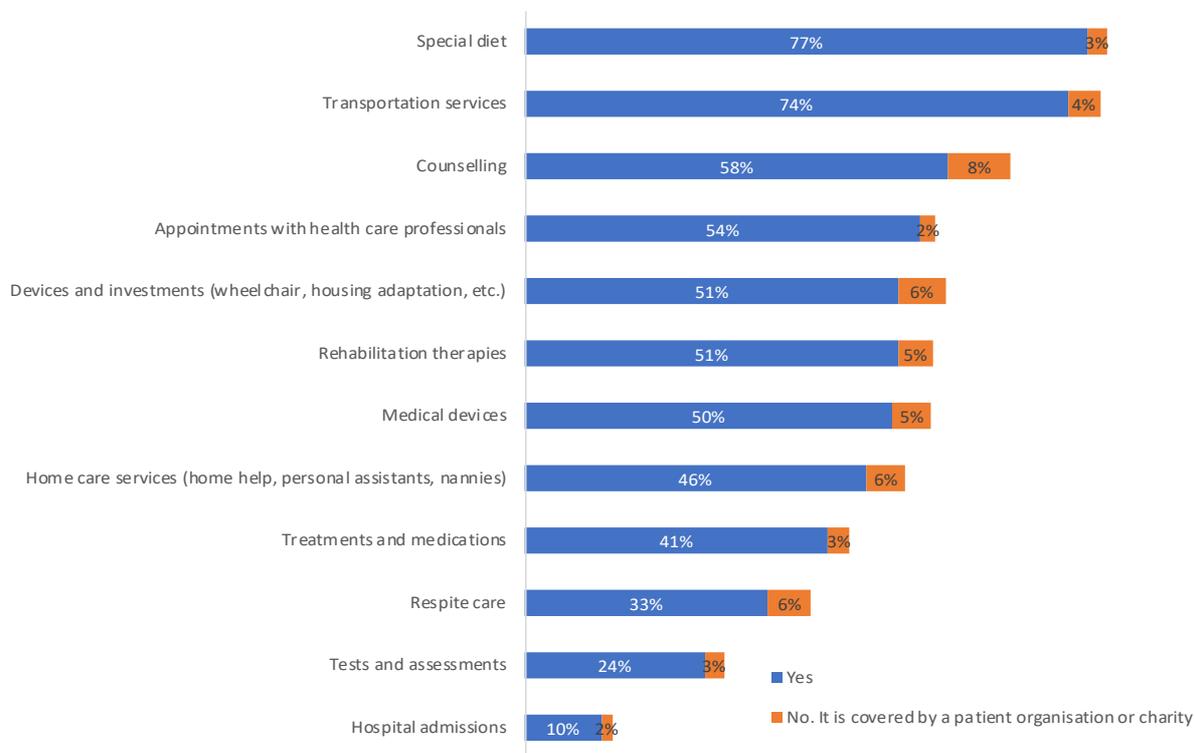


Overall, people did not feel informed at all around the rights related to the consequences of their rare disorder (76%), relevant social services that can help them (73%) or financial help they could be entitled to (71%).

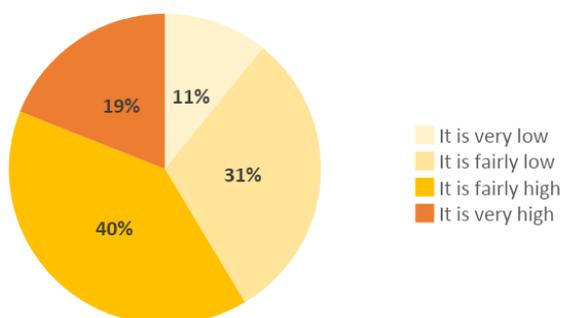
## Cost of living with a rare disorder

There were many costs covered by people living with the rare disorder or their family with approximately half covering some costs.

**Figure 12 Range of privately covered healthcare costs**



**Figure 13 Perception of costs associated with the rare disorder**



The majority (59%) of people felt that the costs associated with the rare disorder were high (Figure 13).

