Rare Disorders New Zealand (RDNZ) response to Manatū Hauora's initial Rare Disorders Strategy (RDS) document[1]

Introduction

RDNZ welcomes Manatū Hauora's initial Rare Disorders Strategy document, and appreciates the opportunity to contribute both as a member of the RDS reference group and as a co-designing partner. We see the RDS as a breakthrough opportunity to significantly improve health and other outcomes for New Zealanders living with rare disorders. We expect the RDS to result in improved access to health and other services and opportunities and to enable people living with rare disorders, to experience pae ora and healthy futures on their terms and as promised for all New Zealanders by the health reforms legislation[2].

As per that legislation, six Pae Ora Healthy Futures Strategies have been developed which "set the direction for a system that is equitable, accessible, cohesive and people-centred"[3]. Although it sits outside of the legislatively required pae ora strategies, RDNZ very much considers the RDS to be the seventh strategy in the suite. We expect that the RDS will be accorded at least the same priority as the Pae Ora Healthy Futures Strategies when decisions are made as to how the strategies will be supported and implemented, including financiallyand through the development of robust action plans and systems and health outcomes accountability processes.

Although RDNZ is but one member of the reference group from which Manatū Hauora has sought feedback we expect to see the role of our organisation embedded as an active co-designing participant, rather than merely being an external observer and commentator.

As requested in the draft RDS document RDNZ's feedback is provided in response to the questions on page 1 of the document. In preparing our response we have elicited feedback from rare disorders support groups, from the RDNZ Board, and collectively from RDNZ staff.

Throughout this document we refer to people with a rare disorder as being those individuals who have a condition or diagnosis or unusual co-presenting undiagnosed symptoms[4], and people living with a rare disorder as being those with a diagnosis or

[1] Manatū Hauora. A rare disorders strategy for Aotearoa New Zealand: initial proposed content and points to consider and provide feedback on. 30 June 2023.
[2] Pae Ora (Healthy Futures) Act 2022.
[3] Pae Ora Strategies. July 2023. https://www.health.govt.nz/new-zealand-health-system/pae-ora-healthy-futures-all-new-zealanders/pae-ora-strategies
[4] Co-presentation of unusual, often different and sometimes rare, symptoms can point to a unifying diagnosis, just as unusual combinations of symptoms and signs in an individual should trigger diagnostic questioning, (Marks R, Robertson S, Latu A, Rare Disorders, NZ Doctor, 27 October 2021).

condition or unusual co-presenting undiagnosed symptoms together with whānau, family and others directly involved with that person's care and support.

We are yet to comment specifically on how the strategy should respond to issues experienced by Māori and whānau living with rare disorders, as we are awaiting the outcome the consultation process being managed by Te Aka Whai Ora in partnership with RDNZ.

Feedback from RDNZ support groups

RDNZ acknowledges and thanks the rare disorders community for the feedback from support group leads, sought by RDNZ as requested by Manatū Hauora.

Feedback from 24 support groups is attached as Appendix B, a summary of the feedback compiled by RDNZ is attached as Appendix A, and a document outlining things going well, barriers, and how groups wished things were for people living with rare disorders is attached as Appendix C

We have reflected the groups' feedback in the body of our submission below, and wish also to highlight the following key themes:

- The need for Manatū Hauora to consult more directly with the rare disorder community rather than only by email through RDNZ, and we appreciate the additional steps that Manatū Hauora has agreed to in response

- It was raised that it is premature to decide upon the aims and focus areas prior to undertaking genuine co-design processes and lived experience expert consultation, as well as assessment of the current situation

- It was noted that 'pae ora' on its own is a meaningless term for many with a rare disorder, and that a definition of what pae ora means specifically for those with a rare disorder is needed. Not only do people living with rare disorders need healthy futures, they also need health, wellbeing, inclusivity, and adequate care, now. The overall timeframe the strategy is working to should not be a barrier to pressing rapidly in any quarters where improvements in access to care and treatment could be made right now

- It was requested that there is an analysis of gaps and shortfalls in the current system. There was concern that without understanding our baseline it is difficult to know where the focus needs to lie, as well as how to measure progress

- There was concern that there was not a section that discussed previous reviews and work streams that have made recommendations in respect of rare disorders, in particular the recent Pharmac review, so that these shortcomings are captured and brought through to the focus areas to ensure those issues are being addressed

- It was also raised that there is very little commentary or evidence presented regarding how New Zealand currently compares internationally in relation to rare disorders care and outcomes, especially those countries that have implemented strategies and action plans for rare disorders

- There was a lot of concern about the statement "the strategy will not do the work or identify specific actions or commitments in focus areas...". It was asked that the strategy identify the gaps in current programmes and activities and the actions to be taken to address those gaps. It was noted the strategy needs to be explicit regarding the parties responsible for addressing those gaps and timing for that, as well as how progress will be measured and how agencies will be held accountable for delivery

- Groups sought recognition that not getting it right for people with rare disorders has a wider societal impact, including a large social and economic impact. It was noted that an absence of equitable and clear pathways and processes for rare disorders diagnosis and management can result in high costs to the health system in the long term. It was highlighted that missing opportunities for early intervention and best-practice care can result in people who are less productive in society, and that this has a spill-on effect to their carers and entire whānau, which is often overlooked and not measured

- Rare disorder support groups hold knowledge of patient experience and have awareness of international practice. Many of the groups noted they have international contacts and relationships with their counterparts and experts in their disorder internationally. They asked to be involved in the development process of standards of care guidelines for New Zealand, and for referral to them to be built into care pathways. Some groups already hold resources that can be utilised more widely and have their own registries.

- The groups emphasised that timely and accurate diagnosis is important but only the beginning. They are asking for a holistic coordinated approach that: recognises the role and responsibilities of family/whānau, including into adulthood; foresees and addresses the implications for education and employment; includes support at times of transition e.g. from paediatric to adult care; is proactive and considers appropriately scheduled reviews and recalls as well as screening for known complications.

- Support groups expect to see real action and measurable progress on the ground for people with rare disorders, with agencies held accountable for delivery of their aspects of the strategy.

To address these concerns, Rare Disorders NZ asks an action plan or implementation plan accompanies the strategy. We would like to see the plans for the creation of

this, including a clear timeframe for delivery and accountability for progress, incorporated in the strategy.

Support group feedback aligns with RDNZ's seven priorities to improve health and wellbeing for people living with a rare disorder and we would like to see these more specifically incorporated in the focus areas. Appendix D provides elaboration on the seven priorities and Appendix E elaborates on RDNZ's proposed centre of excellence, both of which RDNZ recommends for inclusion in the RDS.

Feedback from the RDNZ Board

Key points from an RDNZ Board meeting convened to discuss the consultation draft strategy are as follows:

Manatū Hauora's RDS process

- A board member identified as a member of the RDS reference group representing the Paediatrics Society
- There's a need to bring a selection of stakeholders together to develop the strategy, which could comprise a hui of reference group members plus community group members
- People with rare disorders should be clearly visible in the RDS as its co-authors and co-designers, as opposed to being seen to be on the sidelines.

RDS content

- "I didn't see myself in it" [as a parent caring for a child with a rare disorder]
- Needs to address childhood mobility
- Needs to address the role of carers
- Access to medicines was avoided (apart from an RDNZ reference)
- No mention of education and need for individualised support
- No mention of work place and social support
- Needs to address issues of continuity of care and multiple/repeated requests for personal information
- Don't want to end up with a different set of boxes and barriers that people with rare disorders have to jump through
- Content re Te Tiriti is very general and needs to be tailored to the specific issues being faced by Māori and whānau living with rare disorders
- RDNZ's values need to be articulated in the RDS
- The document is soulless and doesn't reflect the paramountcy of people living with rare disorders, nor the flow on effects to wider family, whānau and others, nor the mental health toll on families and whānau living with rare disorders
- Needs to address impact and costs for the wider community
- Needs to address "temporal equity", noting that life expectancy for people with rare disorders can be short

- Treatment paradigms need to be tailored for rare disorders (what's appropriate for treating a common condition can be disastrous for a rare disorder)
- Needs to recognise international connections and input.

Comments from an RDNZ Board member who was not able to be at the Board meeting, and who might have been expected to have been invited to be a member of the RDS reference group given his clinical and research expertise in genetics, is attached as Appendix F.

Are there additional points that are important to capture?

RDNZ considerers that there are a number of additional points that are important to cover, and that some of the points made should be differently expressed so that the RDS becomes an aspirational – yet achievable – document that focuses on what we can and should be doing rather than making concessions to what we may struggle to achieve. RDNZ's recommendations are set out in Appendix G by way of "post it notes" annotations to the Manatū Hauora draft.

