Making sense of the numbers

Rare Disorders Insights Report

Pathways towards better health outcomes

March 2023

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©BERL Reference No: #6291 March 2023

Executive summary

Rare disorders are often defined as disorders that affect between one in 2,000 people, or one in 3,000 people. While each individual rare disorder is uncommon, in total they are estimated to affect 300 million people worldwide.[1] Rare disorder patients represent a large group in any country's health system, they face more barriers, and suffer significantly more from their conditions than other patients. Therefore, this group needs special attention.

Business and Economic Research Limited (BERL) was tasked by Rare Disorders New Zealand (RDNZ) to develop an insights report on rare disorders in New Zealand. This report is based on publicly available data and a high-level literature scan of recent New Zealand and international studies that discuss the prevalence of rare disorders, as well as burden factors that need to be considered in the development of health policies.

After conducting our research, we concluded that the quantity of data on rare disorders in general in New Zealand is strikingly low. Thus, it is not possible to accurately estimate the prevalence of rare disorders in the country.

Awareness and training

Our high-level literature scan revealed a range of international studies that aimed to estimate the level of awareness of and training regarding rare disorders among health professionals and students.[2] Only one study from New Zealand mentioned the level of knowledge on rare disorders among health professionals, but it was not comprehensive.

The common theme among all studies was that health professionals and students alike demonstrated a very poor level of rare disorders awareness and knowledge. This is concerning as from diagnosis to treatment, awareness and effective training are pre-requisites for a desired health outcome.

Our recommendations include advocacy for the Government health agencies, and education providers, to take further action to include rare disorders in campaigns, activities, and curricula.[3]

Research and data

The lack of data regarding rare disorders in New Zealand is not only a challenge to estimating prevalence, but also for improving diagnosis, treatment, and research. Through our research, we have identified that the classification system for diseases that New Zealand uses does not include most rare disorders. Without data we cannot know the size of the problem.

Since New Zealand is a small country, it depends heavily on international partnerships for research and by not using the internationally recognised system of classification of rare disorders, Orphanet, partnerships are limited.[4]

[1] https://www.rarediseasesinternational.org/wp-content/uploads/2022/01/Final-UN-Text-UN-Resolution-on-Persons-Living-with-a-Rare-Disease-and-their-Families.pdf
[2] "Health professionals and students" is a broad definition referring to all professionals involved in healthcare, including medical, health, and support professionals, and students that are enrolled in courses related to these professions. For the sake of consistency, this term is used throughout the report, substituting for a range of similar terms used in referenced studies.
[3] All mentions of "Government health agencies" refer to the tripartite system of the Ministry of Health, Te Whatu Ora – Health New Zealand, and Te Aka Whai Ora – Māori Health Authority.
[4] https://www.orpha.net/consor/cgi-bin/index.php?lng=EN

Diagnosis and treatment

Our research showed that New Zealand may be performing worse than its peers in diagnosis speed and accuracy. Patients had to visit more doctors to get a diagnosis, and almost two thirds received a wrong diagnosis at least once. This also points towards the need to review the country's National Screening Unit, as is being done in the UK.

Pharmac's own definition of rare disorders, at a prevalence of less than one person in 50,000, means that there is a rare disorder medicines funding gap. In 2019, only 348 rare disorder patients had their specialised medicines funded by Pharmac, while it is estimated that there are from approximately 2,000 to 15,000 people living with a treatable rare disorder in this country.[5]

Another barrier that rare disorder patients face in accessing care in New Zealand is the lack of coordination in the health system. The previous District Health Boards (DHBs) were too fragmented, and while there are some measures in place to promote the sharing of information, they were also too sparse. However, there is a great opportunity for integration and coordination of care with the new country-wide single DHB (Health NZ) introduced in July 2022.[6]

The costs of a failed system

From the literature scan, it is evident that rare disorders incur heavy costs to the people living with them, their families, and health systems. This supports the urgency and need to take action to improve the lives of those affected by rare disorders:

- In New Zealand:
  - There is high utilisation of healthcare services
  - Between 81 to 87 percent of people had seen a specialist or General Practitioner (GP) in the last 70 days
  - Almost one third of respondents (30 percent) were having to undertake self-funding for at least some of their medicines
  - The vast majority of people (70 percent) saw a reduction in income related to a rare disorder
- In Canada:
  - Almost 90 percent have had family finances negatively impacted
  - Almost 80 percent suffer from mental distress
- In the United States:
  - An average rare disorder patient has an annual medical cost that is US$26,887 more than an average patient
  - A lack of treatment for a rare disorder is associated with a 21 percent increase in total costs on a per patient per year basis

[5] https://pharmacreview.health.govt.nz/assets/Uploads/final-report/Pharmac-Review-Final-Report.pdf
[6] https://www.futureofhealth.govt.nz/health-nz/

- In Australia:
    - Rare disorder patients represented two percent of the Western Australia (WA) population, but they represented 4.6 percent of the total number of patients discharged from hospital, and 9.9 percent of all discharges
    - The total cost associated with hospital discharges for rare disorder patients was 10.5 percent of the total WA inpatient hospital expenditure.

Recommendations

We have divided our recommendations into three key areas, and used national frameworks from Australia and the United Kingdom to inform these areas. We have shaped our recommendations to be either part of a national framework or implemented independently. Apart from focusing on the burden factors that need to be considered in the development of health policies, our recommendations also identify the tools and data necessary to estimate the prevalence of rare disorders in New Zealand more accurately.

Awareness and training

- Advocate for the Government health agencies to fund a study with health and support professionals to gather evidence of their level of awareness and knowledge of rare disorders
- Partner with the University of Otago and the University of Auckland to conduct a similar study with healthcare students
- Advocate for the Government health agencies to fund awareness-raising campaigns and events in New Zealand
- Advocate for the University of Auckland and the University of Otago to further include rare disorders in their curricula
- Advocate with The Royal New Zealand College of General Practitioners and The Royal Australasian College of Physicians, for the Medical Council of New Zealand, and the Australian Medical Council, to further include rare disorders in the Continuing Professional Development (CPD) curricula
- Advocate for the Government health agencies to implement digital resources that support health professionals reach a diagnosis and a treatment plan.

Research and data

- Advocate for the Government health agencies to develop a rare disorder patient registry, which uses the Orphanet classification.

Diagnosis and treatment

- Advocate for the Government health agencies to conduct a comprehensive review of the National Screening Unit
- Advocate for the Government health agencies to further develop rare disorder guidelines
- Advocate with the Genetic Health Service NZ for the Government health agencies to allocate additional funding for genetic testing

- Consider the possibility of further advocacy in regards to genome sequencing in newborns, depending on the results from England in regards to the efficiency of genome sequencing in newborns
- Advocate for the Government health agencies to adopt an official definition of rare disorders that is in line with international best practice
- Advocate for Pharmac to expand its rare disorders medicines contestable funding pilot, and to admit that under the current model a significant proportion of people living with a treatable rare disorder are not being covered
- Advocate for the Government health agencies to take advantage of the health system unification reform and implement a fully integrated country-wide information sharing system
- Advocate for the Government health agencies to develop an expert centre for rare disorders in the country.