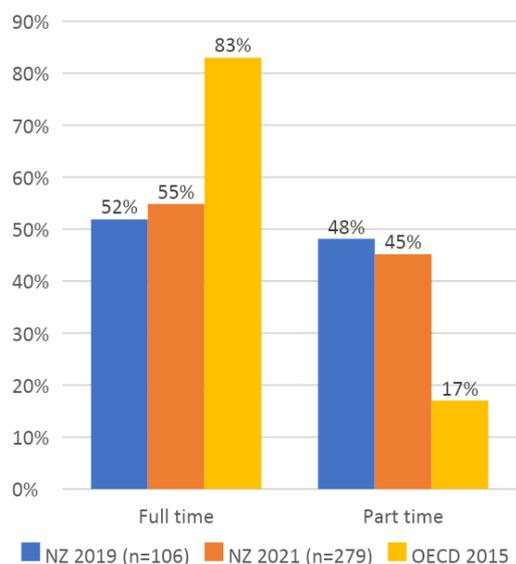
In addition, 54% felt the costs associated with managing their rare disorder were hard to manage.

One in three people with a rare disorder had not been submitted for an official health assessment to determine their level of disability.

Furthermore, of those assessed, four in ten felt their assigned percentage of disability was too low.

## Employment

**Figure 14 Impact on employment:  
Comparison of NZ and OECD**



The proportion of **people in part time employment was higher than against an OECD average** (Figure 14).

*“They made me feel like they didn't want me there due to my disability.”*  
**Person with rare disorder**

**Overall 25% were unemployed, with the majority citing long-term illness or disability.**

The way the person’s employment was affected by their rare disorder included **limiting professional choices, reducing or stopping professional activity and limiting job opportunities**. It **caused a decrease in income for 70% people**.

**15% of people were absent from their work for more than 60 days** due to their rare disorder.

The majority would like adapted accessible work environments that took account of their rare disorder and would like their working hours and responsibilities to remain similar or increase.

## Education

While most children of school age went to a mainstream school with or without adaptation, almost one in five went to a specialised school or had home schooling.

Almost 80% felt the rare disorder had a moderate to significant impact on their learning.

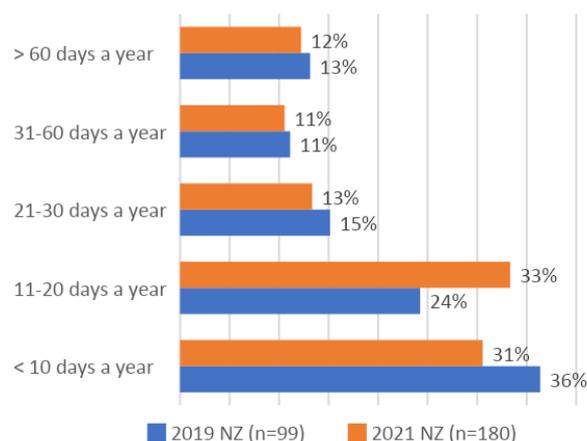
Many believed better Ongoing Resourcing Scheme (ORS) funding access would improve their situation, with 34% receiving ORS funding.

*“What would improve the situation for my son is psychological assessment and increased access to learning support”*

**Parent of child with rare disorder**

Overall there was a range of days that children were absent from school due to their rare disorder in a year with **23% children being absent for more than 30 days per year** (see Figure 15).

**Figure 15 Days absence from school**



## Care Services

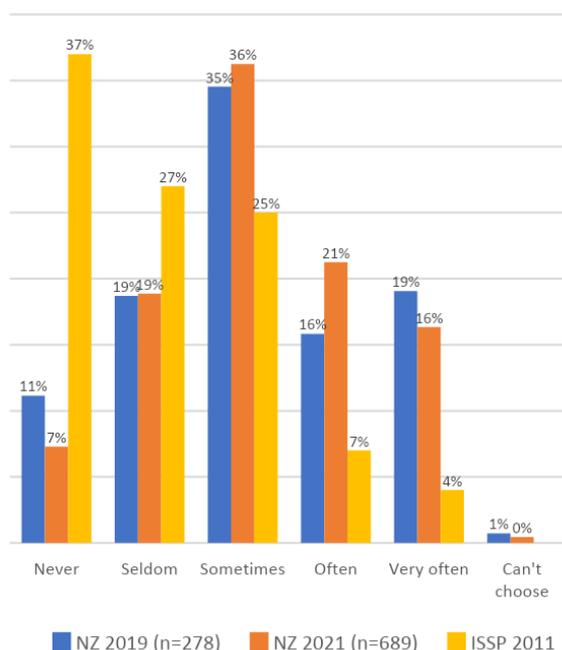
- There was a broad range of hours per week received for rehabilitation services and therapies although mainly between 1-3 hours.
- There were generally more hours received for support for house chores and daily tasks with 13 (3%) of people receiving more than 12 hours.
- 18% of people had had some respite care in the last 12 months which was lower than two years ago (25%). In addition, a further 14% of people felt they could benefit from respite care but did not currently qualify for it.

