Impact of Living with a Rare Disorder in NZ

Why a different approach is needed to improve outcomes for people living with rare disorders, their family and whānau

February 2020

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.

This white paper report was commissioned by Medicines New Zealand

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.

In the largest survey of its kind ever conducted in this country, the **2019 NZ Voice of Rare Disorders Survey** provided an opportunity to understand the real impact for living with or caring for people with rare disorders. Not only improving our evidence base in this area but also providing an impetus for demanding changes that deliver sustainable improvements in health outcomes.

Key findings from survey:

- For 60-75% of people and their families surveyed, the **rare disorder makes a number of everyday activities difficult**.
- The time burden is substantial for people living with a rare disorder and their carers; the **majority requiring over 2 hours per day for care and coordination**.
- High utilisation of healthcare services including specialist and GP visits and diagnostic tests. **One in three people were in hospital for an average of 16 days per year**. **One in twenty people were in ICU for an average of 8 days per year**.
- There were **almost no effective treatments accessible** for the majority of people other than for reducing inflammation.
- Most people living with a rare disorder and their carers consider 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 living with a rare disorder and their carers: this may require modifying work arrangements through part time contracting; or continuing absence from school for children.
- The disorder has **serious effects on social and family life**, **increasing tension** with family members and **triggering isolation** and feelings of being **neglected.**
- Mental health of people living with a rare disorder is worse in comparison with the general population with **one in three often unhappy and depressed** and **feeling they cannot overcome their problems**.

The results painted a picture of isolation, poor treatment access, lack of coordinated care, significant carer impact and for some, being lost in the system. It highlights the need for changes and a number of recommendations have been made:

- Develop an inclusive **consistent definition for rare disorder**. This is essential in order to enable policymakers to develop a national plan for rare disorders.
- Build a collective voice for people living with rare disorders through **wider community engagement**, in particular to include Māori, Pacific and ethnic minorities
- Greater collaboration amongst different stakeholders to **implement patient centric co-ordinated models of care** for better quality life for people with rare disorders.
- Ensure funding mechanisms for medicines for rare disorders consider an economic evaluation method that not only considers direct medical costs and medicine costs but other societal costs.
- Develop a **NZ Rare Disorders Framework and Action Plan.**

Significant challenges remain for people living with a rare disorder

Rare disorders have widespread impact on people living with them, their families and carers, the health system and society (Figure 1). The condition often impacts a range of daily activities and requires a significant amount of time each day for the person and their carer(s) to manage. Many of these conditions are life-long and debilitating and may lead to death at a very 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.

People often report that their condition affects their emotional and mental wellbeing. Depression, stress and anxiety are commonly associated with people with rare disorders with reasons likely to include health and disability status, potentially years of misdiagnosis and difficulty in accessing treatment or healthcare services. People can be disadvantaged when it comes to employment or education due to time taken off to manage their condition, as well as lack of accommodations being made. All of this impacts the financial wellbeing for people with rare disorders and their families.

Figure 1 Complex health and social challenges affecting people living with a rare disorder[1]

While there is a growing body of evidence overseas highlighting the significant impact of living with a rare disorder, the situation in NZ is less well understood. A patient survey was therefore developed with the overall objective being to explore the health impact and social needs of people living with a rare disorder and their carers in this country, specifically by:

- Assessing the impact of rare disorders on mental, social and physical functions, household budget, employment and job careers, family life and well-being;
- Evaluating rare disorder patients access to social and healthcare services and treatments and exploring the challenges surrounding coordination of care.

[1] ANSEA report, 2019

Providing the New Zealand Context

2019 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 2019 was developed[2]. 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, whilst the first to be conducted in NZ, was designed from similar robust patient voice surveys from European countries. This included the EURORDIS Rare Barometer Programme[3] which to date has over 10,000 survey responses and BURQOL-RD[4]. These surveys also improved the overall evidence base of people living with a rare disorder and enabled direct comparison of NZ and European-wide populations of people living with rare disorders. This has been reported on in the results section where possible.

Rare Disorders NZ engaged with over 130 support groups representing different Rare Disorders to participate and engage with the survey. All groups sent personalized e-mails to patients, stating the project objectives and providing a link to a web-based questionnaire. The data collection period ran from 18th November to 6th December 2019.

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

Results

Rare Disorder Diagnosis

Overall there were 288 responses with a confirmed diagnosis included in the primary analysis[2]**.** This represents **the largest ever survey of patient reported outcomes for people with rare disorders in NZ**.

The primary analysis reported on the overall population of people with a confirmed rare disorder diagnosis. There were 122 different diagnoses and a number of rare disorders with > 10 people diagnosed which included Ehlers Danlos Syndromes, Spinal Muscular Atrophy, Trigeminal Neuralgia, Charcot Marie Tooth Disease, Prader-Willi Syndrome and Myotonic Dystrophy

There were also 27 people excluded from primary analysis either because they were undiagnosed or where diagnosis was not confirmed. It is recognised that **early and accurate diagnosis of rare disorder remains important to accessing a range of treatments and healthcare services**.

[2] NZ Voice of Rare Disorders Survey.