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1 Introduction

Business and Economic Research Limited (BERL) was commissioned by Rare Disorders New Zealand (RDNZ) to develop an insights report on rare disorders in New Zealand. This report is based on publicly available data and a high-level literature scan of recent New Zealand and international studies that discuss the prevalence of rare disorders and burden factors that need to be considered in the development of health policies. The recommendations were also shaped by national frameworks from Australia and the United Kingdom (UK).

In New Zealand there is no official definition of what constitutes a rare disorder. International definitions vary, but most definitions consider rare disorders to be disorders that affect between one in 2,000 people, or one in 3,000 people. There are over 7,000 rare disorders, which mostly affect children, and many of them are genetic and inherited.

In many referenced documents in this report, the term “rare disease” is used to refer to “rare disorder”. In this report we use the term “rare disorder”, but in this context these terms are interchangeable.

1.1 Methodology

In order to develop the insights report we:

- Conducted a high-level literature scan on national and international studies about the prevalence of rare disorders
- Conducted a high-level literature scan on national and international studies about the challenges and costs faced by people living with rare disorders, their families, and health systems
- Reviewed national and international rare disorders policy documents, including the national frameworks from Australia and the UK, and Pharmac’s funding models.

From this research, we made recommendations based on the available data and literature. The national frameworks from Australia and the UK also influenced some of our recommendations, where viable. Based on our research, we observed that rare disorders health policies can be divided into three areas. These areas shaped our report:

- Awareness and training
- Research and data
- Diagnosis and treatment.

Data limitations and assumptions

The main limitation of our research is the lack of data and studies specific to the New Zealand context. Furthermore, even when looking internationally, there are only a limited number of studies and data sources around rare disorders.

All the studies paint the same picture: rare disorders have a disproportionate impact on the affected people and the health system as a whole. While limited, studies and data from New Zealand show that the country performs worse than its peers in providing health care for those living with rare disorders. It is also likely that New Zealand suffers from similar problems as the other countries studied.

2 Awareness and training

In this section we explore the scope of awareness and training under rare disorders health policy. We discuss the level of awareness and training by analysing publicly available data from New Zealand and internationally, and explore examples of best practice. Further, we outline the rare disorders awareness and training challenges specific to New Zealand's context.

Awareness and training of health professionals is essential to improving the lives of those living with rare disorders. From diagnosis to treatment, the level of awareness of rare disorders a health professional has is crucial to desired health outcomes. A lack of awareness of rare disorders often contributes to people feeling isolated and misunderstood, as well as to delays in diagnosis and treatment, potentially missing opportunities for early intervention and improved outcomes.[7]

Hence, improving awareness and knowledge relevant to rare disorders in New Zealand through effective training is essential for better health outcomes. Also, by increasing awareness and knowledge, we will get closer to having the necessary data needed to estimate prevalence in the country.

2.1 Existing data

Awareness of rare disorders and subsequent training are essential determinants of a precise diagnosis followed by an effective treatment. There is currently only one publicly available study that includes the level of rare disorders awareness and knowledge among health professionals in New Zealand.[8] Moreover, a series of studies internationally suggest that there is an underwhelming level of awareness around rare disorders amongst health students and professionals globally, which adds to the findings of the study in New Zealand.

New Zealand

The study in New Zealand concluded that health professionals are not well prepared to support the families of those living with a rare disorder, nor are they informed about the disorder and its consequences.[8] The study was based on a survey in which people living with rare disorders, or their carers, answered a series of questions related to their experiences.

Over half (52 percent) of study participants stated that health professionals were poorly prepared to support their families with the consequences of living with a rare disorder. The majority (66 percent) of participants also said that health professionals have a poor degree of knowledge about rare disorders.

Spain

A study conducted in Spain with people with rare disorders and carers found that 79 percent of the participants said they were inappropriately treated by health professionals because of their rare disorder.[9] Over half (56 percent) of those said that the main cause of the poor treatment was a lack of knowledge about their rare disorder. Only 19 percent stated that they had never received inappropriate treatment by health professionals.

[7] https://ojrd.biomedcentral.com/articles/10.1186/s13023-017-0622-4
[8] https://www.raredisorders.org.nz/assets/VOICE-OF-RARE-DISORDERS-White-Paper-February-2021-FINAL.pdf
[9] https://sid-inico.usal.es/idocs/F8/FDO24871/necesidades-sociosanitarias.pdf

Australia

Similarly, a study conducted in Australia with carers of people with rare disorders found that the participants' perceived reasons for diagnostic delays included lack of knowledge about their condition among health professionals (69 percent), and lack of symptom awareness by the family (21 percent).[7]

The findings from a similar study in Australia had comparable results.[10] Over half (54 percent) of study participants reported dissatisfaction with health professionals' level of knowledge and awareness of rare disorders. This study also included a few participants from New Zealand. However, it was not considered a New Zealand study due to the low number of participants from the country (*n*=16) in relation to the total (*n*=301).

The following studies from Poland, China, and Kazakhstan took a different approach and surveyed health professionals and students instead of patients and carers.

Poland

A study conducted in Poland with 165 health professionals who were taking specialisation courses, found that most of these health professionals lacked basic knowledge about the causes and prevalence of rare disorders.[11] The overwhelming majority of participants (95 percent) rated their knowledge of rare disorders as "insufficient" or "very poor". Only five percent of participants stated that they felt prepared for caring for a patient with a rare disorder.

China

Researchers in China performed a study with 224 health professionals, which included a self-assessment survey.[12] Only 5.3 percent of participants said they were moderately or well aware of rare disorders. Most participants (59 percent) thought their hospitals had not paid enough attention to patients with rare disorders. Moreover, this study included interviews with nine rare disorders experts. All of those experts agreed that health professionals generally lacked the rare disease awareness, which was a major reason of misdiagnosis.

Kazakhstan

A study done in Kazakhstan with 207 health students and 101 health professionals had an interesting methodology.[13] Apart from a self-assessment survey, participants had to show their knowledge of rare disorders in a test. Among students, 12 percent rated their knowledge of rare disorders as "very good" or "fair enough", and 27 percent felt prepared for caring for a patient with a rare disorder. Among professionals, 10 percent rated their knowledge of rare disorders as "very good" or "fair enough", and 20 percent felt prepared for caring for a patient with a rare disorder.

There were seven questions in the rare disorders knowledge test. They included the prevalence rate of rare disorders, their causes, the number of patients, and the most affected age groups. Participants performed poorly; in six of the questions, the vast majority gave a wrong answer. The rare disorders knowledge test done in the Kazakhstan study confirmed the conclusion from the self-assessment survey: both students and professionals had an underwhelming level of rare disorders awareness and knowledge. The studies mentioned in this section all paint the same picture.

[10] https://bmcprimcare.biomedcentral.com/articles/10.1186/s12875-016-0488-x
[11] https://ojrd.biomedcentral.com/articles/10.1186/s13023-021-02023-9/tables/4
[12] https://ojrd.biomedcentral.com/articles/10.1186/s13023-021-01788-3
[13] https://www.frontiersin.org/articles/10.3389/fpubh.2022.872648/full

The level of rare disorders awareness and knowledge is poor in every country in which they were conducted.