## Family life, stress and wellbeing

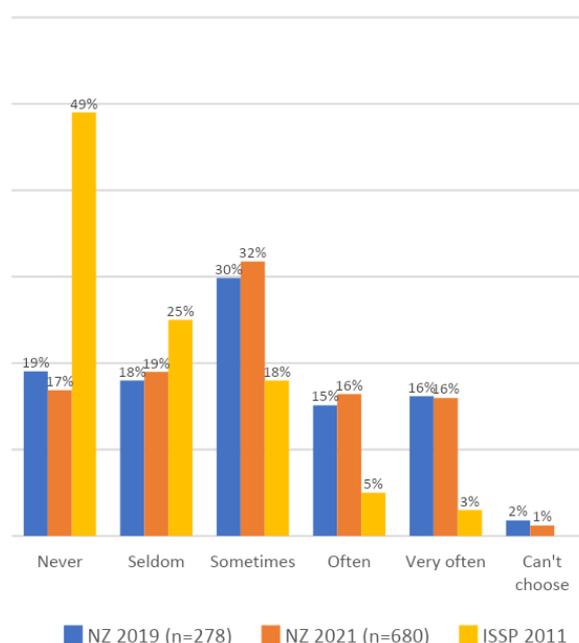
Since the symptoms started the **majority of people have experienced increased tension between family members (57%) and isolation from family and friends (69%)** amplified by or caused by their rare disorder. With the added uncertainty and tension from the COVID-19 pandemic this creates a serious 'cumulative trauma' situation for many living with a rare disorder and yet there are no specific flags, actions or pathways for access other than GP.

Concerningly a further **one in three respondents often felt unhappy and depressed and felt they could not overcome their problems** (Figure 15, Figure 16). These issues were much higher than in general populations (based on International Social Survey Programme, 2011).

**Figure 16 Extent person felt unhappy and/or depressed in last month: Comparison to general population**



**Figure 17 Extent person felt they could not overcome their problems in last month: Comparison to general population**



Recognition and inclusion of mental health and wellbeing support need to be part of integrated holistic health care.

## 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.

A significant fact is that rare is many and represents a growing community of scale. There are likely to be more than 6,000 rare disorders affecting around 300,000 New Zealanders,<sup>8</sup> and precision healthcare and genomic technologies present opportunities to reveal even greater numbers of rare disorders previously misdiagnosed or not diagnosed at all. 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 policy development.<sup>9</sup> This population of persons impacted by rare disorders needs to be included and accounted for in future focused plans in a comprehensive manner.

We also need to look at the changing global context and international expectations, taking shared learnings from other countries to reduce the impact for people living with a rare disorder in New Zealand. A number of these opportunities are clarified here.

### 1. Need for wider community engagement, in particular with Māori, Pacific and ethnic minorities

Small numbers of people living with a wide variety of rare disorders creates repetitive challenges on our health and welfare systems, creates an economic burden and often leads to feelings of isolation, yet having a National Health Strategy for rare disorders with appropriate action plans offers cohesive solutions and cost savings that other countries have long appreciated. Living with a rare condition in a sparsely populated country like New Zealand emphasises this need rather than excludes it, and the cohesion and value-add of a national peak body for rare disorders is clear. The unified voice of rare disorders that acknowledges Te Tiriti obligations and includes sensible future health policy is the only way forward.

This includes active engagement with tangata whenua, along with serving our Pacific communities and ethnic minorities to ensure inclusion in a community facing the same challenges to offer a sense of belonging and hope. Harnessing and nucleating the wisdom of the team of 300,000 could make an enormous difference to the all too common experience of living with something rare.

This survey offers a vehicle for a significant and unified voice that is mostly dismissed in New Zealand and with the evidence of the largest survey ever conducted in Aotearoa New Zealand this voice can no longer be ignored.

The health and disability reform acknowledges that providers and organisations can struggle to engage consumers in a meaningful way. Given this, it is heartening to see that Rare Disorders New Zealand currently engages with over 125 support groups representing over 20,000 people living with different rare disorders. This is an increase from 2019 with an associated increased survey response and almost doubling of diagnoses reported, plus an increased breadth of diagnoses being included in the 2021 survey.