[3] https://www.eurordis.org/rare-barometer-programme

[4] 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 2 Most common rare disease diagnoses in *Voice of Rare Disorders Survey*

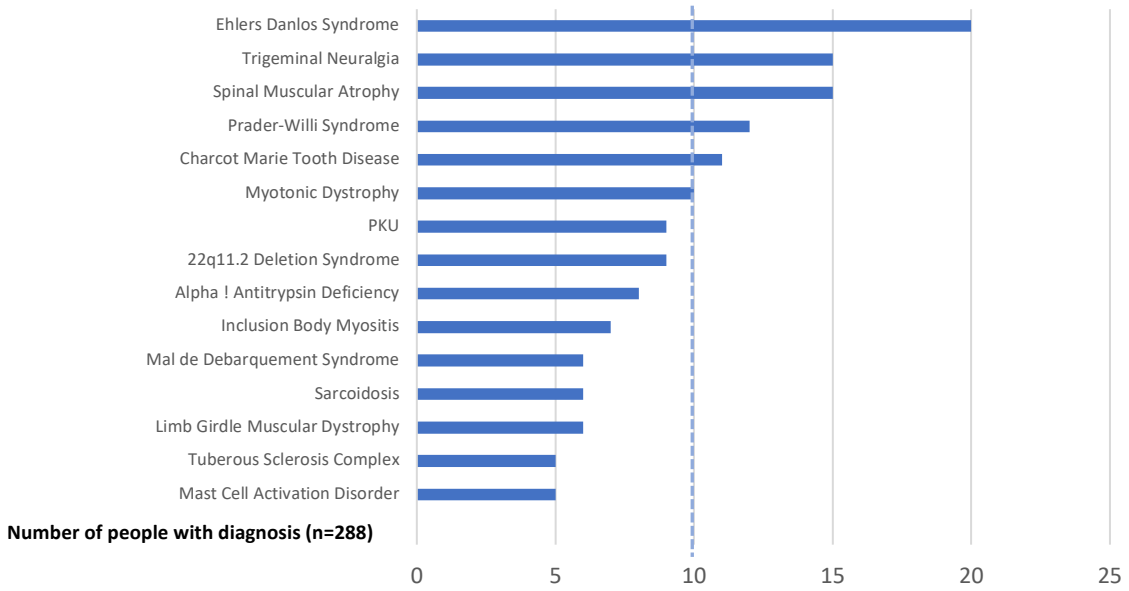
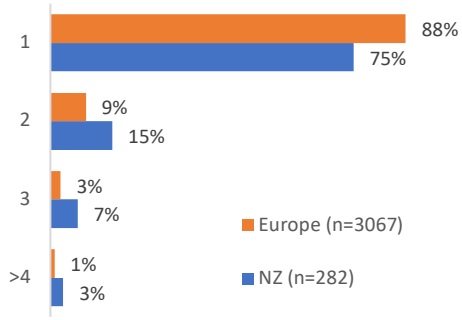


Figure 3 Comparison of number of people living with a rare disorder in European and New Zealand households



Results from the New Zealand surveys (see Fig. 3) highlighted that while the majority of households included one person with a rare disease, for 25% of cases there were 2 or more people. Comparatively this was much higher than in Europe where it is almost half of that number (13%). **In New Zealand, for one in ten households there were 3 or more people living with a rare disease..** 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[5]

The overwhelming majority of people that responded described themselves as being New Zealand European at 84%. By contrast the percentage of people that identified as being Maori or Pacific Peoples or Asian at 5%, 2% and 1% respectively, was lower than National Census estimates.

In line with population the majority of responses were from Auckland, Canterbury and Wellington regions. 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.

[5] 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)" <nzdotstat.stats.govt.nz>.

The majority of responses (63%) were provided by people living with a rare disorder, which was similar to Europe (56%) (Figure 4). Of these, **almost one in five had an additional role, either as a parent, sibling or spouse of another person living with a rare disorder.**

Overall 33% of the New Zealand responses were from parents of a child living with a rare disorder. Only 4% of response came from those who had additional roles including as spouse of person living with a rare disorder.

Figure 4 Relationship to person with rare disorder: Comparison of NZ and European survey responses

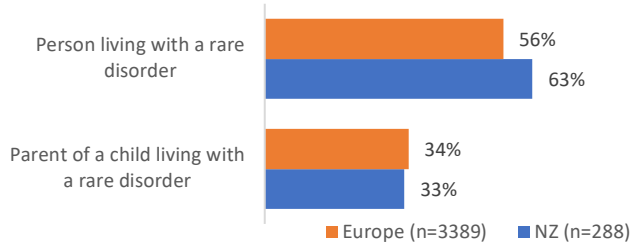
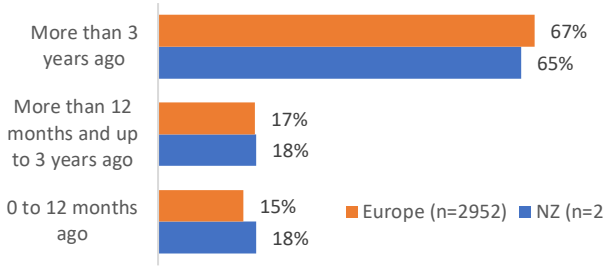