We haven't necessarily repeated all of our annotations in this the body of our submission, and we do therefore ask that the authors of subsequent versions of the RDS take particular note of Appendix G's detailed observations and feedback.

Why have a rare disorders strategy?

New Zealand is a signatory to the United Nations resolution on "Addressing the Challenges of Persons Living with a Rare Disease and their Families"[5]. RDNZ believes that this is a further powerful reason for having an RDS and which should be articulated as part of the strategy.

The resolution recognises "the need to promote and protect the human rights of all persons, including [those] living with a rare disease … many of whom are children, by ensuring equal opportunities to achieve their optimal potential development and to fully, equally and meaningfully participate in society".

Clause 1 calls upon Member States "to strengthen health systems … which 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 mental health, to enhance health equity and equality, end discrimination and stigma, eliminate gaps in coverage and create a more inclusive society".

Furthermore, clause 2 "encourages Member States to adopt gender-sensitive national strategies, action plans and legislation, to contribute to the well-being of persons living with a rare disease and their families, including on the protection and

[5]United Nations Resolution on "Addressing the Challenges of Persons Living with a Rare Disease and their Families." December 2021. 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

enjoyment of their human rights, consistent with their obligations under international law".

RDNZ broadly agrees with the remainder of the reasons for having an RDS, with the following provisos:

- "Pae ora" needs to be articulated specifically for people living with a rare disorder. A child, for example, who is disabled from an early age as a result of a life shortening rare disorder will not necessarily be looking forward to a healthy future, and neither will their family. The UN resolution refers to achievement of optimal potential development and the right to the highest attainable standard of physical and mental health, and RDNZ recommends that pae ora be articulated in these and similar terms

- It should also be noted that the achievement of pae ora expressed in these terms may be an investment in New Zealand's economy as people with rare disorders who receive appropriate and timely clinical interventions will have a better chance of being able to productively contribute to the country's economic well being. Newborn screening and timely therapies for conditions such as SMA and infantile onset Pompe disease, funding of Trikafta for cystic fibrosis, and education strategies for parents and teachers of fragile X children [6] are all examples of how people can have opportunities to enjoy productive lives as a direct result of appropriate interventions.

- The "impacts of rare disorders" should reference the challenges that families face in navigating the health and other systems, and we illustrate in Appendix H the specifics of what this looks like for one family. Individuals, carers and whanāu, are currently carrying the load of filling the gaps and navigating a system that is not designed with rare disorders in mind. The RDS should address this

- Unrecognised and undiagnosed conditions should be discussed in the context of rare disorders (rather than non-rare conditions). The point which should be highlighted in this section is the diagnostic odyssey which is experienced by many who live with a rare disorder, and which results from the combined effects of inadequate clinical knowledge, clinical blind spots and unavailability of and lack of access to current and emerging diagnostic capabilities including genetic testing. This section could also acknowledge that some conditions which have hitherto been considered common will become recognised as rare as better scientific understanding emerges of their specific genetic influences.

[6] No Longer Fragile: Education strategies for parents and teachers of fragile X children. Education | Fragile X (Fragile X is the world's leading cause of inherited intellectual disability and the leading identifiable cause of autism)

- The majority of people in the 2021 'Impact of Living with a Rare Disorder in Aotearoa New Zealand' survey conducted by Rare Disorders NZ had challenges in getting a diagnosis for their rare disorder: over half took longer than 1 year to get a diagnosis and for one in five the time taken to get a diagnosis was over 10 years. The majority (64%), had to visit 3 or more doctors to get a diagnosis and for one in ten over 10 doctors were visited. 62% people with a rare disorder were misdiagnosed at least once before final diagnosis was confirmed. One in ten were misdiagnosed at least twice.

- The section on Te Tiriti o Waitangi responsibilities appears light, and RDNZ looks forward to seeing this being more specific to rare disorders following Te Aka Whai Ora's consultation with Māori and whānau who live with rare disorders.

- The section on equity should specifically address what this means for people living with rare disorders rather than people with disabilities. We acknowledge the overlaps, but note that health equity issues for disabled people are covered in the provisional health of disabled people strategy[7] and shouldn't need to be re-traversed in the RDS. RDNZ commends Eurordis content[8] which affirms that "… people have equal rights from birth. Recognising and protecting these rights requires efforts from wider society which are targeted and proportionate to the needs of people living with a rare condition. Achieving equity … must be about addressing the specific vulnerabilities of the rare disease population…for people living with a rare disease, this means the assurance of social opportunity, non-discrimination in education and work, and equitable access to health, social care, diagnosis and treatment".

What are rare disorders?

RDNZ supports the proposed definition.

Aims and principles

RDNZ agrees with the key elements (aims and principles) proposed for the strategy, subject to tighter definition of what pae ora looks like for people living with rare disorders (as articulated above) and inclusion of the following additional principle:

Supports health and other sector implementation of actions and accountability for health system and pae ora outcomes.

[7] Provisional Health of Disabled People Strategy. Manatū Hauora. July 2023. https://www.health.govt.nz/system/files/documents/publications/provisional-health-of-disabled-people-strategy-jul23.pdf

[8] Rare Disease Day 2023: Our global community comes together for a more equitable world. Eurordis. February 2023. https://www.eurordis.org/rare-disease-day-2023-our-global-community-comes-together/

Additional opportunities that could be built on

- Te Whatu Ora's emerging programme of National Clinical Networks[9], described as "joined-up clinical leadership comprised of diverse expert voices to drive system shifts through development of national standards and models of care". They will involve hospital and primary care experts from across professional disciplines working with consumers and whānau, to influence how we prioritise and drive system change with a focus on identifying ways to address variation in service quality and outcomes, addressing equity, and developing innovative, efficient, and evidence-based solutions that will inform investments and workforce planning and be applied nationally. RDNZ hopes that on the back of the RDS rare disorders will feature in the next programme of network development, which would be consistent with our current advocacy programme in support of a rare disorders centre of excellence

- Progress in implementing the government's Health Research Strategy[10] including the recommendations of the July 2022 Enhancing Aotearoa New Zealand Clinical Trials report[11]. One of the key deficits associated with the health sector's inability to support people with rare disorders to have better, healthier futures is lack of knowledge. A New Zealand rare disorders research programme which draws upon research programmes internationally will give the health sector the means to develop best practice knowledge and apply it appropriately in New Zealand clinical, te ao Māori and other settings

- The Ministry of Education Highest Support Needs Review[12]. In November 2022 the Education Minister announced that the Ministry of Education was to work alongside Whaikaha to design and develop a new system and report back to the Minister mid-2023[13]. Although the report to the Minister is apparently yet to be released the Ministry of Education has developed a change programme which the RDS could build on

- The body of health information work being undertaken by Hira[14], which encompasses the SNOMED CT National Release Centre[15]. This is significant

[9] National Clinical Networks. Te Whatu Ora. https://www.tewhatuora.govt.nz/whats-happening/what-to-expect/national-clinical-networks/
[10] New Zealand Health Research Strategy 2017-2027. Ministry of Health. June 2017. https://www.health.govt.nz/system/files/documents/publications/nz-health-research-strategy-jun17.pdf
[11] Enhancing Aotearoa New Zealand Clinical Trials. Liggins Institute. July 2022. https://cdn.auckland.ac.nz/assets/liggins/docs/HP8537%20-%20LIG_Clinical%20Trials_FINAL_v6.pdf
[12] Highest Support Needs Change Programme. Ministry of Education. https://www.education.govt.nz/our-work/changes-in-education/highest-support-needs-change-programme/
[13] New model to better support kids with the highest needs. Associate Minister of Education. November 2022. https://www.beehive.govt.nz/release/new-model-better-support-kids-highest-needs
[14] https://www.tewhatuora.govt.nz/our-health-system/digital-health/hira-connecting-health-information/#what-is-hira
[15] https://www.tewhatuora.govt.nz/our-health-system/digital-health/snomed-ct-national-release-centre/

because it creates opportunities for collecting New Zealand data on the prevalence and incidence of rare disorders specifically and collectively, thus providing important information to the health and other sectors as to which and how services should be delivered. Further elaboration is provided in Appendix I