Wider engagement with key stakeholders beyond people living with rare disorders also reinforces a person-centred focus, including with key peak bodies, governments, researchers, clinicians and industry. Examples over the last two years include with the Genomics think-tank, Carer's Alliance, Neurological Alliance and Parent to Parent. Despite this, there remained a low percentage of people responding in the survey that identified as being Māori or Pasifika or Asian. While we could not conclude on rurality and socioeconomic status, these groups are

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<sup>8</sup> <https://www.raredisease.org.uk/what-is-a-rare-disease/>

<sup>9</sup> 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

typically associated with poorer health outcomes due to impact of colonisation and discrimination. This highlights the need to create new ways of inclusion for Māori with increased engagement with these populations, For this reason a temporary role of Māori Engagement Officer at Rare Disorders NZ has been established.

## 2. Implement person centred co-ordinated models of care

People living with rare disorders often have complex needs and face unique barriers of access to care, treatment, education, employment and mental health. Clinicians also face the challenge of limited awareness, information or connections to specialists with expert knowledge on rare disorders. This is highlighted in an often long and convoluted pathway to diagnosis with many having long delays, cycling through multiple specialists and being misdiagnosed at least twice.

This long 'diagnostic odyssey' impacts mental health and wellbeing, with many being told it must all be in their head. Additionally, a health system that is underpinned by confirmation of a person's diagnosis, being able to tick a box or having a disorder that is well known in order to access services, creates challenges which negatively impact those with rare conditions. These multiple system issues and complexities mean a planned approach to healthcare service delivery is required to achieve equitable outcomes through new integrated, aligned models of care that recognise these realities.

Expecting there will be inclusion with no recognition is unrealistic and it will take inspirational leadership to realise that rare disorders require a strategy that can then align, connect and dovetail into health models we already have in place.

It is also critical that evolved models of care are underpinned by rare disorder data that has been specifically gathered. While this survey is an important step in collecting valuable data about the collective experience, it is acknowledged data limitations across the health system create invisibility within treasury, health systems and for policy analysts and politicians who then disregard this sizeable population in future planning.

## 3. Alternative assessment pathways for medicine evaluation

Access to medicines for rare disorders needs to take into consideration small patient populations, limited data and progressive disease. Analysis requires a societal approach, not a health sector approach - taking into consideration not only the impact on the health care system, but also impacts and cost due to loss of parental income, need for social supports, vehicle modifications, equipment, support in preschool and school etc. Limitations of data collection include challenges obtaining such quality of life information required for Health Technology Assessments (HTAs), as well as assessing impact for the family, whānau and wider society. This leaves the rare disorder community as a vulnerable population, invisible to policy analysts.

There is increasing disparity in access to treatment for rare disorders around the world, with New Zealand at the bottom of 20 comparable OECD countries. Only 5 modern medicines to treat rare disease have been publicly funded between 2011-2020 in New Zealand.<sup>10</sup> By comparison over the same period 45 modern medicines for rare disorders have 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.<sup>12</sup>

Given this difference it is important to understand alternative assessment pathways and cost-sharing models implemented overseas that could support better access to modern, life changing medicines. Increasingly many

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<sup>10</sup> IQVIA (2021). A Decade of Modern Medicines: An international comparison 2011-2020. IQVIA International Inc., November 2021.

seek patient input at pivotal assessment stages and request clinical expertise for specific disease types. The following models operate alternative medicine evaluation pathways for people living with a rare disorder:

- **England NICE Model:** The UK has created a more appropriate and flexible framework for bringing the treatment for rare disorders to patients with clear timelines and transparency. Their innovative licensing and access pathway provides accelerated patient access to safe and innovative medicines. In Scotland and UK, NICE published an increased threshold of £100,000 per Quality Adjusted Life Year (QALY) gained for drugs for “very rare” disorders.
- **Scotland PACE:** This has an additional layer in the appraisal process giving patient groups and clinicians a strong collaborative voice in Scottish Medicines Consortium decision making.<sup>11</sup>
- **Australia Life Saving Drugs Programme (LDSP):** Currently funds 14 different life-saving medicines for nine very rare disorders through the LSDP including high cost medicines that do not meet criteria to be funded on the PBS.<sup>12</sup> A recent review has recommended the establishment of a Centre for Precision Medicine and Rare Diseases, the Health Technology Assessment (HTA) process for cell and gene therapies be simplified to establish a clear and certain pathway for such therapies. The objective of the Centre would be to ensure that the capacity of the Department of Health is enhanced to provide Australians with timely access to new drugs and novel medical technologies, including for rare diseases.<sup>13</sup>

These alternative models use assessment criteria which recognise the societal aspect and the specific challenges accessing medicines for small populations such as those with rare disorders. These challenges include, higher costs to complete clinical trials due to smaller clinical trial populations, less people to pay for the medicines once production commences and costly non-transparent application processes.