Figure 5 Time since diagnosis: Comparison of NZ and European survey responses

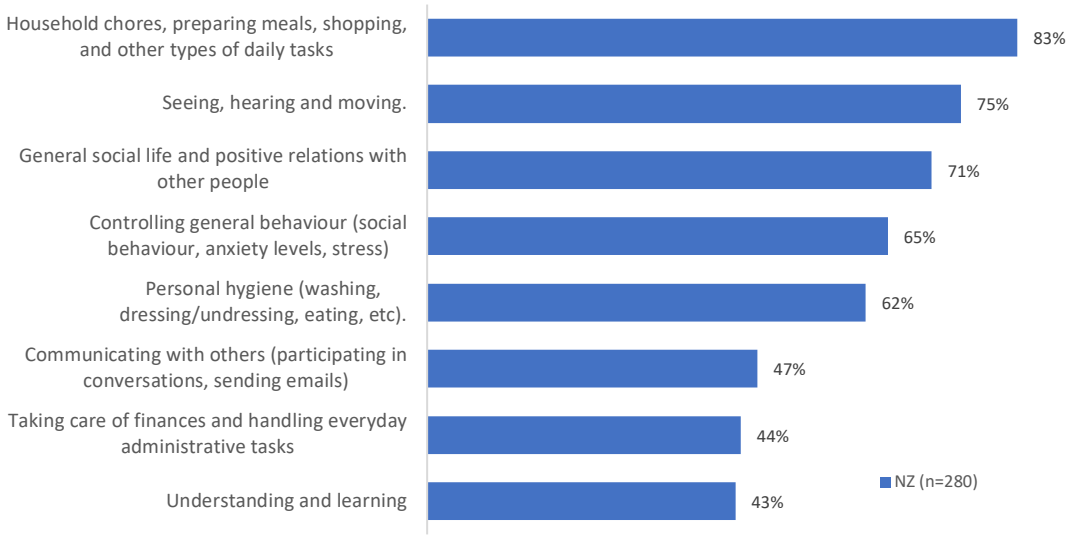


Overall over 80% of New Zealand people living with a rare disorder had received their diagnosis at least one year ago, with the majority of people (65%) having had a diagnosis from more than 3 years (see Figure 5). This is similar to Europe.

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 (e.g. capacity to carry out daily tasks, motor and sensorial functioning, personal care).

Figure 6 Activities of daily living that provide greatest difficulty



TOTAL % = some difficulty % + lot of difficulty %

Overall, almost 65% reported that the disorder impacted a lot on their health and everyday life, which was greater than for Europe.

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

Figure 7 Impact on health and everyday life: Comparison of NZ and European survey responses

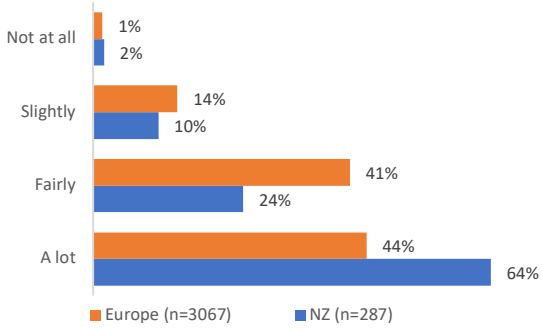
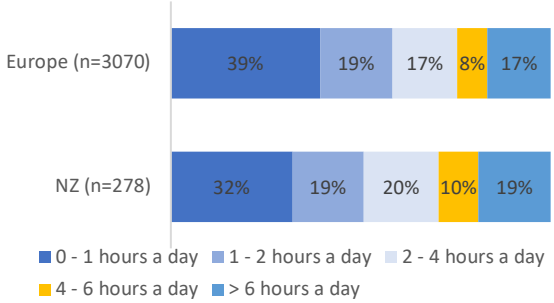


Figure 8 Daily time investment for illness-related tasks: Comparison of NZ and European survey responses



49% 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 40 minutes per day was invested in illness related tasks. This was 20 minutes longer than in Europe.

Healthcare services utilisation and cost estimates

Table 1 Healthcare service utilisation and cost estimates for people living with rare disorders

Service	Utilisation rate (%)	Total Annual frequency	Cost per test/visit/day[6]	Average cost per patient
Specialist visits	81%	6.4 visits	$250	$1,594
Diagnostic tests	75%	6.8 tests		
GP visits	88%	5.0 visits	$80	$400
Nurse visits	47%	2.6 visits	$40	$104
ED visits	38%	1.6 visits	$370	$560
Admissions	38%	1.1 admissions		
Inpatient days	36%	15.8 days	$1,200	$18,940
ICU Inpatient days	5%	7.9 days	$5,500	$43,410

[6] 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

There was high utilisation of healthcare services with **a large number of people having seen a specialist or GPs over the last 70 days (81-88% utilisation rate by respondents)**. There were also one in three people with a rare disorder that presented at the hospital in the last year. **They spent almost 16 days as an inpatient in the last year and, for one in twenty people, almost 8 days in ICU.**

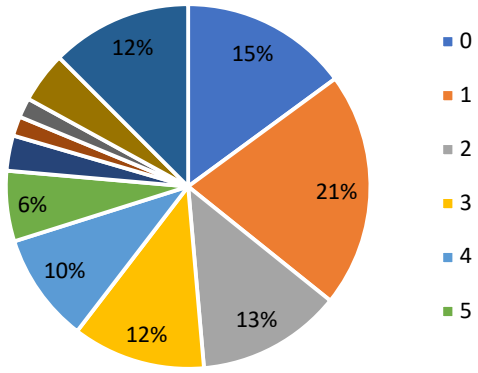
Medicines

The range of treatments people reported taking were limited and mainly focused on reducing inflammation.