Studies that collected information from people with rare disorders and their carers, and studies that collected information from health students and professionals, both reached the same conclusion. There is a lack of rare disorders awareness and training across a wide selection of countries.

2.2 Actions to improve awareness and training

In this section we explore what other countries have done to improve awareness and training, especially in the UK and Australia. Both of those countries have developed national strategies for rare disorders, in which they outline actions aimed at improving awareness and training.

2.2.1 Education for health professionals

Australia

Australia has indicated, through its National Strategic Action Plan for Rare Diseases, the importance of improving awareness and training.[14] One of the actions outlined in the plan is to develop a national rare disorders workforce strategy. This action will focus on identifying gaps in the workforce, and ensuring collaboration and consultation between education providers, professional bodies, and other key stakeholders. The goal is to ensure that the health system can maintain the supply of capable workers who support patients with rare disorders.

Another action stated is to equip and encourage health professionals to consider, investigate, and refer for a potential rare disorder diagnosis. This action consists of the development and promotion of guidelines to provide support for clinicians in identifying possible rare disorders in patients with complex symptoms. Another element of the presented action is the promotion of the use of the digital repository by health professionals. This will support health professionals to identify rare disorders by providing them with information, data, and images of specific rare disorders.

England

The English rare disorders action plan includes actions to develop an innovative digital education resource, and to include rare disorders in health professional education and training frameworks.[15] For instance, Health Education England (HEE) is developing GeNotes, a digital educational resource, to help health professionals make the right genomics decisions by providing clinical information to support patient care. The educational content in GeNotes can be integrated into other digital platforms that health professionals already use, providing them with easily accessible information on rare disorders.

England's action plan also encompasses an empowering action to improve the curricula of health professional education and training frameworks. HEE will work with people living with rare disorders, professional organisations, and curriculum developers to determine how to better include rare disorders in those training frameworks.

[14] https://www.health.gov.au/sites/default/files/documents/2020/03/national-strategic-action-plan-for-rare-diseases.pdf

[15] https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/1057534/England-Rare-Diseases-Action-Plan-2022.pdf

Aside from being part of the actions outlined in the plan, HEE has already been promoting awareness and knowledge of rare disorders among NHS staff with its Genomics Education Programme. The programme offers a range of taught and online courses on genomics, including rare disorders.

2.2.2 Awareness in the wider society

Australia

As part of the national strategy, the Department of Health intends to develop and maintain an accessible digital repository to detail available care and support, and to provide general rare disorder information.[14] The repository will be promoted to rare disorder organisations, for the distribution to people with rare disorders and their families.

Another interesting action is the promotion of targeted awareness to support people in their preparation for conception. This includes information on preventive measures, and rare disorders testing and screening. In general, the government aims to promote awareness in wider society by funding national media campaigns.

Northern Ireland

Actions to increase overall rare disorder awareness are included in the Northern Ireland action plan.[16] The Department of Health intends to deliver a range of webinars, information sessions and outreach support meetings. They have also announced the hosting of at least two rare disease focused science, technology, engineering and mathematics initiatives in 2022. The Department of Health has also stated its intention to participate in bi-monthly all-Ireland rare disease webinars, and plan for quarterly rare disease discovery research meetings with both professional and public components.

2.3 Recommendations for improving awareness and training

As discussed, there is only one study in which the level of rare disorder awareness and knowledge among health professionals in New Zealand is measured. Also, that study is only based on the experiences of people with rare disorders and their carers.

We recommend Rare Disorders NZ (RDNZ) advocate for the Government health agencies to fund a study on health professionals in order to better understand their level of awareness and knowledge of rare disorders.[3] Such a study should include a self-assessment questionnaire and a knowledge test, and these would be compared to reach a more trustworthy conclusion. We also recommend RDNZ to seek a partnership with the University of Otago and the University of Auckland, with the goal of conducting a similar survey with health students.

The main action from the Government health agencies to promote awareness of rare disorders in the country is providing funds to RDNZ. RDNZ is the main actor in promoting rare disorders knowledge and awareness in the wider society. More could be done by the Ministry around improving awareness and training for health professionals.

We recommend RDNZ also advocate for the Government health agencies to fund awareness-raising campaigns and events in New Zealand. One of these events could be a rare disorders case competition for tertiary students. A case competition is where students form small teams that compete to solve challenging problems.[17]

[16] https://www.health-ni.gov.uk/sites/default/files/publications/health/doh-ni-rare-diseases-action-plan-2223.pdf
[17] https://managementconsulted.com/case-competition/

Teams focus on an overarching research question and are tasked with developing a solution that they present to a panel of judges, who evaluate the recommendations. The main objective of having a case competition is to get students thinking about the theme of the competition, being aware that this is a problem, and sparking their interest in further studying about rare disorders. It is an awareness-raising action that effectively targets future health professionals, without huge financial resources.

New Zealand should also bring its training in rare disorders for health students and professionals up to international standards. In countries with similar health outcomes for people with rare disorder patients to New Zealand, new policies have been introduced to improve training and improve outcomes. We recommend RDNZ advocates for the University of Auckland and the University of Otago to further include rare disorders in their curricula.

Additionally, improving Continuing Professional Development (CPD) for doctors is needed. Apart from generally needing to improve training for New Zealand trained doctors, over 40 percent of doctors registered in New Zealand completed their qualifications overseas.[18] Therefore, we recommend RDNZ advocates with The Royal New Zealand College of General Practitioners, The Royal Australasian College of Physicians, the Medical Council of New Zealand, and the Australian Medical Council, to further include rare disorders in the CPD curricula. Furthermore, this could be achieved by following England's steps and creating a working group with patient organisations, curricula developers, and professional organisations to discuss how to better approach rare disorders content in health education.

Lastly, we recommend that RDNZ advocates for the Government health agencies to implement digital resources that support health professionals to reach a diagnosis and a treatment plan. These digital resources could include a digital repository with varied and relevant information about rare disorders, based on existing resources from institutions like Orphanet, NORD, and GARD.[4] [19] [20] Health professionals cannot reasonably know about every rare disorder, as there are over 7,000 of them. However, if they had access to those digital resources they could more easily reach a diagnosis, and recommend a treatment faster and more accurately.

[18] https://www.mcnz.org.nz/registration/getting-registered/
[19] https://rarediseases.org/for-patients-and-families/information-resources/physician-guides/
[20] https://rarediseases.info.nih.gov/

3 Research and data

In this section, we explore the scope of research and data in relation to rare disorders. We discuss how research is funded and stimulated internationally, and what could be implemented in New Zealand. We also analysed the importance of data collection in rare disorders health policy.

There are some topic-specific challenges given the size of the population with a specific rare disorder. However, improving rare disorders research and data collection in New Zealand is a pre-requisite for accurately estimating prevalence.

3.1 Barriers to research

Research for the development of new treatments for rare disorders often face the same obstacles. For instance, a study using data from the European Commission's Community Register of Medicinal Products found that, on average, 438 patients participated in clinical studies for new orphan medicines to be approved in the European Union.[21] The small number of patients for each rare disorder poses a significant challenge for research, which is further aggravated in New Zealand given the country's small population.