- The July 2023 Australian recommendations for a national approach to rare disorders data.[16] The report states that a nationally coordinated and systemic approach to the collection and use of rare disorders (RD) data, including registries, is a key priority of the Australian Government's National Strategic Action Plan for Rare Diseases and recommends how this priority can be best addressed. It explores the landscape of Australian rare disease registries (RDR) and databases and responds to the United Nations Resolution on 'Addressing the Challenges of Persons Living with a Rare Disease and their Families'. Specifically, point 5 encourages Member States to: 'Collect, analyse and disseminate disaggregated data on persons living with a rare disease, including by income, sex, age, race, ethnicity, migration status, disability, geographical location and other characteristics relevant in national contexts, where applicable, to identify discrimination and to assess progress towards the improvement of the status of persons living with a rare disease.' Like Australia, rare disorders are not routinely counted or recorded in New Zealand and we rely on extrapolations from international data sets. "To ensure evidenced-based planning and an accurate contextual understanding of the economic burden of RD, it is necessary to systematically count RD. This can be achieved by adopting national routine coding of RD patients at the point of care and extracting data from existing registries and health records to capture existing patients". RDNZ recommends that the Australian report be considered in full in the development of the New Zealand RDS

- Emerging developments in the fields of language and image based artificial intelligence models which could assist with more rapid diagnosis of unusual co-presenting undiagnosed symptoms

- Professor Karen McBride-Henry and Dr Tara Officer, Research Trust of Victoria University of Wellington recently HRC funded research project "Invisible inequity: Healthcare insights from people with rare disorders"

- Manatū Hauora's report on precision health, exploring opportunities and challenges to predict, prevent, diagnose, and treat health needs more precisely in Aotearoa New Zealand[17]

[16] Recommendations for a National Approach to Rare Disease Data. Findings from an Audit of Australian Rare Disease Registries. Rare Voices Australia and Monash University. July 2023. https://rarevoices.org.au/wp-content/uploads/2023/08/RecommendationsforaNationalApproach_RareDiseaseData_August2023.pdf

[17] Precision health: exploring opportunities and challenges to predict, prevent, diagnose, and treat health needs more precisely in Aotearoa New Zealand. Manatū Hauora. August 2023. Precision health: exploring

- Development of rare disorders strategies in other comparable countries.

RDS focus areas

RDNZ supports the inclusion of all of the focus areas identified in the RDS consultation draft. However, in terms of the importance for achieving pae ora for people and whānau with rare disorders we would prefer critical health services, disability and whānau support and coordination, and funding, assessment and prioritisation to appear at the top of the list as these are the things that most directly and immediately impact on the lives of people living with rare disorders.

We further recommend that RDNZ's seven priority areas (which the draft RDS lists under another heading) and proposals for a centre of excellence are included in this section, as set out in our concluding recommendations below.

We note that many of the groups included information in their feedback about what is important to them that the strategy and any subsequent implementation or action plan achieves. We expect that this information, along with responses from the May 2023 survey RDNZ carried out will be utilised as the focus areas are expanded.

Important possibilities not yet identified

Although the consultation draft referred to the pae ora strategies, given that they were yet to be released there was no substantial elaboration. Now that they have been released RDNZ would expect that the RDS would leverage off these where appropriate, and we have made a number of recommendations for their inclusion in the next section.

Recommendations

RDNZ recommends that the RDS includes the following content:

1. DIAGNOSIS

Health entities will ensure early and accurate diagnosis of rare disorders through:

- Equitable access to a range of diagnostic tools/tests, supported by policy.
- A newborn screening program which is robust and funded to keep up with international best practices and availability of new medicines.
- Providing a pathway for carrier testing. This involves testing people who are known to be at increased risk of being carriers of a specific inherited disorder. This may be because a relative is known to be a carrier or has the condition or certain genetic conditions might be more prevalent in their community.

opportunities and challenges to predict, prevent, diagnose, and treat health needs more precisely in Aotearoa New Zealand. | Ministry of Health NZ

- Providing a pathway for genetic testing once a patent presents with undiagnosed symptoms.
- Putting protocols in place to identify people with no diagnosis, ensuring that a lack of diagnosis does not create a barrier to treatment.
- Making high-quality diagnostic tests accessible through common, clinically agreed systems or pathways.
- Developing policy that supports timely and equitable access to new and emerging health technologies.
- Equitable access to peri-conception genetic testing and counselling for those with an increased chance of being carriers of rare disorders

2. PLANNED PATHWAYS FOR CLINICAL CARE

Health entities will deliver co-ordinated and integrated pathways for cohesive healthcare by

- Developing a care pathway for rare disorders including diagnosis and genetic testing for people with unusual co-presenting undiagnosed symptoms.
- Developing standards of care documents for specific rare disorders to be implemented within the health system.
- Delivering services which seamlessly support people living with a rare disorder through life-stage transitions, including from childhood to adolescence, and on to adulthood and older age
- Targeting awareness and education for people in their preparation for conception and pregnancy.
- Identifying people with unusual co-presenting undiagnosed symptoms for priority access to a specialised diagnostic response.
- Coordinating rare disorder care and support that is integrated, while being person and family-centred.
- Implementing clearly coordinated pathways throughout health, disability and other systems.
- Ensuring that rare disorder care and support systems address mental health and wellbeing.
- Promulgation of guidelines that address the specific needs of people unusual co-presenting undiagnosed symptoms

3. ACCESS TO DISABILITY AND SOCIAL SUPPORTS

Health and other government entities will implement mechanisms to ensure appropriate access to disability and social supports by;

- Ensuring those with unusual co-presenting undiagnosed symptoms are included.
- Developing an easily accessible pathway to information on support services available to those with a rare disorder.
- Developing a pathway for those with a rare disorder within the education system.
- Developing a pathway for those with a rare disorder leaving the education system.
- Removing the administrative burden of proving the disorder is ongoing for conditions that are lifelong.
- Ensuring that people with rare disorders that include both health conditions and disabilities and/or hidden disabilities are able to access support that is comparable to people who have an isolated or more visible disability.

4. RARE DISORDER MEDICINES

Health entities will provide equitable access to modern rare disorder medicines through a specific assessment pathway including:

- Future-proofing a pathway for new and innovative modern medicine for those with a rare disorder, for example gene therapy.
- Updating the factors considered in cost-benefit analyses to include the wider health system, social support system, and society costs and benefits.
- Funding PHARMAC so that it is able to fund new and innovative modern medicines in a way that is consistent with best international practice
- Developing and implementing a medicines strategy to include rare disorders, gene therapy and innovative modern medicine.
- Developing and implementing policy that supports timely and equitable access to new, emerging and best available health technologies.
- Ensure people with a rare disorder have equitable access to medicines with demonstrated clinical benefit for a rare disorder,

5. RESEARCH

Health and other government entities will deliver a coordinated and funded programme of research for rare disorders including:

- Development and implementation of a national research strategy for rare disorders
- Development and implementation of a pathway for those with a rare disorder to participate in both national and international research.

12

- Robust investment in rare disorder research in New Zealand
- Translation of research and innovation into clinical care and vice versa so that clinical care informs research and innovation.
- Partnerships between researchers and clinicians in research into rare disorders.
- Clinical teams' collection and recording of data, contributing to research and evidence-building.
- Involvement of Māori and Pacific peoples at every level of development, implementation and governance of genomic research as per *Te Mata Ira*[18] guidelines.
- Development and implementation of policy which maintains sovereignty for Māori over their health data to ensure that Māori and iwi aspirations are realised.

6. NATIONAL RARE DISORDER REGISTRY (DATA COLLECTION)

Health, disability, education and other government entities will capture relevant data on rare disorders in New Zealand. This will include.

- embedding **Orphanet Coding (ORPHACODES)** or an internationally recognised equivalent into the health system. This can/should be done in a way that is compatible with other key coding systems (e.g. SNOMED, ICD-10/11)
- Supporting the availability of computerised prompts to help GPs diagnose a rare disorder when a rare disorder has not previously been considered.
- Developing rare disorders registries and ensuring that they mirror the ethnic demographics of Aotearoa so that they are generalisable and can aid in the diagnosis of Māori, Asian and Pacific peoples.

7. WORKFORCE DEVELOPMENT

Health, disability, education and other government entities will plan and provide training on rare disorders for health and other professionals and support staff which will include:

- Creation of effective rare disorder clinical networks that connect to international research and best practice.
- Developing and implementing pathways to work with international partners.

[18] Te Mata Ira Guidelines for Genomic Research with Māori. Te Mata Hautū Taketake – Māori & Indigenous Governance Centre University of Waikato. October 2016. https://www.waikato.ac.nz/__data/assets/pdf_file/0018/321534/Te-Mata-Ira-Genome-Research-Guidelines.pdf

- Identification of existing gaps in the rare disorder workforce and development and implementation of a national rare disorder workforce strategy.
- Equipping and encouraging frontline health and other professionals to consider, investigate and refer for a potential rare disorder diagnosis.
- Embedding rare disorders in medical training, including diagnosis pathways and undiagnosed ones.
- Promoting to healthcare professionals the importance of 'thinking rare' when presenting with symptoms.
- Promoting to healthcare professionals the importance of taking the whole picture of a condition into account when seeing different specialists
- Strengthening healthcare professionals understanding of tikanga Māori especially pertaining to whakapapa, human tissues and genetic material.