Of people surveyed, 43 (15%) people were not taking any medicine related to their disease at all. An additional 131 (46%) people were only taking between 1-3 medicines, the majority of these for pain and inflammation. By contrast, there were 13 (5%) people taking at least 9 medicines related to their disease.

This is not surprising given references in the literature that 95% of rare disorders do not have effective treatments available

Figure 9 Number of medicines being taken by people



In total there were a total of 711 medicines being taken with the main types being for pain and inflammation. The most commonly reported were ibuprofen for 105 (36%) people, salbutamol for 48 (16%) people, prednisolone for 44 (15%) and melatonin for 38 (13%) people. There were a smaller number of people taking a broad range of different treatments including antibiotics, antipsychotics, anti-depressants vitamins (C, D, E).

Coordination of Care

The majority felt that organising care was difficult to manage. This included finding information on your disorder, understanding your rights and administrative procedures (66%) and finding the right professionals (58%). The time required to organise care varied from a little to a lot of time.

Over 60% felt that **communication and information exchange between different service providers was poor** (Figure 10).

While various healthcare professionals were a main first source of information, the **internet was also an important primary source.**

Figure 10 Quality of communication between service providers: Comparison of NZ and European survey responses

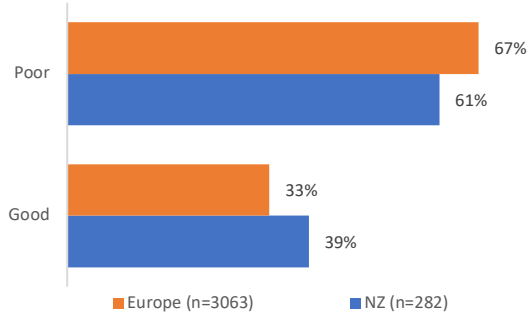
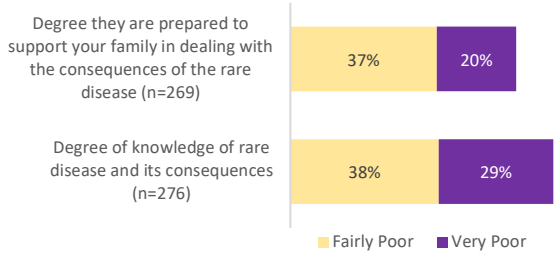


Figure 11 Level of support and knowledge from support services



A majority of people said that **professionals from social, local and support services are not well prepared to support their family**, nor do not know about the disorder and its consequences (Figure 11).

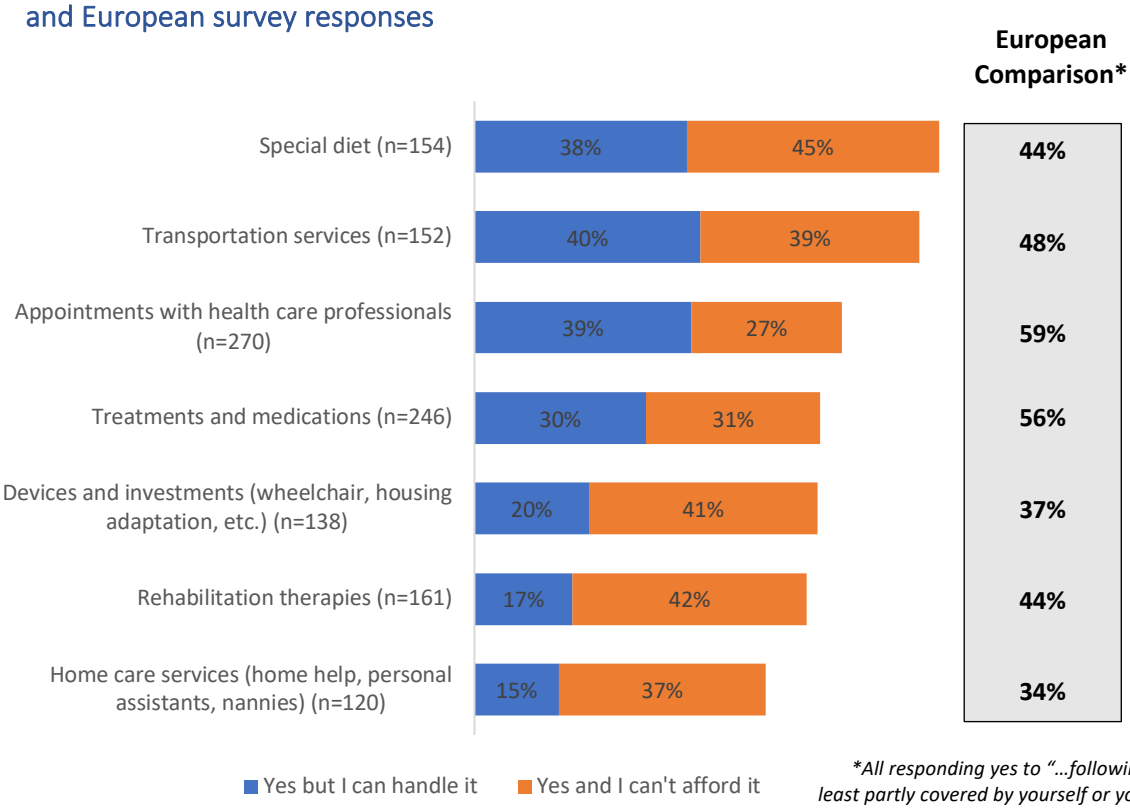
Overall, people did not feel informed at all around the rights related to the consequences of their disorder or financial help they could be entitled to.