Additionally, research on new rare disorder treatments is limited by the increased disparity between financial risks and returns. Research institutions often take high risks when investing in new rare disorders treatments. Apart from high research costs, institutions may suffer from the lack of patients available for clinical trials. Also, those institutions can have difficulties in meeting governments' regulatory standards, which can require more comprehensive clinical trials for the approval and financing of new treatments.

Apart from the high risks mentioned, which can put off new research, rare disorder treatments are currently very costly to governments and patients. Since the market for each specific rare disorder treatment is small, pharmaceutical companies have to recover their initial research costs from a small number of buyers. It is also common for rare disorder treatments to have only one provider with monopoly powers. This market environment contributes to the high cost and low competition among rare disorder treatments.

3.2 Barriers to data collection

Data collection is paramount for improving the lives of those living with rare disorders. An integrated and accessible database can aid health professionals in providing an accurate diagnosis and effective treatment to patients, and provide valuable information for researchers and policymakers. ICD-10 is the most used diseases classification system, yet it is not used in most countries, and in many it is only used for mortality statistics.[22,23] Also some countries, for example the United States, Canada, and Australia, have made their own modifications to the system, creating new variants, and compromising compatibility.

[21] https://journals.plos.org/plosmedicine/article?id=10.1371/journal.pmed.1001407#:~:text=The%20average%20number%20of%20patients,that%20might%20otherwise%20be%20unprofitable)
[22] https://icd.who.int/browse10/2019/en#/
[23] https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7960170/

Without a common classification system, it becomes difficult to conduct international research on rare disorders. The low numbers of people living with a specific rare disorder in a single country worsens this problem. Moreover, only a small fraction of rare disorders have their own codes in the ICD-10 system. Therefore, it is not only an issue of adopting a common nomenclature system, but also of adopting a system that gives visibility to rare disorders.

3.3 What can be done to stimulate rare disorder research and data collection?

Although the challenges to research are significant, a number of countries have implemented policies to mitigate it.

Australia

Australia has committed to taking actions for improving rare disorder research in its national action plan.[14] These actions include ensuring people's lived experiences drives research, by encouraging collaboration between researchers and people living with rare disorders through workshops, conferences, and consumer reference groups. Moreover, the plan outlines a focus on international collaborative research, as the government will provide financial incentives for research teams that collaborate internationally. Aimed at mitigating the lack of patients for clinical trials, the Australian Government has announced the goal of increasing the economies of scale of research by operating multi-trial sites.

Orphanet is one of the most prominent non-profit organisations with the mission of improving the lives of those with a rare disorder.[24] It has created the most comprehensive classification system for rare disorders based on using codes called "Orphacodes", preferred terms, synonyms, and definitions.[25] To improve data collection The Australian Institute of Health and Welfare (AIHW) will adopt the Australian National Congenital Anomalies Register (NCAR), including rare disease coding (the Orphacodes).

United Kingdom

In its national rare disorders action plan, Wales has outlined actions to overcome the barriers to research and data collection.[26] For example, £3.4m of new funding from the Medical Research Council and Welsh Government will establish the Wales Genomic Medicine Centre. This virtual Centre will work across Wales with the National Health Service All Wales Medical Genetics Service and experts from Welsh tertiary institutions, to both recruit Welsh patients with rare disorders, and their families, into the programme, and also to interpret and apply genomic data in healthcare.

Wales is also focused on stimulating patient participation in international research. Their action plan outlines the need to develop a risk-proportional permission system to enable researchers and patients to participate in international studies.

The Northern Ireland Clinical Research Network, the Northern Ireland Cancer Trials Network, and other similar institutions, will work with the Department of Health to establish a current baseline for the number of rare disorder studies openly recruiting patients.[16] The goal is to increase awareness and participation through targeted communication.

[24] https://www.orpha.net/consor/cgi-bin/Education_AboutOrphanet.php?lng=EN
[25] http://www.rd-code.eu/introduction/
[26] https://gov.wales/sites/default/files/publications/2019-01/welsh-rare-diseases-implementation-plan-july-2017_0.pdf

In Scotland, the Scottish Health Research Register (SHARE) has created a database of up to one million people available for participation in research.[27] The system is linked to health records, which allows it to easily identify the best candidates for a research study.

New Zealand

In New Zealand, there are a few patient registries for clinical research. Some examples are: the Neuromuscular Disease Registry, the Neuro-Genetic Registry, and the Motor Neurone Disease Registry.[28] [29] [30] However, the lack of a centralised registry, or a registry specific for rare disorders, limits patient awareness and participation in clinical research.

3.4 Recommendations for improving research and data collection

New Zealand uses Australia's version of the ICD-10 system, ICD-10-AM. This is a great limitation to the country's capacity to accurately record data from those diagnosed with rare disorders, and to use that data in international research. We recommend that RDNZ advocates for the Government health agencies to develop a rare disorder patient registry, which uses the Orphanet classification.[4] This would improve data collection and accuracy, and benefit international collaboration.

Also, people could benefit from better communication between care providers as the Orphanet system is implemented, improving diagnosis and treatment. People living with rare disorders may gain visibility, and have a louder voice, giving them more support to advocate for new policies. Lastly, recommendations from section 4 that involve advocating for improving diagnosis, would also feed into data collection and research.

[27] https://www.gov.scot/publications/rare-rare-disease/
[28] https://treat-nmd.org/patient-registry/nmd-registry-new-zealand/
[29] https://www.mda.org.nz/Our-Research/Registry--Biobank
[30] https://mnd.org.nz/research/take-part-in-research/mnd-registry/

4 Diagnosis and treatment

Research, data, awareness, and training all feed into diagnosis and treatment, and so it is the scope that depends the most on the others. In this section, we explore how different countries approach screening and testing, how rare disorder treatments are funded, and how co-ordination of care can deliver better outcomes. Furthermore, by increasing diagnosis speed and accuracy, New Zealand will be one step closer to reaching an accurate prevalence estimate.

4.1 Screening and diagnostic testing

New Zealand

With approximately 7,000 rare disorders, and a set health budget, it is challenging to decide which conditions will be screened for. In New Zealand, newborns are screened for 23 metabolic conditions, and antenatal screening for Down syndrome, Edward's Syndrome, Patau syndrome, and other rare genetic disorders is available. Since most rare disorders are genetic (72 percent), the Genetic Health Service NZ (GHSNZ) is relevant for diagnosis.[31] [32] GHSNZ is a publicly funded network of clinics that provide:

- Diagnostic assessment of genetic disorders
- Diagnostic, pre-conception, prenatal, or pre-symptomatic tests for genetic conditions
- Genetic counselling and management advice for the extended family of affected individuals.

Patients can be referred from General Practitioners (GPs), hospital specialists, and self-referrals from members of families who have received a letter from genetic services suggesting a review. There are only clear guidelines on the referral process for hereditary cancers. For other conditions health professionals will refer patients based on their observations alone.

While New Zealand does screen for a fair amount of conditions, and has a publicly funded network of genetic clinics, the country's performance in rare disorders diagnosis is poor.