RDNZ notes that the Provisional Health of Disabled People Strategy (PHDPS) uniquely makes a number of references to rare disorders, and we therefore recommend that the RDS includes the following directly paraphrased PHDPS content:

- There must be clearer pathways to accessing specialist diagnoses for rare disorders and for adults accessing diagnoses for rare disorders normally diagnosed during childhood. Waiting for a diagnosis can delay access to other support services, which is detrimental to overall health and wellbeing. This is particularly the case when a health care diagnosis can strengthen an application for funding for support in a non-health related area (page 17)
- To address gaps in the health system's ability to meet the health and wellbeing needs of people with rare disorders in Aotearoa New Zealand … there must be a robust health system data and evidence base that identifies their health and wellbeing needs, and tailors support accordingly (page 20)
- Health entities need to … invest in identifying unmet need, early diagnosis and intervention (page 30)
- Health entities need to … partner with people living with rare disorders and their whānau to collect robust, meaningful, timely and accurate data to be able to determine their health and wellbeing needs and plan and monitor system performance. This includes commissioning and supporting the creation and maintenance of data frameworks and regularly reporting on accurate disaggregated data. Disaggregated data that maps to health data for rare disorders will increase opportunities for service improvement (page 41)

As a member of New Zealand's Carers Alliance RDNZ advocates for improved support for carers of people with rare disorders, and supports the recommendations of the

'The State of Caring in Aotearoa' report[19] (see precis in Appendix J). We note therefore the commentary in the Women's Health Strategy[20] on unpaid carers, which is directly applicable to the unpaid carers who support people with rare disorders, and which should in turn therefore be directly reflected in the RDS:

- "Unpaid carers are people who care for friends, family, whānau, and aiga members with a disability, health condition or illness who need help with everyday living. Women make up 63% of unpaid carers, with older women as the largest group. Unpaid carers are less likely to be partnered and more likely to be sole parents. Younger carers are more likely to be Māori and Pacific. Caring is associated with reduced health and wellbeing and has an economic cost, including loss of income and lost opportunities to participate in education (Ministry of Social Development 2019)" (p 23)

- In the future… system complexity will be "reduced for women who are caring and navigating different services on behalf of whānau with complex needs [including those with rare disorders]. For carers, this looks like receiving better, more coordinated support from the health and social systems, as well as feeling less stressed and more connected to their communities…this means that health entities will need to provide better support for carers, including improving accessibility of respite care. They will also need to partner with other agencies, including ACC, Whaikaha | the Ministry of Disabled People, and the Ministry of Social Development to make it easier for carers to navigate on behalf of those they care for" (pp 39,40)

Rare Disorders Centre of Excellence

As we look ahead to the implementation of the National Rare Disorders Strategy and how resources available across the health sector can be used in efforts to improve health system responsiveness for people and whanau with rare disorders as soon as possible, Rare Disorders NZ has identified a key opportunity for maximum leverage and progress for rare disorders through the establishment of a Rare Disorders Centre of Excellence in New Zealand. In a number of other countries 'Rare Disease Centres of Excellence' have been established that act as hubs of expertise and promote best practice, bringing together experts from multiple specialities to reduce the time to diagnosis and improve the availability and coordination of multi-specialty clinical care.

Strong support for a Rare Disorder Centre of Excellence was identified in the previously supplied May 2023 survey of RDNZ support group leads regarding what

[19] The State of Caring in Aotearoa. Carers NZ and the Carers Alliance. August 2022. https://carers.net.nz/wp-content/uploads/2022/07/State-of-Caring-Report-Aug2022.pdf
[20] Women's Health Strategy. Manatū Hauora. July 2023. https://www.health.govt.nz/system/files/documents/publications/womens-health-strategy-jul23.pdf

they would love to see change in New Zealand for people with rare disorders, as well as what the biggest barriers people with the disorder and/or their carers are facing.

RDNZ commends this approach as a key enabler which will realise the aspirations of the RDS.

16

Appendix A

Summary of feedback from support group leads on 'A rare disorders strategy for Aotearoa New Zealand, Initial proposed content and points to consider and provide feedback on', as at 31 July 2023

The support group leads were appreciative of the opportunity to give feedback, however, some raised concern and frustration. This centred around not having been involved earlier, about the limitations of email only, and about how genuine the consultation was, with comparison to it being a tick box exercise to say it had been carried out.

A strong desire to have more than just 'a voice' and to be included in a co-design process was communicated by multiple groups.

Many groups also requested more transparency in what the process for developing the strategy will involve, including around the following:

- What the next steps are
- What other agencies are involved in the strategy development
- What expert engagement is being undertaken
- What the schedule for the preparation of the strategy is
- What the planned timeframes for deliverables are.

It was raised that it is premature to decide upon the aims and focus areas prior to undertaking genuine co-design processes and lived experience expert consultation, as well as assessment of the current situation.

There were suggestions to incorporate and acknowledge the United Nations Convention on the Rights of Persons with Disabilities (UNCRPD), Māori models of health, the social model of disability, and to consider looking at strategy development through the five I's framework.

Feedback in response to the questions posed by the Ministry has been summarised under the subheadings below.

• Are there additional points that are important to capture?

o In the background (why have a rare disorders strategy?)

Groups raised that the background material was quite vague and that they would like to see it include a larger evidence base.

It was noted that 'pae ora' on its own is a meaningless term for many with a rare disorder, and that a definition of what pae ora means specifically for those with a rare disorder is needed.

It was requested that there is an analysis of gaps and shortfalls in the current system. There was concern that without understanding our baseline it is difficult to know where the focus needs to lie, as well as how to measure progress.

There was concern that there was not a section that discussed previous reviews and work streams that have made recommendations in respect of rare disorders, in particular the recent Pharmac review, so that these shortcomings are captured and brought through to the focus areas to ensure those issues are being addressed.

It was also raised that there is very little commentary or evidence presented regarding how New Zealand currently compares internationally in relation to rare disorders care and outcomes,

especially against those countries that have implemented strategies and action plans for rare disorders.

There was general agreement that it is important to improve equity of outcomes within the rare disorders community, with many noting regional variability in the availability of treatment and services.

Groups sought recognition that not getting it right for people with rare disorders has a wider societal impact, including a large social and economic impact. It was noted that an absence of equitable and clear pathways and processes for rare disorders diagnosis and management can result in high costs to the health system in the long term. It was highlighted that missing opportunities for early intervention and best-practice care can result in people who are less productive in society, and that this has a spill-on effect to their carers and entire whānau, which is often overlooked and not measured.

o In the proposed definition (what are rare disorders?)

Feedback was generally supportive of the definition with some groups suggesting including a definition of 'ultra-rare' of 1 in 50,000.

• Do you agree with the key elements (aims and principles) proposed for a strategy?

Groups supported the key elements (aims and principles), noting that many found it very general and vague.

There was a lot of concern about the statement "the strategy will not do the work or identify specific actions or commitments in focus areas."

It was asked that the strategy identify the gaps in current programmes and activities and the actions to be taken to address those gaps. It was noted the strategy needs to be explicit regarding the parties responsible for addressing those gaps and timing for that, as well as how progress will be measured and how agencies will be held accountable for delivery.

It was emphasised that the strategy must provide sufficient detail to enable agencies to be clear about their roles and responsibilities, for progress to be measured (and how this will be done), and accountability for delivery to be determined. Our groups expect to see real action and measurable progress on the ground for people with rare disorders, with agencies held accountable for delivery of their aspects of the strategy.

To address these concerns, Rare Disorders NZ asks an action plan or implementation plan accompanies the strategy. We would like to see the plans for the creation of this, including a clear timeframe for delivery and accountability for progress, incorporated in the strategy.

• Are there additional opportunities that could be built on? (doing the work to develop a strategy)

It was suggested that the strategy builds on the findings and recommendations of the Pharmac Review published in 2022.

It was noted that expert clinicians are already working in the New Zealand health system and that they should be brought together to create a centre of excellence whether in person or virtually.

Expansions to the point “progress being made internationally, such as through clinical and research collaborations and virtual specialist networks” were suggested. It was emphasised that we should be building on work from other countries and reviewing rare disorder strategies and action plans that exist internationally, analysing what is working well and what isn’t.