Cost of living with a rare disorder

There were many costs covered by people living with the rare disorder or their family. Overall the percentage of people needing to privately cover at least some of their healthcare costs were higher in NZ than in Europe.

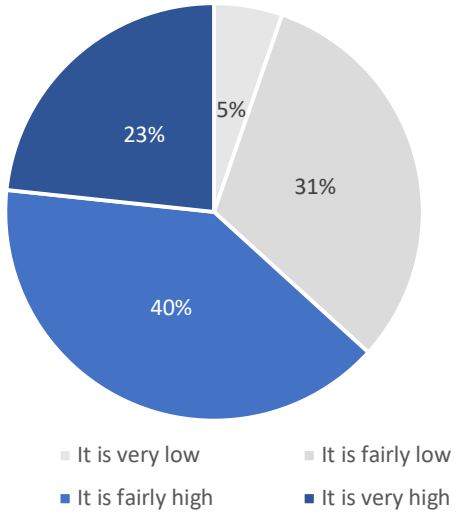
More than one in three people covered but could not afford home care, rehabilitation services, devices and investments including wheelchairs and housing adaptation.

Figure 12 Range and affordability of privately covered healthcare costs : Comparison of NZ and European survey responses



**All responding yes to "…following costs at least partly covered by yourself or your family"*

Figure 13 Perception of costs associated with the rare disorder

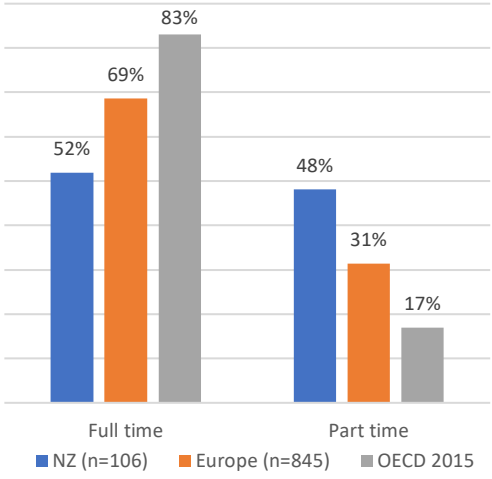


The majority of people felt that the costs associated with the rare disorder were high.

Overall 58% felt the costs associated with managing their rare disorder were hard to manage.

Employment

Figure 14 Impact on employment: Comparison of NZ and European survey responses



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

"I was well trained in a great job until I needed to be home to care for my son with SMA. I am unable to work because of his needs"

Overall 28% 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**, **limiting job opportunities** and reducing or stopping professional activity. It **caused a decrease in income for 80% people**.

While the majority of people were absent from work for less than 20 days due to the rare disorder, there were 15% of people who were absent from their work for more than 60 days.

Between 15-25% people did not have a work environment that was specifically adapted but needed it.

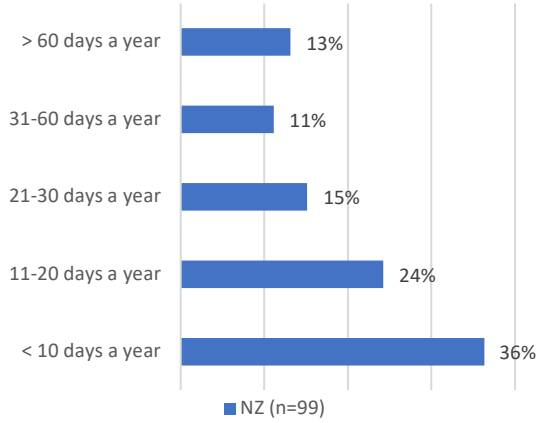
If the work environment was adapted to accommodate their rare disorder then the majority
would like their working hours and responsibilities to remain similar or increase.

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. The majority that required, adaptation and integration felt it was good.