Results from a study done with rare disorders patients and their families found that 32 percent of patients waited over 5 years to get a diagnosis.[8]

Around two thirds (64 percent) had to visit three or more doctors to get a diagnosis, and 10 percent had to visit over ten doctors. Also, 62 percent of people were misdiagnosed at least once, and 10 percent were misdiagnosed at least twice.

Overseas studies indicate that New Zealand may be performing worse than its peers in screening and diagnosis. According to a study done in Canada, about 20 percent of patients waited between six and 14 years to get a diagnosis, and 60 percent consulted three to over 20 specialists on the way to a diagnosis.[33] Furthermore, the European Organization for Rare Diseases survey found that 25 percent of patients had to wait between 5 and 30 years for a diagnosis, and 40 percent received an initial erroneous diagnosis. Finally, a study in Australia found that 69 percent of the participants consulted with three or more doctors to get a diagnosis, and 27 percent initially received a wrong diagnosis.[7]

[31] https://www.raredisorders.org.nz/about-rare-disorders/facts-and-figures/#:~:text=72%25%20of%20rare%20diseases%20are,have%20no%20approved%20drug%20treatments
[32] https://www.geneticshealthservice.org.nz/
[33] https://ojrd.biomedcentral.com/track/pdf/10.1186/s13023-017-0618-0.pdf

United Kingdom

The United Kingdom (UK) outlines the actions it will take to improve screening and testing in the rare disorder action plans of its four countries. The UK National Screening Committee (UK NSC) advises Ministers and the National Health Service (NHS) in all four UK countries on all aspects of screening. England's action plan include establishing a UK NSC Bloodspot Task Group, which will identify practical and innovative approaches to facilitate research and evidence which will inform evaluations of blood spot screening.[15] Furthermore, part of the plan is to use these evaluations to reform the UK NSC, with collaboration between researchers and stakeholders, including rare disorder patient groups.

Genomics England and the NHS are also leading a programme to explore the risks and benefits of whole genome sequencing in newborns. This programme involves sequencing the genomes of up to 100,000 newborns. If the programme is successful, and whole genome sequencing is incorporated into NHS screening programmes, it would potentially greatly increase the speed and accuracy of rare disorders diagnosis.

Australia

In the Australian rare disorder action plan, the proposed actions to improve diagnosis and screening include:

- Further development in genomics technology
- A flag in health information systems when someone presents with an undiagnosed rare disorder
- Developing guidelines to aid doctors in rare disorders diagnosis.[14]

4.1.1 Recommendations for improving diagnosis in New Zealand

Enhancing rare disorders diagnosis is paramount for improving the lives of those living with rare disorders. As was previously discussed, New Zealand does have structures that allow for rare disorder diagnosis, but they need to be improved as the country's health system is slow and often inaccurate in providing rare disorder diagnosis.

We recommend that RDNZ advocates for the Government health agencies to conduct a comprehensive review of the National Screening Unit.[34] This review should be opened for the participation of rare disorder patient organisations, health professional associations, and other interested stakeholders. One of the goals of the review would be to provide guidance to the Government health agencies on what actions can be taken to increase early rare disorder detection.

We recommend that RDNZ advocates for the Government health agencies to further develop rare disorder guidelines. These guidelines would make it easier for health professionals to identify rare disorders, and know which specialist to refer the patient to. This is in line with a previous recommendation in the awareness and training section, which involved the development and adoption of digital resources, to improve rare disorder awareness, and information availability, for health professionals.

Finally, we recommend that RDNZ advocates with Genetic Health Service NZ for the Government health agencies to allocate additional funding for genetic testing. The additional funding could provide a greater quantity of tests, but also allow for Genetic Health Service NZ to be up to date with the latest technological developments in the fast-growing field of genomics.

[34] https://www.nsu.govt.nz/

In the UK and Australia, the public health sector is giving greater attention to developments in genomics, which can be ground-breaking for rare disorder diagnostics. We also recommend that RDNZ considers the possibility of further advocacy in regard to genome sequencing in newborns, depending on the results from England. New Zealand should not fall further behind its peers in genetic testing.

4.2 Treatment funding

Although less than five percent of rare disorders have a specific treatment, having access to treatment can be life-changing for people living with rare disorders. As was brought up in section 3, rare disorder medicines, or orphan drugs, are often significantly more expensive than regular medicines. Therefore, rare disorder patients are unlikely to have the financial means to afford the treatment, making them heavily dependent on public funding.

In New Zealand, Pharmac is the agency that makes decisions about which medicines will be publicly funded.[35] Pharmac is the only agency of its kind in the world that decides which medicines will be funded based on a fixed budget. The Minister of Health and the Government set Pharmac's budget and appoint the board. While the agency is accountable by the Ministry of Heath, it holds a great degree of independence, and has been able to promote competition among pharmaceutical companies through its funding channels. Over time, Pharmac has been able to negotiate better prices, allowing for more medicines to be funded.

4.2.1 Funding channels

There are three channels of funding through Pharmac:[36]

The Pharmaceutical Schedule listing process:

This is the standard process for deciding which medicines will be funded at the population level. When deciding which medicines will be funded Pharmac takes into account:

- The health needs of the person
- The availability and suitability of existing medicines
- The health needs of others.

The Exceptional Circumstances Framework:

This is a pathway for considering medicines funding applications for individual patients that fall outside of the Pharmaceutical Schedule. This pathway follows the Named Patient Pharmaceutical Assessment (NPPA).[37] Under this framework, Pharmac will take a two-step-approach when considering the application:

Step One:

a. Does the person have exceptional clinical circumstances?

b. Has the person tried all funded alternative treatments?

c. Has Pharmac previously considered the treatment for funding?

[35] https://pharmac.govt.nz/
[36] https://pharmac.govt.nz/assets/2019-Report-Funding-Medicines-for-Rare-Disorders-PDF-version.pdf
[37] https://pharmac.govt.nz/assets/nppa-patient-info.pdf

If the answers to questions “a” and “b” are “yes”, and the answer to question “c” is “no”, the application goes forward.

In Step Two, the application is assessed using factors of consideration divided into four dimensions: need, health benefits, costs and savings, and suitability. Finally, the application is assessed using three different levels of impact: the person; the person’s family, whānau, and wider society; and the broader health system.

The rare disorders medicines contestable funding pilot:

This is a pilot process initiated to stimulate competition amongst suppliers of rare disorders medicines. Pharmac released a request for proposal (RFP) seeking funding applications for rare disorders medicines. The agency set aside $5 million per year for this funding channel, and through it Pharmac received 28 bids and managed to fund 10 new orphan drugs.

4.2.2 The funding gap

Pharmac claims that its current funding pathways are effective in funding medicines for patients with rare disorders, however, data from Pharmac shows that only 102 medicines were funded for 348 patients in 2018.

A relevant issue with defining funding for orphan drugs is the lack of data around rare disorders prevalence in New Zealand. Delays and inaccuracies in diagnosis, which is impacted by the lack of rare disorder awareness and training, result in significant underreporting. Moreover, the lack of data and studies around the prevalence of rare disorder in New Zealand makes it difficult for stakeholders to precisely estimate the size of the problem.