It was noted that many areas of research are widespread internationally and there are several multi-site international studies that take place. Establishing links with organisations and networks carrying out this work would enable our clinicians to access knowledge, promote trial participation and to enable evaluation of effectiveness for Māori and Pacific populations.

There are many large organisations internationally who have well established databases and guides for Rare Disorders that can be useful and guide medical professionals here in their responses to their patients.

Rare disorder support groups hold knowledge of patient experience and have awareness of international practice. Many of the groups noted they have international contacts and relationships with their counterparts and experts in their disorder internationally. They asked to be involved in the development process of standards of care guidelines for New Zealand. Some groups already hold resources that can be utilised more widely and have their own registries.

• Focus areas

It was expressed that the ease of making progress, barriers and timeframes would largely depend on the resources allocated and decisions made by government.

There was a lot of scepticism regarding the statement “In New Zealand, we have good assessment and prioritisation of individual products (such as medicines and medical devices) and certain services (such as surgical points systems), and some examples of integrated assessment of treatments, supports, products and services (such as for child cancer)”, with many groups noting that New Zealand’s international ranking for access to modern medicine is very poor.

We note that many of the groups included information in their feedback about what is important to them that the strategy and any subsequent implementation or action plan achieves. This area of the feedback aligns with Rare Disorder New Zealand’s seven priorities to improve health and wellbeing for people living with a rare disorder and we would like to see these more specifically incorporated in the focus areas.

This feedback has been added to the answers, themed by priority area, from Support Group Leads to the May 2023 survey hosted by Rare Disorders NZ regarding what is going well, what the perceived barriers are and things that the groups wished for. This information is presented in the document titled ‘Rare Disorder NZ’s Support Group Leads’ summarised comments on what is going well, what the barriers are, and what they wish to change for rare disorder care in New Zealand, grouped by Rare Disorder NZ priority area’. The additions from the feedback to the Ministry of Health’s consultation document are in blue.

• What important possibilities are not yet identified?

We noted the following areas were repeatedly discussed throughout the feedback from our groups:

- Development of a centre/centres of excellence. Rare Disorders NZ has attached further information regarding this.
- International collaboration including the development or joining of international virtual networks for health professionals, and for research- including clinical trials.

- Support groups are an under utilised resource- Support groups would like to see referral to them built into care pathways so everyone with the disorder has the opportunity to engage with them.
- Interim measures- Not only do people living with rare disorders need healthy futures, they also need health, wellbeing, inclusivity, and adequate care, now. The overall timeframe the strategy is working to should not be a barrier to pressing rapidly in any quarters where improvements in access to care and treatment could be made right now.
- A whole of life approach – Groups emphasised that timely and accurate diagnosis is important but only the beginning. They are asking for a holistic coordinated approach that:
    - recognises the role and responsibilities of family/whānau, including into adulthood
    - foresees and addresses the implications for education and employment
    - includes support at times of transition e.g. from paediatric to adult care
    - is proactive and considers appropriately scheduled reviews and recalls as well as screening for known complications.
- Funding- The strategy and subsequent action/implementation plan must be well resourced so that its implementation can be expedited. We expect to see discussion on how and when it is anticipated funding and resources will be allocated to support the implementation of the strategy.

Appendix B

Responses from Rare Disorders Community on Rare Disorder Strategy

Support group name	Response
Head and Neck Cancer Support Network NZ	Thank you for the opportunity to give feedback on the RDS. I think it is fine as is. There are areas of concern, including developing a workforce that is Rare Disorders capable, and managing the patient experience, or the workflow for those with RD. I think they can wait for an implementation plan rather than a strategy.
ANE International	I note that this document includes approximately 20%/2 pages relating to Treaty of Waitangi. Rare disorders are not specific to race and it should go without saying that everyone gets opportunity for the same level of treatment. One thing in our experience – not everyone fits into 'a box.' We have battled for years to find support that meets our children's needs – we firmly believe the majority of parents/caregivers simply give up. Our children have a very rare genetic condition and require a custom tailored support package. The majority of those truly battling with sick children/serious conditions simply do not have the time, energy or resources to seek the help they want and need. And definitely not to join 'extra-curricular' groups/organisations.
ANCA vasculitis NZ and Scleroderma NZ	**Definition of rare disorders** Scleroderma and ANCA associated vasculitis and many other rare disorders are clearly rarer than 1 in 2,000. The prevalence of disorders are not precisely known. Some disorders have geographical differences in prevalence. There may be some where it is not clear if they affect fewer than or equal to 1 in 2000 persons. This definition suggests a precision that is not obtainable. We do not have a good suggestion to address this. Adding a word before 'affects' could be considered, such as plausible or could. The uncertainty in prevalence estimates could be expressed with 95% confidence intervals. Would a disorder with a prevalence estimate of 1.05 in 2,000 persons with a 95% confidence interval of 0.8 to 1.3 be considered a rare disorder? Although the best estimate of its prevalence is more than 1 in 2,000, it is reasonable plausible that the prevalence could be less. **Rare disorder medicines** A pharmaceutical company was not applying for Medsafe approval for a drug for a rare disease. Would this issue be addressed by "Rare disorder medicines – equitable access to modern rare disorder medicines through a specific assessment pathway"?
Rett NZ	Thank you for the opportunity to submit to your strategy for Aotearoa families with Rare Disorders. 1.A Brief History Of Working together with and For Each other. Twenty two years ago about ten of the groups with Rare Disorders combined to give evidence at the Royal Commission on Genetic Modification in New Zealand. Each group had been in their own silos working through the pre internet search for information. We agreed to share any information which would be useful to other groups and support each other in their needs for clear and early diagnosis and access to treatment and education. We have learnt from other groups and the internet has been a powerful tool in dissemination of information.

	2.Collection of Accurate Data Collection of data has been a major problem. If a patient is in hospital the data is collected however, it is not collected from the first point of contact the General Practitioner. For good policy making decisions this data is imperative to reach those in the wider community. Auckland has most of the numbers because of its population, it is difficult for families in Gore or Tokoroa. 3.Development of Clinical Networks This was a dream for many years however, we do understand the cost to cope with the geographic spread. The Disorders with higher numbers have benefited from these networks. RettNZ 's ad hoc solution has been to have biennial conferences (Delayed by covid and what the effect would be on fragile bodies). We have engaged expert speakers and kindly Paediatric neurologists to assess and advise. 4.Early and free Diagnosis Recently one of our families needed to ask for genetic testing for several months for a specialist to undertake genetic testing for Rett. Their daughter's diagnosis was delayed, and the family had to pay for it. We had understood genetic testing to be free and it should be free. Unless you have been in this situation you do not realise the wated hours wondering what is happening with a degenerative condition and what is the right course of action. The alleviation of guilt. This family wanted to have another child which they have since had and we are all enjoying. It was a sad and unnecessary delay. 5. Educational Opportunities With children with multiple disabilities and often severe seizures schooling can be a problem, it would be good to have approved Medical Websites for Nurses Teachers and Caregivers on subjects such as dealing with anxiety and seizures. The great advance in the last twelve years has been the girls with Retts have been able to use Tobii machines for eye pointing for communication in writing and reading. It has been liberating for the girls and their parents and teachers and the Doctors who now believe they do understand, however, their neuronal impulses cause delays in expressive movements 6 Access to Drug Therapy . A solution from a brilliant Medicinal Chemist Distinguished Prof Dame Margaret Brimble of Auckland University. Dreaming of being able to alleviate some of the traumatic symptoms in Rett has seemed a long way off. However, on the 12 March 2023 the FDA in the USA gave permission for Daybue to be used on those with Rett from 2 years old to be used in the USA. WE are following the USA individual parent stories which show a recovery of some of the milestones which were lost during the periods of regression. Of course we are dreaming for a time when Daybue will be available in Aotearoa. It would appear from the Clinical studies the earlier the drug is obtained the more efficacy it has. On Friday 14.07.23 it was agreed with the parent company Neuren that their US partner Arcadia would now be responsible for distribution to the rest of the world. We are waiting to see when this may apply in Australia and New Zealand. What a wonderful story it would be if those that suffered from Rett were able to be helped by this molecule carefully and lovingly created by a brilliant NZ Woman Scientist. Margaret's achievement is being lauded by Prestigious Chemistry Societies all over the world. Some of the awards are of almost Nobel Prize equivalents. Margaret's drug was intended for TBI and became successful with Rett.