Overall there was a range of days that children were absent from school in a year with **24% children being cared for absent for more that 30 days per year.**

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. Overall the largest response (35%) was for those people that don't qualify but would benefit
- There were generally more hours received for support for house chores and daily tasks with 18 people receiving more than 12 hours. Again the largest category (44%) were for those that don't qualify but would benefit
- 45% people had some respite care in the last 12 months with a range of care. A further 20% people felt they could benefit but did not qualify for respite care

Family life, stress and wellbeing

Since the symptoms started the **majority of people have experienced increased tension between family members (55%) and isolation from family and friends (68%)**

Overall around **one in three people often felt unhappy and depressed and felt they could not overcome their problems** (Figure 16, Figure 17). In addition, almost another one in three people sometimes had these same feelings. These issues were much higher than in general populations (based on International Social Survey Programme, 2011)

Figure 16 Extent that person felt unhappy and/or depressed in last month: Comparison of NZ and European survey responses

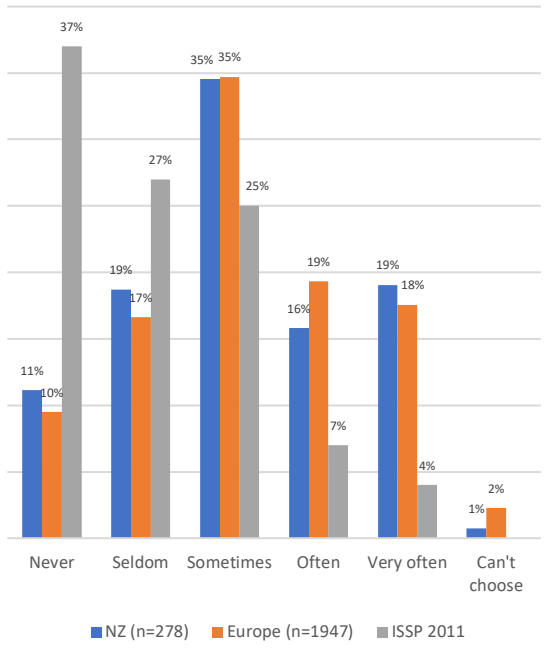
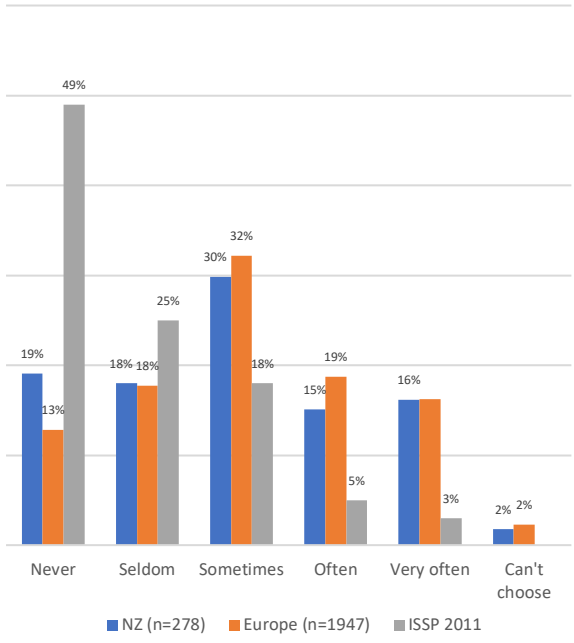


Figure 17 Extent that person felt they could not overcome their problems in last month: Comparison of NZ and European survey responses



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. When measured against a European reference population living with rare disorders[7] the impact was similar to or worse for people in NZ. It paints a picture of isolation, poor treatment access, lack of coordinated care and for some, being lost in the system.

Given this, we can look to other countries to provide insight and direction, strategic options and approaches that can be taken in reducing the impact for people living with a rare disorder living in NZ. A number of these have been considered here.

1. Common definition of rare disorder in NZ

At present New Zealand does not have an official definition of rare disorders, whereas in Europe a rare disorder (disease) is defined as a health condition that affects 1 in 2000 people or less. If we were to use this definition, there are likely to be more then 7,000 rare disorders affecting around 300,000 New Zealanders.[8]

There is significant heterogeneity in rare disorder prevalence that needs to be considered in policy development. Taking NZ's population, rare disorder prevalence can vary from as high as 2,500 people down to as little as one individual being affected.[9] Table 2 highlights the different prevalence intervals (excluding rare cancers and infectious diseases), showing that while the majority of the prevalent population (96%) are accounted for by approximately 400 rare disorders, more than 3,000 rare disorders have less than 50 people affected.

Table 2 Prevalent rare disorder population in NZ

Rare Disorder Prevalence intervals	NZ Prevalent population	Number of rare disorders[9]	Estimated number of people[10]
1-5/10,000	500-2500	149 (4.2%)	223,000 (74%)
1-9/100,000	50-499	241 (6.7%)	66,000 (22%)
1-9/1000,000	5-49	164 (4.6%)	4,400 (1.5%)
<1/1,000,000	<5	3,031 (84.5%)	7,500 (2.5%)

Inability to establish a national definition of rare disorder is likely to reduce the motivation to develop policies that lead to meaningful improvements in health outcomes. It can also result in inconsistencies and frustration across the health system. For example, with NZ funding processes and policy settings for rare disorder medicines, PHARMAC define 'rare' based on 1 per 50,000 people, or those disorders with an identifiable and measurable patient population of less than 90 patients.[11] In Australia, England and Scotland this is an ultra-rare definition. This rare definition has an impact by limiting medicines available under this policy to benefit, at most, one in ten people with rare disorders (Table 2).