Using international estimates is the only way in which we can currently estimate the prevalence of people living with rare disorders in New Zealand, including those living with rare disorders for which there is s treatment. The table below uses a range of prevalence rates from different sources, and two estimates of the rate of people living with a treatable rare disorder. The main point to take away from the table is the fact that, no matter which set of estimates are used,

There is a major gap between the current number of people receiving funded rare disorder treatments from Pharmac, and the total number of people living with a treatable rare disorder in New Zealand.

Table 1 Estimated number of New Zealanders with rare disorders, and percentage with treatable rare disorder

Prevalence (percent)	Estimated number of people with a rare disorder	Estimated number of people with a treatable rare disorder (2.4%)	Estimated number of people with a treatable rare disorder (5%)
1.5	76,839	1,884	3,842
2.0	102,452	2,459	5,123
3.0	153,678	3,688	7,684
4.0	204,904	4,918	10,245
5.0	256,130	6,147	12,807
6.2	317,601	7,622	15,880

Source: https://pharmacreview.health.govt.nz/assets/Uploads/final-report/Pharmac-Review-Final-Report.pdf

Estimates used internationally vary considerably from 1.5 to 6.2 percent of the population. This is largely due to the variability in rare disorder definitions used, as well as the lack of data. If these estimates were applied to Statistics New Zealand's estimate of the New Zealand population in 2021, the number of people living with rare disorders would be between 76,839 and 317,601 (Table 1).

However, a study using rare disorders prevalence data from Orphanet concluded that a conservative, evidence-based estimate for the population prevalence of rare disorders was between 3.5 and 5.9 percent.[38] This range is derived from data on 67.6 percent of the known rare disorders; and using the European prevalence definition of five per 10,000 people.

Therefore, the Orphanet study suggests that estimates on the higher end of the range are more accurate. This means that between approximately 180,000 and approximately 300,000 people living with rare disorders in New Zealand is a reasonable estimation of prevalence. Table 1 also supports the argument that further study and data collection is needed to estimate the prevalence of people living with rare disorders in New Zealand, because even with the Orphanet estimates, the range is wide.

Another reason for the funding gap is the lack of an official definition of rare disorders in New Zealand. While there is no agreed international definition, the average threshold is a prevalence of one in 2,500 people. Australia and Scotland also have a definition for ultra-rare disorders, which is a prevalence of one or less in 50,000 people. Despite not having an official definition, Pharmac has its own definition of rare disorders, which is a prevalence of less than one in 50,000 people in New Zealand.

4.3 Recommendations on addressing the funding gap

It is clear that by considering a definition of rare disorder that is widely different to the ones used in other countries, and because of the lack of prevalence data and diagnosis, Pharmac underestimates the size of the problem.

On another point, if more attention was given to rare disorder diagnosis Pharmac may be in a better negotiating position. Pharmac negotiates with suppliers on price, and runs a range of procurement process to ensure competitive pricing. It's possible that, as the number of people diagnosed with rare disorders increases, Pharmac would be in a position to negotiate larger quantities of each medicine, which may lower the unit price.

We recommend that RDNZ advocates for the Government health agencies to adopt an official definition of rare disorders that is in line with international best practice. Furthermore, we recommend that RDNZ advocates for Pharmac to expand its rare disorders medicines contestable funding pilot, and to admit that under the current model a significant proportion of people living with a treatable rare disorder are not being covered. Finally, recommendations from previous sections that involve advocating for improving diagnosis and data collection, would also work to help Pharmac to allocate funding for rare disorder treatments.

4.4 Coordination of care

Coordination of care is another relevant aspect for people living with rare disorders. Because of the complex nature of their health problems, they often have to visit a wide range of health professionals from when they first try to obtain a diagnosis, until they receive treatment.

[38] https://www.nature.com/articles/s41431-019-0508-0

Miscommunication between different health service providers can delay diagnosis and negatively impact treatment. In this section we pull international examples of actions to improve coordination of care, explain the domestic context, and explore options for enhancing coordination of care in New Zealand.

United Kingdom

The action plans from the four UK countries showcase a range of actions with the goal of enhancing coordination of care, and some of those actions could be adapted and also implemented in New Zealand. In England, the Department of Health and Social Care has stated its intention of developing digital approaches to rare disorders care, including virtual consultations.[15]

This can be a particular advantage for some rare disease patients who may live a long way from centres specialising in their condition, or who may not need a face-to-face appointment on every occasion, or who may find travel difficult. The adoption of digital approaches has the potential to improve coordination of care by allowing rare disorder patients to access the care of multiple specialists without the need to travel long distances.

Another action is to support more integrated care across the system. The government introduced integrated care systems (ICSs) in 2018. These are partnerships between the organisations that meet health and care needs across an area, with the goal of coordinating services, but they are not mandatory. In order to expand the ICSs, the Health and Care Bill proposed to build on the work of the existing non-statutory ICSs by requiring the creation of integrated care partnerships in each local system area.

There are a few common actions across the four UK nations. One is to develop expert centres for rare disorders that follow the same UK-wide standards. Another action is to work with international partners to connect with their expert centres for rare disorders, and thus form an international collaborative network.[15] [16] [26] [27]

Australia

Although Australia's rare disorder action plan does not have coordination of care as one of its broader themes, the country did take action to improve its coordination of care for rare disorders. Rare Care Centre, an expert centre for rare disorders, is being developed at Perth Children's Hospital. The Centre will deliver improved awareness and early identification of children with potential rare disorders, and enhanced referrals to support earlier and more accurate diagnosis. It will also provide improved support and care coordination. As well as better access to community resources, clinical trials, and research.[39]

New Zealand

In New Zealand, rare disorder patients have expressed the view that coordination of care needs to be improved. In a survey, 52 percent said that the quality of communication between service providers was poor.[8] In that same survey, a patient commented how they have to be constantly repeating themselves to different doctors, and how they wished doctors had access to their latest data.

[39] https://www.cahs.health.wa.gov.au/News/2022/02/25/Rare-diseases-centre-at-PCH

New Zealand does have examples of good practice in information sharing between service providers. However, these are limited due to their fragmentation, and the fact that there is no country-wide information sharing system.

For example the GP2GP system, used by the majority of GPs across the country, allows GPs to transfer patient records electronically. The electronic Shared Care Record View (eSCRV) is present in all DHBs in the South Island. The eSCRV holds patients' information on their conditions, test results, medications, allergies, and correspondence between services. GPs and all public hospitals in the South Island can access that information. District Health Boards (DHBs) have used the Picture Archiving and Communication System (PACS) to store and share x-ray images. However, DHBs are grouped together in regional PACS archives instead of being together in a national network.

The current measures to promote information sharing are too fragmented and regionalised. Furthermore, New Zealand's health system is currently undergoing major systemic reform.[6] The 20 DHBs have been dismantled and replaced by a single national entity, Health New Zealand. This could allow for great advancements in coordination of care, as it would be easier to share information within a single entity.

4.5 Recommendations for better coordination of care

We recommend that RDNZ advocates for the Government health agencies to take advantage of the health system unification reform, and implement a fully integrated country-wide information sharing system. This system could be based on the South Island's eSCRV.