	As soon as Neuron announces their strategy ,we are optimistic that this world leading event will help those with Rett in Aotearoa. 7. Thank you for including the Groups with Rare Disorders in your study. It means so much since working alone, then in groups and then Together.
FACSNZ (Foetal Anti-Convulsant Syndrome NZ)	Tēnā koutou, RE: Rare Disorders Strategy for Aotearoa New Zealand Thank you for sending through the draft document. FACSNZ are disappointed at the fact that we only had a week turnaround period, which is over a long weekend, celebrating Matariki. This has resulted in us not being able to collaborate a response from our FACS community, particularly when it is also school holidays and a lot of our community have tamariki still at school. So FACSNZ are submitting our feedback acknowledging this limitation, and our limited response as we too have had prior commitments. Some general comments we would like to provide prior to delving into the particulars of the document are that there has been no acknowledgement or mention of the United Nations Convention on the Rights of Persons with Disabilities (UNCRPD). If we look at the definition of the social model of UNCRPD we see that "Persons with disabilities include those who have long-term physical, mental, intellectual or sensory impairments which in interaction with various barriers may hinder their full and effective participation in society on an equal basis with others."1 Just by this social definition alone, the rare disorders community do have a disability, however they may not chose to identify as having a disability, and that is their right. Another general comment is that we would suggest that the use of the Five I's Framework, an holistic and integrated approach to health care with ethics at its heart2 should be pivotal to the structure of the Rare Disorders strategy. The Five I's Framework was designed by the Health Consumer Advocacy Alliance. "The Five 'I's Framework … is an holistic and integrated approach to health care with ethics at its heart. The core tenets are: **Interdisciplinary:** Medical professionals from a range of disciplines working together with the aim of organising and co-ordinating health treatments and care services. Providing an environment that supports co-operation, respect and collaboration between colleagues and health practitioners to provide seamless, continuous care. **Integrated:** A healthcare approach that takes account of the whole person, including the social determinants of health, their past, present and future. Holistic practice that considers the interconnectedness and interrelationship of the different parts of the body as well as mental, spiritual and physical wellbeing, while utilising all appropriate therapies. **Interrelated:** Building and fostering relationships with team members. Valuing and validating personal and individual strengths to achieve desirable patient outcomes,

	effective communication and provision of quality care. Information sharing across the team and with consumers/patients. **Intergenerational:** Actively promoting, valuing, fostering and encouraging intergenerational communication with patients/consumers and whānau. Understanding the impact of intergenerational trauma, adopting a trauma informed approach to health care. **Inclusive:** Validating identity, diversity and culture. Being aware of unconscious bias. Ensuring accessibility for all to a range of community services that support mental health and wellbeing. Meeting the needs of individual consumers/patients and whānau, providing patient centred care." "The Five 'I's Framework is applicable to all health care settings and services. In practice the Five 'I's Framework involves: • Providing an environment that supports co-operation, open discussion and transparency, and respect between colleagues and health professionals. Problems should be challenged, and issues should be navigated in a safe space, in a respectful way. If for any reason this is not possible, outside help should be sought to ensure that problems are resolved in this manner. • Questioning if there are specific barriers for consumers seeking help within a practitioner's own practice. Gaining an understanding of the impact of intergenerational trauma to better understand how unconscious bias can affect provision of treatment and health care services, by choosing to actively listen and learn from patients and their communities. • Seeing and treating the patient in a holistic manner, as a 'whole' person rather than the health problem they present with, as opposed to the prevailing reductionist approach, where the complexity of individual health needs can be ignored. • Learning how others embrace mental, spiritual, and physical well-being in treatment, so their patients and their whānau are at the centre of care. Creating opportunities for staff so they are aware of, recognise, and acknowledge the holistic approach, and the importance that reflection has in changing practice to improve healthcare outcomes. This framework aligns with four Māori models of health, Te Whare Tapa Whā, Meihana model, Te Pae Māhutonga and Te Wheke model, which underpin an holistic approach to healthcare and encompass spirituality as a key element and core foundation. For many Māori, the major deficiency in modern health services is taha wairau, the spiritual dimension; none of the elements illustrated within these models can be separated from the socioeconomic circumstances and social determinants that lead to such significant disparities in health. By understanding our health system's flaws and inadequacies, and its strengths, we can embrace a more holistic approach to care. We can learn a lot from looking at health care through a different lens, such as the Māori models of health and the Five 'I's Framework. Adopting the Five 'I's Framework and using an interdisciplinary, trauma informed approach to care would be beneficial for both patients and health professionals."

	Why have a rare disorder strategy? In the second paragraph in the sentence "Limited clinical knowledge can lead to…" after the to we would make the suggestion of "not being seen" before continuing to say "misdiagnosis, long wait times for diagnosis…" **Impacts of rare disorders** In the second paragraph, second to last line it states, "While health service utilisation is high…" after the word accessing, we would suggest you add in the words "any and/or" before "sufficient". **Unrecognised and undiagnosed conditions** FACSNZ would suggest the sentence in the first paragraph where it states, "Relatively common conditions, like foetal alcohol spectrum disorder or many neurodiverse conditions, have not been well understood by health practitioners nor been a high focus of research or service improvements until recently." Whilst we appreciate this is a very relative area of discussion, we believe it does not belong in the Rare Disorders strategy, nor have experts by experience in these areas been consulted in regards to this point. **What a strategy will provide** When it says "The strategy is being developed in partnership with health entities and Whaikaha, as well as with RDNZ and experts from clinical, science, cultural and lived experience viewpoints." FACSNZ have a few comments. This draft strategy is the very first opportunity we have had to have any discussions into what the strategy might include. When the old CEO of RDNZ was first in discussions with Manatū Hauora there were a number of rare disorder community leads saying that they wanted to be part of the co-design as they felt a range of people with experience viewpoints, needed to be involved in the co-design. We are experts by experience and we should be around the table. The world partnership is being used, except it should be co-design. Using this word and the meaning behind it is more in alignment with Te Tāhū Hauora, Te tikanga mō te mahi tahi a ngā hinonga hauora ki ngā kiritaki me ngā whānau | Code of expectations for health entities' engagement with consumers and whānau, and the World Health Organization, Global Patient Safety Action Plan 2021-2030. How is Whaikaha being engaged, as when we have had feedback from RDNZ about engagement with Whaikaha they have indicated none – limited. **Doing the work to develop a strategy** In the second dot point it says "…including gathering evidence…". We would suggest using a different word instead of evidence as it can have negative connotations, so we recommend something such as information. Under dot point 8 FACSNZ would recommend that at the end of the sentence an addition of, as set out in the Five I's Framework. We would recommend an additional dot point that would be: • Utilising the Te tikanga mō te mahi tahi a ngā hinonga hauora ki ngā kiritaki me ngā whānau | Code of expectations for health entities' engagement with consumers and whānau, and the World Health Organization, Global Patient Safety Action Plan 2021-2030.

	Capturing voices of the rare disorders community We would like to suggest additional recommendations such as: • National Centre of Excellence, that has the inclusion of rare disorders. • National registry of rare disorders. • Data collection. • Equitable outcome for all. • Specialist centre where you have access to the right specialists at the right time using the Five 5's Framework. • Carer support. • Disability access. • A holistic approach that includes every point of a rare disorder person, whānau may have interaction e.g. health, education, disability, and local, tertiary and secondary health settings, to name a few. **Identifying focus areas for the strategy** "The strategy will not do the work or identify specific actions or commitments in focus areas." This statement is alarming because if there is no commitments then how can there be changes? **Digital solutions and enabling** There is a huge problem in Aotearoa New Zealand with digital as we have different electronic systems and they do not all talk to one another. Without resolving this it is like putting band aids on a broken arm, no significant difference will come, and it will only make matters worse. The second dot point, sorry we do not even know what this means. **Critical health services** Implementation of the Five 5's Framework is pivotal here. At the end of the first paragraph where a list is being given to some of the specialist areas, we recommend the addition of psychiatrist. **Disability and whānau support and coordination** Spoke of the UNCRPD at the beginning of this submission/feedback. The last sentence of this section we recommend the addition of people with disabilities as well. Some of the carers who are trying to engage in this journey are disabled themselves, which makes equity extremely difficult. **Knowledge building** We recommend an additional dot point: • The whole of Aotearoa New Zealand **Funding, assessment and prioritisation**