[7] Juggling care and daily life The balancing act of the rare disease community. A Rare Barometer survey. May 2017

[8] https://www.raredisease.org.uk/what-is-a-rare-disease/

[9] 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

[10] https://www.stats.govt.nz, Population clock estimate December 2019

[11] 2019-Report-Funding-Medicines-for-Rare-Disorders

By contrast, where countries have consistently defined rare disorders, it has enabled policy development and planning. As an example, the recently announced collaboration agreement between Rare Diseases International (RDI) and World Health Organization will initially focus on harmonising rare disorder definitions internationally, enabling development of a global network of centres of excellence for rare disorders.[12]

2. Wider community engagement, in particular to include Māori, Pacific and ethnic minorities

Small numbers of people living with various rare disorders creates many challenges. This includes engaging with the widest range of communities to understand the impact of living with their specific rare disorder, as well as providing a truly collective voice to advocate for better and more equitable access to healthcare services and treatments. As an example, this survey provided an important voice for a number of people in the NZ rare disorder community which can be repeated over time to measure improvements in health outcomes.

It is also critical to prioritise populations to reduce potential inequities in the health system on the basis of ethnicity, rurality or socioeconomic status. The current state was highlighted in the survey findings given a disproportionately low engagement with people that described themselves as Maori, Pacific or Asian. While we could not conclude on rurality and socioeconomic status, these groups are typically associated with poorer health outcomes. This suggests an opportunity to test new ways to increase engagement with these populations, using culturally appropriate methods so all people are included and connected. This is essential to leave no one behind, part of the wider UN WHO 'right to the highest attainable standard of health' and universal health coverage (UHC).[13]

Wider engagement with key stakeholders beyond people living with rare disorder and their advocates are to reinforce a person-centred focus, including with key peak bodies, governments, researchers, clinicians and industry to promote rare disorder, diagnosis, access to services and treatments, data collection and coordinated care.

3. Develop person centred co-ordinated models of care for people living with a rare disorder

People living with rare disorder often have complex needs and face unique challenges and barriers of access to care, treatment, education, employment, mental health. Clinicians also face the challenge of limited awareness, information or connections to specialists with knowledge on rare disorders. These multiple system issues and less understood complexities mean a planned approach to healthcare management and social support is required to achieve equitable outcomes through new models of care.

This approach must have effective person centred care which acknowledges their needs, has room to hear their voice and involves them in the structure of mechanisms for healthcare and social inclusion. 'Nothing about us without us'. The best people to direct solution-focused care frameworks must be those that have walked the journey and faced the barriers and challenges.

Developing new models of care requires a co-ordinated 'holistic' approach, enabling inter-disciplinary services and effective collaboration between all health professionals, community service providers, Ministry of Social Development and Ministry of Health. People living with a rare disorder should be supported by a healthcare system that leaves no one behind. It should remove structural barriers that prevent cohesive healthcare, providing navigation for rare and complex cases, access to the right treatment, access to appropriate specialists and co-operation, no matter where you live in the country.

[12] https://www.rarediseasesinternational.org/rdi-signs-memorandum-of-understanding-with-the-world-health-organization/

[13] https://www.who.int/news-room/fact-sheets/detail/human-rights-and-health

Models of care should also reflect the wider International perspective, especially with the emergence of centres of excellence. Given this access to centres of excellence for specific 'grouped' disorders (metabolic, skeletal, neurological, etc.) which link to international centres should be considered.

4. Rare Disorder medicine evaluation to consider wider societal impact

Access to medicines for rare disorders need to take into consideration their small patient populations, progressive disease and limited data. Just some of the challenges for the development of innovative medicines include the quantity and quality of clinical evidence given size of patient populations, the number of rare disorders that disproportionately affect children, or lack of data to demonstrate the wider societal impact due to rare disorders that could be mitigated through effective treatments.

In NZ, PHARMAC requires ultra-rare disorder medicines substantially improve patients absolute or relative age specific life expectancy and/or quality of life as a direct consequence of the treatment.[11] However collection of clinical data including quality of life information required for these Health Technology Assessments (HTAs) are often challenging, especially for children that are over-represented in rare disorders. It is also important in the context of rare disorders that the wider impact of the family, whānau and wider society is a factor for consideration when evaluating these medicines.

The UK has created a more appropriate and flexible framework for bringing the treatment for rare disorders to patients. In Scotland and UK, NICE defines an incremental cost- effectiveness ratio (ICER) threshold of £30,000 per quality-adjusted life year (QALY) gained for medicines. In April 2017, NICE published an increased threshold of ICER of £100,000 per QALY gained for drugs for "very rare" disorders.[1]

In Australia, the Government currently funds 14 different life-saving medicines for nine very rare disorders through the Life Saving Drugs Programme (LSDP). Medicines funded through this program include high cost medicines that do not meet criteria to be funded on the Pharmaceutical Benefits Scheme (PBS). In 2017-18 they invested $128 million with the program benefiting 400 Australian patients.[14] This is in addition to a range of effective treatments for rare disorders funded through the Highly Specialised Drugs Programme (Section 100) of the PBS.