Also, we recommend that RDNZ advocates for the Government health agencies to develop an expert centre for rare disorders in the country. Since rare disorders are more common in children, this centre could be part of Auckland's Starship Children's Hospital, which is already the leading provider of paediatric care in the country, and has a range of specialists. This expert centre for rare disorders should also be developed aiming at international collaboration with other expert centres, especially with the ones in the UK and Australia. By having an expert centre for rare disorders, New Zealand would be keeping pace with the leading countries in rare disorders care. International collaboration would also mitigate our small population issue, particularly for ultra-rare disorders.

5 The costs of a failed system

In this section, we explore the cost of inaction. We outline a range of studies from New Zealand and internationally that bring up different direct and indirect costs related to rare disorders. The main take away point is that the costs of rare disorders are significantly higher than the costs of more common diseases, and that people living with rare disorders, and their carers, carry a heavy burden.[40] Lastly, the goal of this section is to demonstrate that continuing to ignore the problem is costly for the health system and society in general, and therefore immediate action must be taken.

5.1 The costs to the health system

While rare disorder diagnosis and treatments can be expensive, failing to properly diagnose and treat rare disorder patients is also costly, as these patients have to spend many nights under hospital care to manage their symptoms. Since caring for rare disorder patients is costly, actions are needed to make the health system more efficient in managing these conditions.

Australia

A study from Australia that ran from 1999 until 2010 with a cohort of 61,279 hospital patients, found that cohort members represented two percent of the Western Australia (WA) population. [41] Despite that, the cohort represented 4.6 percent of the total number of patients discharged from hospital and 9.9 percent of all discharges in WA during that period. The mean number of discharges per patient during 2010 for the cohort was twice that of all people discharged from hospital in WA. However, considering only rare-disorder-related discharges, the mean number of discharges per patient was only slightly more than that of all people discharged from hospital in WA.

The mean length of stay for all discharges during 2010 for the cohort was 3.6 days, and for the subset of rare-disorder-related discharges, it was 5.5 days. For comparison, the mean length of stay for all hospital discharges in WA in 2010 was 2.9 days.

The total cost associated with hospital discharges for the cohort during 2010 was AU$394,947,610, which was 10.5 percent of the total WA inpatient hospital expenditure. By considering only rare-disorder-related discharges, the cost was AU$173,322,256, which was 4.6 percent of the total WA inpatient hospital expenditure. Therefore, this study shows that rare disorder patients cost significantly more to the health system, and that these patients also suffer disproportionately more than patients with common diseases.

United States

A study conducted in the United States (US) estimated the overall economic burden of 379 rare disorders to be greater than US$966 billion in 2019, including US$418 billion in direct medical costs, and US$548 billion in indirect and non-medical costs absorbed directly by patients and their families.[42] Since this estimate is based on a subset of 379 rare disorders rather than the over 7,000 known rare disorders, the economic impact estimate represents a lower bound estimate and is not generalisable to all rare disorders. The total excess medical cost associated with the 379 rare disorders is an estimated US$418 billion in 2019, with a per-person excess cost of US$26,887.

[40] The term “common diseases” refers to diseases that would not be classified as “rare” under any definition. Examples include Influenza, bronchitis, and diabetes.
[41] https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5440569/
[42] https://ojrd.biomedcentral.com/track/pdf/10.1186/s13023-022-02299-5.pdf

This means that an average rare disorder patient has an annual medical cost that is US$26,887 more than an average patient.

Another study from the US found that the burden of rare disorders is approximately ten times higher than common diseases on a per patient per year (PPPY) basis.[43] Some of the finding include:

- The burden was generally driven by direct and mortality costs
- A lack of treatment for a rare disorder is associated with a 21 percent increase in total costs PPPY
- The average costs with and without treatment:
    - Direct costs: US$63,000 PPPY with treatment vs. US$118,000 PPPY without treatment
    - Indirect costs: US$40,000 PPPY with treatment vs. US$73,000 PPPY without treatment
    - Mortality costs: US$36,000 PPPY with treatment vs. US$49,000 PPPY without treatment

Hence, apart from showing that costs related to rare disorders are much higher than costs related to general diseases, this study highlights that failing to treat rare disorder patients incurs even higher costs.

5.2 The costs to patients and their carers

It is not only the financial costs of rare disorders to the health system that matter. People living with rare disorders and their carers have their lives negatively impacted in various ways, for example potentially experiencing limited social lives and unequitable financial outcomes.

New Zealand

A study completed in New Zealand with rare disorder patients and their carers found many insights around the difficulties that this group face.[8] The findings include:

- Almost 55 percent reported that the disorder impacted a lot on their health and everyday life
- For 60-75 percent of people and their families surveyed, the rare disorder makes a number of everyday activities difficult (communicating, controlling behaviour, social life)
- 32 percent required more than two hours per day for illness-related daily tasks including hygiene, helping with house chores, moving the person and administration of treatments
- There is high utilisation of healthcare services
- 81 to 87 percent of people had seen a specialist or GP in the last 70 days
- One in three rare disorder patients had a hospital stay in the last year (2020).
- On average they spent almost 13 days as an inpatient in 2020 and, for one in 17 people, almost seven days in intensive care (ICU)
- Almost one third of respondents (30 percent) were having to undertake self-funding for at least some of their medicines
- The majority (59 percent) of people felt that the costs associated with the rare disorder were high

[43] https://chiesirarediseases.com/assets/pdf/chiesiglobalrarediseases.whitepaper-feb.-2022_production-proof.pdf

- In addition, 54 percent felt the costs associated with managing their rare disorder were hard to manage
- Overall, 25 percent were unemployed, with the majority citing long-term illness or disability
- The vast majority of people (70 percent) saw a reduction in income related to a rare disorder
- Almost 80 percent felt the rare disorder had a moderate to significant impact on their learning, with 23 of percent children being absent from school for more than 30 days per year
- The majority of people have experienced increased tension between family members (57percent) and isolation from family and friends (69 percent)
- One in three respondents often felt unhappy and depressed and felt they could not overcome their problems
- One participant commented that the life changing treatments they need are not publicly funded, and they will have to move to Australia to access them.

This study outlines the variety of challenges rare disorder patients and their families face in New Zealand, and how the current system does not work in their favour. All of those problems will not be solved, unless new policies are put in place.

United Kingdom

A study from the UK found that rare disorder patients and their carers incur heavy costs that are often not measured.[44] These costs include:

- Costs associated with appointments:
  - Time off work and reduced income
  - Childcare
  - Travel
  - Accommodation
  - Accessible vehicles and transport options
- Financial costs associated with wider condition management:
  - Private healthcare
  - Childcare and respite
  - Specialist activities and equipment
  - Prescriptions
  - Fees for informal helpers and carers
  - Disruption to employment and income
- Time costs:
  - Time off work
  - Time spent coordinating care and the various agencies and appointments involved
  - Time spent fighting to access care and support

[44] https://www.geneticalliance.org.uk/media/2502/hidden-costs-full-report_21916-v2-1.pdf

- Psychosocial, health, and wellbeing costs:
    - Disruption to schooling, employment and personal time
    - Impact on relationships and social life
    - Isolation
    - Mental health.