	All of the rare disorders should be prioritised as we all have disabilities, irrespective of whether or not we identify as having one, and as such we should be receiving equitable care, which we know we aren't. The second paragraph concerns FACSNZ a lot. Where it states "In New Zealand, we have good assessment and prioritisation of individual products (such as medicines and medical devices)…". This simply is not true. FACSNZ might be one of the medicine groups the strategy has referred to as Fetal Anticonvulsant Syndrome (FACS) is caused by exposure to anti-seizure medicines during pregnancy which can result in being permanently harmed and in some cases death. Everything that has been achieved in this space has been because of the hard work, determination, and passion of FACSNZ. We have collaborated at an international, national, local, and individual level to achieve what has been done. There is definitely not any assessment, or prioritisation going on with FACS in Aotearoa New Zealand unless our tamariki are going through the ACC system, in which some cases it can take up to 4 years to have a decision made. As for medical devices again like the mention of Foetal Alcohol Spectrum Disorder earlier, unless there has been experts by experience involved with this rare disorder strategy, which they should not be, there is no relevance for this comment. Our Executive Officer knows personally some extremely knowledgeable people with mesh injuries who would not agree with that statement above. We recommend the removal of this whole paragraph, or list specifically the medicines you are referring to, assuring that an expert by experience has been consulted with as well. **Research and evaluation** In the second paragraph where it states "…especially for responsiveness to Māori and Pacific populations…" we would recommend the addition of disabled in this sentence as well.
Waldenström's Macroglobulinemia patient support group.	Thank you for the opportunity to submit feedback on the initial draft content and points for the draft New Zealand Rare Disorders Strategy. We are pleased to see Manatū Hauora is leading the development of a Rare Disorders Strategy in response to the recent recommendations from the 2022 independent Pharmac review and ongoing advocacy work of Rare Disorders New Zealand (RDNZ), groups and individuals. The incorporation of the voices and recommendations from RDNZ, groups and individuals will aide in the strength of the strategy. We understand strategy development and implementation is a long process. While Manatū Hauora have noted that the strategy is expected to be available within the next 12 months, we acknowledge that the successful implementation of strategy objectives is a further long term process. We therefore want to use this opportunity to advocate for urgent interim support for the unmet need of people in Aotearoa living with rare disorders. People with rare disorders have been unjustly disadvantaged and require additional support and resources now, concurrently to strategy development and implementation. We understand that this request is not standard procedure for populations affected by future strategy development. However just because something is normal, it does not mean it is acceptable. When it comes to rare disorders (and many disorders for that matter) it often feels this normality of funding timelines, limited medicine access, misdiagnosis and living with the fear, uncertainty, marginalisation, and often debilitating symptoms, is so inherent that is accepted and tolerated. It is this that we wish to see challenged and changed through the strategy and with interim support.

	To streamline our feedback, please note when referring to people with rare disorders, this includes people and whānau and discussions regarding medicines also include medical devices. The underlying theme for our feedback surrounds interim support and challenging the status quo. We have offered some brief solutions to our suggestions that we hope the strategy will address, some of which we understand are outside the general scope of what is normal. However, we hope Manatū Hauora will keep an open mind and think outside the box to find solutions to obtain the best outcomes for people living with rare disorders. **Draft Strategy Questions and feedback** **1. Are there additional points that are important to capture?** a) In the background (why have a rare disorders strategy?) This section needs to include current relevant information. Under the Impact of rare disorders section of the consultation draft, the document quotes an Australian 2010 study. From a quick search there is a multitude of more recent and relevant data that can be used here including but not limited to New Zealand information such as Impact of Living with a Rare Disorder in Aotearoa New Zealand (2022) and Rare Disorders Insights Report: Pathways Towards Better Health Outcomes (2023). Providing a powerful compelling New Zealand context is critical to strategy direction, development, acceptance and adherence. For example the statistic that there are more New Zealanders collectively living with rare disorders than there are with diabetes is striking. b) In the proposed definition (what are rare disorders?) The definition is comprehensive and inclusive. The adoption of the universal definition also provides significant benefits for our collective international efforts across all areas from data collection and clinical services to research and evaluation. **2. Do you agree with the key elements (aims and principles) proposed for a strategy?** I note the proposed aims of the strategy are: ○ Pae ora for people and whānau living with rare disorders ○ Pae ora for Māori, Pacific peoples and disabled people living with rare disorders and for those living in rural and other diverse communities We understand this may be implied, however we want to draw attention to the words used. According to the Pae Ora (Healthy Futures) Bill, pae ora is defined as 'healthy futures'. Not only do people living with rare disorders need healthy futures, they also need health, wellbeing, inclusivity, and adequate care, now. This ties in with our underlying feedback for the urgent need of *interim support* to help people with rare disorders, *now*. Regarding the strategy **principles**, we feel these are comprehensive and inclusive. We feel the addition of "seeks out evidence", "health system challenges that come with rarity", "gives voice" and "collaborative" are going to be of particular importance considering the nature of the strategy.

	3. Are there additional opportunities that could be built on? Due to the short timeframe for consultation, we have only provided comment on a few focus areas. However, we agree with the comprehensive list of focus areas and deem them all as important to the achieving of pae ora for people with rare disorders. While we do not have specific comments on current work underway or the length of time to carry out the suggested focus areas, we believe the question of 'how easy it is to make progress' and 'are there significant barriers' are a matter of perspective. Regardless of the initiative, we need to prioritise this and find a way to make it happen. The 'ease' of making progress and the 'barriers that must be overcome' should not themselves become a barrier. We're certain that you will have the support from many organisations, groups, and individuals to assist with strategy development and implementation to help overcome any perceived barriers. This strategy is important to us, and we are willing and happy to collaborate. Unfortunately for people living with rare disorders, we have not had a national strategy to help elevate and prioritise their unmet need. Many people have suffered, physically, mentally, spiritually and socially waiting for the time for when their unmet needs would be acknowledged, addressed, and supported. However, luckily for Manatū Hauora there is a multitude of comprehensive rare disorder strategies from comparable countries (please refer to Appendix). While our strategy must be individualised to our specific needs in Aotearoa, these international strategies will help provide a robust evidence base and strategies to start with. Of particular interest, the countries listed in the appendix are the 19 of the 20 countries (alongside New Zealand) that were analysed in the 2019 International Comparisons of Modern Medicines Report). While there were many interesting insights from the report, one of the conclusions stated: *"All 19 OECD comparator countries publicly funded significantly more modern medicines between 2011-2017 than New Zealand across a wide range of significant health conditions such as cancer, diabetes cardiovascular disease, arthritis and rare diseases. On average these countries were also more efficient (faster) than New Zealand in making these funding decisions. Overall, New Zealand compared very poorly against its peer OECD countries, being ranked in last place out of the 20 comparable countries for access to modern medicines".* The fact these comparable OECD countries have a rare disorder strategy in place may well be a contributor to their greater funding of rare disorder medicines. Additionally the frightful statistics from the Global Access to New Medicines Report (2023), ranking New Zealand the lowest in respect to access to new medicines in the full 38 OECD countries was sobering to see. While these statistics and information highlight the extensive gaps between other OECD countries and Aotearoa in respect to not only the development of a rare disorder strategy, but to funding of medicines, we can view this draft strategy as an exciting opportunity and win for people with rare disorders to soon achieve pae ora in Aotearoa. We agree with the seven priorities that RDNZ have set out to improve health and wellbeing for people living with a rare disorder and feel there is ample opportunity to incorporate these within the focus areas outlined in the draft strategy.

	Focus area: Funding, assessment and prioritisation We urge Manatū Hauora to use this strategy development as an opportunity to review the current practices, strengths, weaknesses and opportunities within Pharmac in relation to medicine access for rare disorders. We have included some specific examples we would like to see investigated / considered within the strategy. We have seen in aforementioned documents the discrepancies in medicines funded for rare disorders compared to more prevalent disorders. The following is an excerpt from the Pharmac rare disorders webpage: *"Suppliers of rare disorders medicines don't need Medsafe approval before applying to Pharmac (Principle 1 notwithstanding). This differs from the funding application process for most other medicines. However, the medicine must be Medsafeapproved before a final decision can be made to fund it. This flexibility enables us to consider funding applications before a Medsafe approves a medicine".* It is pleasing to see Pharmac accept applications for rare disorder medicines that are not Medsafe approved. This is an important step towards equitable outcomes. We are however not sure of the timelines / processes relating to Medsafe approval and fear these may render this above principle invalid. Furthermore the strategy needs to ensure that the likes of Pharmac increase the accessibility of medicines that are not Medsafe approved, if the reason for declining the funding application is based on limited clinical evidence. Considering the rarity of rare disorders, clinical trials and research may be limited. However patients with rare disorders deserve to have the option to access recommended medicines (albeit not Medsafe approved) if it means this provides them with a greater quality or longevity of life, especially if there is no other options available. Is there an opportunity within the strategy to investigate the possibility to have provisional Medsafe approval if Medsafe approval timelines are still limiting and restricting access? Some rare disorders share many commonalities and clinical features with more common disorders. There is an opportunity here to use this information to help inform the suitability of a treatment for said rare disorder and widen medicine indications (on a case by case, temporary and or provisional basis) to increase access. It is evident that there is a significant amount of New Zealanders (and populations throughout the world), rare disorder or not that are accessing generic medicines overseas due to lack of funding and or availability of the medicine back home. While they may take precautions to ensure the medicine they are sourcing is of high quality and comparable in composition to its standard version, this highlights the sheer demand, need, desire, desperation and or hope to improve their quality and longevity of life. People with rare disorders (and all people) should not be out of pocket, nor should they have to navigate uncharted waters of importation of generic medicines in order to achieve pae ora. Our system has simply let them down. On this note, relevant for rare disorder treatment (and general), there is an opportunity for this strategy to allow for the provision of special criteria for rare disorder generic medicines. Pharmac should be supported to investigate the option of funding generic