In this 2021 survey the range of medicines people with rare disorders reported taking were extremely limited and mainly focused on symptom management such as reducing pain and inflammation. While the majority did not even know if they were missing out on innovative medicines, those most frequently cited for relevant rare disorders (Trikafta and Spinraza) have all been highlighted in the media and advocated for by patient communities. They may be good candidates to assess alternative pathways for medicine evaluation.

## 4. Develop New Zealand National Rare Disorder Health Strategy

Systemic change is needed within healthcare to achieve the best outcomes for those living with a rare disorder. The overall goal of Pae Ora (Healthy Futures) must include those on the margins who face the deepest challenges within a system designed for common conditions. The evidence from this survey is clear and the time for change is now with blueprints of success from other countries available – it simply needs commitment and inspired leadership to enact a brighter future for all people with rare disorders.

A New Zealand Rare Disorder Health Strategy would create a roadmap and pathways that ensure people living with rare disorders are not left behind. Priorities could be determined like shown in the UK action plan which highlights the importance of raising awareness of rare diseases among healthcare professionals and supporting clinicians, plus capturing vital data on rare conditions.

The goal of a comprehensive plan is to reduce current inequities; to listen and hear the experts who face barriers in a system not designed with their needs in mind, and create a transformational health system that is equitable and leaves no-one behind. The rare disorder voice can only be heard if it is acknowledged and we desperately need such recognition as a starting point for constructive answers and authentic engagement.

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<sup>11</sup> <https://www.scottishmedicines.org.uk/how-we-decide/pace/>

<sup>12</sup> <https://www.health.gov.au/ministers/the-hon-greg-hunt-mp/media/medicine-for-rare-disease-made-free-on-the-life-saving-drugs-program>

<sup>13</sup> The New Frontier - Delivering better health for all Australians, November 2021 (<https://www.health.gov.au/ministers/the-hon-greg-hunt-mp/media/medicine-for-rare-disease-made-free-on-the-life-saving-drugs-program>)

**Table 2** Country plans based on the UK strategy

Country	Plans published	Year of issue	Issuing body
England	UK Strategy for rare Diseases: Implementation plan for England	Jan 2018	Department of Health and Social Care
Scotland	It's Not Rare to have a Rare Disease	June 2014	Scottish government and other stakeholders
Wales	Welsh Implementation Plan for Rare Diseases	February 2015	National Implementation Group and other stakeholders
Northern Ireland	Providing High Quality Care for people affected by Rare Diseases	October 2015	Department of Health in Northern Ireland

This has been echoed by the Australian National Strategic Action Plan for Rare Diseases, where Australian academics called for an action plan in 2010 and it launched in 2020 with support from the Federal Government. The ongoing associated action plan has led to evidenced economic and health improvements, conversely New Zealand has no specific policy and is falling further behind these countries.

A national strategy and action plan for rare disorders would acknowledge current systemic barriers so it can resolve them. It would offer a clear and comprehensive plan for the future that ensures our health and disability services, and workforce are educated, prepared and resourced to support those with uncommon conditions.

## Final Remarks

In New Zealand a shift in mindset is needed for rare disorders to be regarded as a significant unmet need within our public health system. Unlike common illnesses such as cancer, rare disorders are not 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 that is significant. Taking this wider impact into account, we estimate **thirty percent of our population is impacted by a rare disorder in some way**.

Today, many people living with a rare disorder in this country remain marginalised and largely invisible, with no clear strategy for inclusion within the healthcare system, despite being a significant part of the population. It is disappointing that there have been no significant improvements over the last two years, and this survey highlights the ripple effect and extensive impact rare disorders can have throughout our society.

This has meant re-iterating some of the conclusions from the previous survey, with a number of changes required in order to support effective policy that leads to improved health outcomes for people with rare disorders in NZ. This white paper includes the following recommendations:

- **Recommendation 1: Continue to widen engagement with communities** of people living with a rare disorder to provide a collective voice, in particular with Māori, Pacific and ethnic minorities.
- **Recommendation 2:** Increase collaboration amongst different stakeholders to **implement person centred co-ordinated models of care** for better quality life for people with rare disorders, including those that are yet to be diagnosed.
- **Recommendation 3:** Promote **alternative assessment pathways** to provide better access to modern, life changing medicines for people with rare disorders.
- **Recommendation 4:** Continue to advocate for development of a **New Zealand Rare Disorders Health Strategy and Action Plan** aligned with other current healthcare implementation plans.