Given this, new approaches are recommended in NZ to support economic evaluation of medicines for rare disorders including: bespoke data collection, simple economic modelling methods that reflect evidence available at the time of the assessment; and identifying what is 'good enough' to inform decision making. [15] A method that not only considers direct medical and medicine costs but the wider costs for family, whānau and society in its consideration.

5. Develop NZ National Rare Disorder Framework

Effective policy development requires acknowledgement and awareness of the common challenges faced by people living with a rare disorder, along with a commitment to address these challenges through a New Zealand Rare Disorder Framework and Action Plan. This would ensure people living with rare disorders and clinicians play a central role in decision-making processes, reduce inequities, consider a wider definition of a persons' quality of life to also the impact on carers. The plan would be comprehensive with no part excluded and aligned with other current healthcare implementation plans.

[14] https://www.health.gov.au/ministers/the-hon-greg-hunt-mp/media/medicine-for-rare-disease-made-free-on-the-life-saving-drugs-program

[15] Sampson, C., and Garau, M., 2019. How Should We Measure Quality of Life Impact in Rare Disease? Recent Learnings in Spinal Muscular Atrophy. OHE Briefing, London: Office of Health Economics. Available at: https://www.ohe.org/publications/howshould-we-measure-quality-life-impact-rare-disease-recent-learnings-spinal-muscular

We can take inspiration from the experience of the UK, that published its first national plan titled "UK Strategy for Rare Diseases" in 2013. The plan presented a high-level framework which contained 51 commitments, with a set vision by 2020 to improve lives of patients living with rare disorders. Main areas covered were empowering people affected by rare disorders; identifying and preventing rare disorders; diagnosis and early intervention; coordination of care and the role of research.[1] This led to the publication of country level plans, including Scotland with "It's Rare not to have a Rare Disease".

Table 3 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 now been echoed by the newly formulated Australian National Strategic Action Plan for Rare Diseases, drafted in 2019 following extensive stakeholder consultation. This was seen as a critical step towards a National Rare Disease Framework and effective policy with pillars focused on Awareness and Education; Care and Support; and Research.[16]

New Zealand lags far behind these countries in developing policy that leads to improvements in diagnosis, access to medicines and healthcare services. A NZ Rare Disorders Action Plan should acknowledge barriers for thousands of vulnerable children and adults and reduce inequities that exist. It should promote a co-ordinated 'holistic' approach to care that includes access to health and disability services based on clinical need, adequate respite care and carer support, workforce training – including genetic counsellors, genetic biostatistician, trained specialists, allied health professionals and lab workers.

It is important that any Rare Disorders Action Plan align with and ideally inform other related healthcare implementation plans. This could be the case following the recently launched Carers' Strategy Action Plan which now includes recognition of carer contributions, carer navigation of support and services, wellbeing support and balance of role with paid work or study. It emphasises target populations (Māori, Pacific, young and older carers) with a family, whānau, aiga-centred implementation approach.[17] However while there is not a plan in place the many voices of people with rare disorders will not be heard.

[16] https://www.rarevoices.org.au

[17] https://www.msd.govt.nz/about-msd-and-our-work/newsroom/2019/new-carers-strategy-action-plan-launched.html

Final Remarks

"No country can claim to have achieved universal healthcare if it has not adequately and equitably met the needs of those with rare diseases." Helen Clark, UN Development Programme, 2016[18]

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.

In New Zealand a shift in mindset is needed for rare disorders to be regarded as a significant unmet need within health policy frameworks. The reality is that rare disorders are not viewed as a collective, unlike those common illnesses such as cancer, but considered in isolation. The irony is the fact that 'Rare is many', with up to 300,000 people living with a rare disorder in NZ and with a collective impact for them, their family, whānau and society that is significant.

This has highlighted the importance of a number of changes that are 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:** Develop an inclusive **consistent definition for rare disorder**. This is essential in order to enable policymakers to develop a national plan for rare disorders.
- **Recommendation 2:** Build a collective voice for people living with rare disorders through **wider community engagement**, in particular to include Māori, Pacific and ethnic minorities
- **Recommendation 3:** Greater collaboration amongst different stakeholders to **implement patient centric co-ordinated models of care** for better quality life for people with rare disorders and mitigate obstacles faced by these patients.
- **Recommendation 4:** Ensure funding mechanisms for medicines for rare disorders consider an economic evaluation method that not only considers direct medical costs and medicine costs but other societal costs.
- **Recommendation 5:** Develop a **NZ Rare Disorders Framework and Action Plan,** aligned with other current healthcare implementation plans.

[18] Statement from UN Development Programme Administrator Helen Clark to the International Conference on Rare Diseases & Orphan Drugs, Cape Town, 20 October 2016.