We can take away from this study that there is a range of hidden costs related to rare disorders that are often not given their weight in discussions around rare disorder policies.

Canada

A study conducted in Canada with carers of rare disorder patients found that they also carry heavy burdens from their duties.[45] The study found that:

- Almost 90 percent have had family finances negatively impacted
- 68 percent have had their work performance negatively impacted
- 71 percent had to quit their jobs or seek flexible arrangements
- 41 percent miss six or more days of work a month due to caregiving responsibilities
- Almost 80 percent suffer from mental health issues
- Half are only able to engage in social activities less than once a month.

Therefore, this study from Canada is in line with the others. People living with rare disorders and their carers face similar challenges in the countries studied. Those challenges are often hidden and not recognised by policymakers and wider society. This provides another argument for the need of increasing awareness around rare disorders.

[45] https://www.raredisorders.ca/canadas-rare-disease-caregivers-under-immense-stress-struggling-with-mental-health-issues-isolation-and-financial-burden/

6 Recommendations

In this section, we outline the recommendations at a broad level. One of the main findings is that it is not possible to estimate the prevalence of rare disorders in New Zealand with any level of accuracy. This is due to a lack of publicly available data and current data limitations.

Taking this limitation into consideration, our recommendations are based on actions to mitigate the overall challenges faced by people living with rare disorders, and also on actions that will develop the tools needed to accurately estimate prevalence in New Zealand. We shaped our recommendations so that they could be part of a broader rare disorders national framework, or could be implemented individually.

A common supportive document for our recommendations is Resolution 76/132 adopted by the United Nations General Assembly, of which New Zealand is a signatory.[1] Members agreed, based on a series of conventions including the Universal Declaration of Human Rights, actions should be taken to improve the lives of those living with rare disorders.

The document:

- Calls upon member states to strengthen health systems, notably in terms of primary health care, in order to provide universal access to a wide range of healthcare services that are safe, of quality, accessible, available and affordable, timely, and clinically and financially integrated. These services will help to empower persons living with a rare disease in addressing their physical and mental health needs to realize their human rights, including their right to the highest attainable standard of physical and mental health. They will also enhance health equity and equality, end discrimination and stigma, eliminate gaps in coverage and create a more inclusive society
- Encourages member states to adopt gender-sensitive national strategies, action plans and legislation, that contribute to the well-being of persons living with a rare disease and their families, and that promote the protection and enjoyment of their human rights. These actions are consistent with the member states' obligations under international law.

Our recommendations are also in line with actions promoted by the World Health Organisation (WHO). The WHO has signed a memorandum of understanding (MoU) with Rare Diseases International (RDI).[46] RDI is an international organisation that brings together national and regional associations of patients with rare disorders, as well as international federations and multi-stakeholder groups to achieve greater equity for the global community of people living with rare disorders.[47] In the MoU, the WHO pledges to collaborate with RDI for the development of a Collaborative Global Network for Rare Diseases (CGN), and an operational definition of rare diseases.

Based on our research and the evidence available, our recommendations are focused on advocacy for change and or action in three areas: awareness and training; research and data; and diagnosis and treatment.

Advocate for actions that will increase awareness and improve training

- Advocate for the Government health agencies to fund a study with health and support professionals to gather evidence of their level of awareness and knowledge of rare disorders
- Partner with the University of Otago and the University of Auckland to conduct a similar study with healthcare students:

[46] https://www.rarediseasesinternational.org/working-with-the-who/
[47] https://www.rarediseasesinternational.org/fr/vision-et-mission/

  - Similar international studies found that health and support professionals, and students have a poor level of awareness and knowledge of rare disorders
  - These studies will allow the Government health agencies and other stakeholders to better understand the size of the problem
- Advocate for the Government health agencies to fund awareness-raising campaigns and events in New Zealand:
  - One of these events could be a rare disorders case competition for tertiary students
- Advocate for the University of Auckland and the University of Otago to further include rare disorders in their curricula
- Advocate with The Royal New Zealand College of General Practitioners and The Royal Australasian College of Physicians, for the Medical Council of New Zealand, and the Australian Medical Council, to further include rare disorders in the Continuing Professional Development (CPD) curricula:
  - New policies were introduced, to improve training, in countries with similar negative health outcomes for rare disorder patients to New Zealand
  - CPD needs to further include rare disorders as current healthcare professionals are not fully meeting patients' needs, and also over 40 percent of doctors registered in New Zealand completed their qualifications overseas
  - It would be beneficial to take England's example and create a working group including patient organisations, curricula developers, and professional organisations, to discuss how to better approach rare disorders in education
- Advocate for the Government health agencies to implement digital resources that support health professionals reach a diagnosis and a treatment plan:
  - These digital resources could include a digital repository with relevant information about rare disorders
  - This recommendation would also contribute to improving diagnosis and treatment.

Advocate for actions that will improve research and data collection

- Advocate for the Government health agencies to develop a rare disorder patient registry, which uses the Orphanet classification. This would:
  - Improve the way data is collected and classified
  - Make international research and collaboration easier
  - Increase diagnosis speed and accuracy.

Advocate for actions that will improve diagnosis and treatment

- Advocate for the Government health agencies to conduct a comprehensive review of the National Screening Unit:
  - This review should include all interested stakeholders
  - Its main goal would be to provide guidance to the Government health agencies on what actions can be taken to increase early detection of rare disorders

- Advocate for the Government health agencies to further develop rare disorder guidelines:
    - The guidelines would make it easier for health professionals to identify rare disorders, and know which specialist to refer the patient to
- Advocate with the Genetic Health Service NZ for the Government health agencies to allocate additional funding for genetic testing:
    - Extra funding would allow for the service to expand and increase speed and the number of diagnosis
    - The Genetic Health Service NZ also needs to be up to date with the latest technological developments in the fast-growing field of genomics
- Consider the possibility of further advocacy in regards to genome sequencing in newborns, depending on the results from England in regards to the efficiency of genome sequencing in newborns:
    - If proven effective, genome sequencing in newborns could greatly improve diagnosis
- Advocate for the Government health agencies to adopt an official definition of rare disorders that is in line with international best practice:
    - An official definition would give greater visibility to rare disorders
    - This could encourage Pharmac to change its own definition
- Advocate for Pharmac to expand its rare disorders medicines contestable funding pilot, and to admit that under the current model a significant proportion of people living with a treatable rare disorder are not being covered:
    - To solve the problem it first needs to be recognised
    - Pharmac could fund a greater number of life-changing treatments for people living with rare disorders
    - The high social costs of rare disorders may decrease
- Advocate for the Government health agencies to take advantage of the health system unification reform and implement a fully integrated country-wide information sharing system:
    - This system could be based on the South Island's electronic Shared Care Record View (eSCRV)
    - This would significantly improve coordination of care
- Advocate for the Government health agencies to develop an expert centre for rare disorders in the country:
    - This centre could be part of Auckland's Starship Children's Hospital
    - A dedicated centre put New Zealand in line with its peers
    - Such a centre would also enable greater international collaboration and research.