	medicines from overseas, that are a fraction of the price, and that meet certain criteria (e.g. are legally and ethically sourced, manufactured, tested and distributed; undergo comprehensive testing to confirm composition and performance; and hold relevant current manufacturing certifications such as GMP. Perhaps this is a way in the interim to assist in the accessing of life saving treatment options for people with rare disorders who have been waiting long enough. We are aware this suggestion challenges the status quo. Nevertheless we encourage the health sector to think outside the box for solutions. We also acknowledge this option is not a desirable consideration for the Pharmaceutical companies of the original medicine. Though, if the generic version meets established rigorous criteria set by Medsafe/Pharmac and any other regulatory bodies, why can this not be considered as an interim option (or permanent, if we're being brutally honest) to support people with rare disorders access life saving medicines. Furthermore, we acknowledge the challenges underpinning the funding (or lack thereof) of medicines in Aotearoa is multifaceted and is not a reflection of the many compassionate individuals who work in these funding and regulatory bodies. We acknowledge they are faced with tough decisions, within a limited budget and have an immense task on their hands. The strategy needs to enable / encourage wider investigation, collaboration and negotiation with pharmaceutical companies to offer competitive, affordable and acceptable prices. Pharmaceuticals for rare disorders is an ideal place to start this intensified investigation, collaboration, and negotiation. New Zealanders are disadvantaged due to the current insufficient and inefficient processes and timelines at Pharmac. These timelines are unacceptable. It is our understanding that Pharmac is aware of their shortcomings, and their position in relation to other comparable countries (especially in reference to the aforementioned 2019 International Comparisons of Modern Medicines Report and the Global Access to New Medicines Report (2023). We are glad to hear that Pharmac is addressing this and recommend they devise an interim solution to support access to medicines for people living with rare disorders. **Focus area: Knowledge building** We hope to see the principles of the strategy reflected in the adoption of a direction that enables physicians to have greater collaboration with rare disorder experts. This ties in with all the principles, *notably Supports health practitioners and providers to provide quality care, is informed by and seeks out evidence, is collaborative and built on partnerships and* RDNZs priority of workforce development. One of the issues noted within many reports highlighting the challenges of living with a rare disorder is the limited knowledge / experience of physicians due to the rarity of the disorder. This means physicians are often relying solely on New Zealand clinical practice guidelines, which for rare disorders (if they are available) may not be updated regularly enough to reflect updated international evidence. The revision and updating of rare disorder clinical practice guidelines needs to be included in one of the first stages of strategy implementation. In the interim, and even after this is addressed in the strategy, physicians need to consult with international experts. This cost needs to be covered under the health system. We

	need to remove barriers to physicians seeking expert advice, and ensure they have the resources required to do so. We acknowledge the immense pressure physicians are under. The strategy needs to ensure the provision of resources and incentives for physicians to engage in enhancing rare disorder knowledge. This consultation/collaboration with international rare disorder experts as noted above, if not already considered within the Medical Council of New Zealand's recertification requirements for practicing physicians could be built into and contribute towards a physicians Continuing Professional Development (CPD) Requirements. The strategy talks about 'the future'. We suggest there is a current urgent unmet need and that support in the interim needs to be provided to organisations such as RDNZ until the strategy is not only in place, but the objectives of the strategy have been implemented and met to a satisfactory standard. The support offered through RDNZ helps patients with rare disorders have a voice, access support and increase their chances of accessing expert advice and treatment options. The services that RDNZ provide aligns with the aims of the draft strategy. Experts in the relevant rare disorder (whether local or international) need to be given a seat at the table for all rare disorder Pharmac applications. We appreciate the applications are reviewed by relevant special advisory committees for comment/priority ranking. However in the case of rare disorders, clinical advice from experts in the rare disorder needs to be sought and supported by the strategy. It is imperative that this factor (rare disorder expert consulting) is utilised not only within the Assess and Consult Pharmac application stages, but at the outset at the Review and Advice priority ranking stages. Clinicians, who may be experts in their broad fields for common disorders, are invaluable in providing clinical information from an evidence based and professional perspective (from drawing on the many patients that they see with said conditions) cannot be expected to comment/grade rare disorders which they have not seen/treated and in some cases heard of. Likewise, alongside enhancing consultation with experts, we expect the strategy to support the need for increased consultation with people living with rare disorders at the initial stages in rare disorder Pharmac application assessments, and not left until the latter final consultation stages of the application process. Not only is it important for Pharmac to hear directly from patients, it is important for patients to be able to connect and liaise with others walking in their shoes. The strategy needs to elevate the presence and support of local rare disorder patient support networks. **Focus area: Research and evaluation** It is encouraging to see the increase in international efforts and interest in rare disorders. We are glad to see research collaboration noted as a key area. This collaboration is key, not only in research but in all stages in the prevention, diagnosis, treatment and management of rare disorders in Aotearoa. The strategy needs to consider how to provide incentives for rare disorders research in Aotearoa.

	Concluding comments Overall, we are happy with the initial proposed content for the draft strategy. We feel the aims and principles align with what people with rare disorders want and need. The focus areas are comprehensive and when implemented will complement each other for a synergistic multifaceted approach to enhancing health outcomes for people with rare disorders. While as mentioned, there may be many barriers to achieving what we hope the strategy will achieve, we hope Manatū Hauora is confident and dedicated to think outside the box to creatively find effective solutions to overcome these barriers. Collaboration and unity throughout all stages of strategy development and implementation is key. We hope Manatū Hauora takes our urgent request of interim support seriously and considers how we can get the ball rolling on supporting those with rare disorders while we continue on this long (but exciting) journey to the successful implementation of a Rare Disorders Strategy for Aotearoa. We are grateful for the opportunity to provide feedback at the early stages of this draft strategy and commend Manatū Hauora for starting progress towards this.
Occulo dento digital dysplasia (ODDD)	I was wondering why we were sent a document that has obviously been prepared and worked on for some time, and reflects well established themes (that the Ministry don't want widely distributed)? no doubt the result of the lobbyist activity which some groups and individuals contributed to and are given 2 days to make comment back. IS this a change of approach by the management group (transparency?) , reflecting another new hire in the top role or because the Ministry or yourselves needed the "community was consulted box" ticked.
Unique NZ	I wasn't able to share the document to the group unfortunately but I did ask within the group for feedback on the seven priorities that RDNZ had identified, as well as what is important to them for a Rare Disoders Strategy for Aotearoa New Zealand. These are the responses I got, so I hope they are useful. 1. Do you think modern medicine and alternative therapies? 2. Education and training for healthcare professionals is certainly important. I had access to my daughter's medical notes several years ago and discovered a medical professional had noted her as having Down Syndrome. I complained about this and an amendment was put in her hospital file. 3. Mention of VOUS - variant of unknown significance, as a diagnosis isn't exact or clean, and changes over time, and no diagnosis is a barrier to communication and can impede getting support. 4. I also feel so tired when I hear of another strategy. The biggest issue we have faced is staff shortages. If there were enough medical professionals they'd have enough time to work through clinical pathways, disability supports, and advocate for the medicines needed. The staff we've had who weren't stretched for time helped us make the most progress. A complex system will always be complex. Ideally it would be the medical staff working through issues in the system rather than the parents or the patient. 5. Transition support from school and beyond 6. What I would like is access to medical treatment like Botox as there is not any other available treatment due to other medical issues. 7. I actually find that even though Paediatrics did the testing for Chromosome Disorders its often dismissed when your Child has a Medical Crisis. The

	Orthopedic Specialist was first one in over the year to consider there could be links to the Chromosome Duplication. I do think Parents are often dismissed with their concerns because their Child doesn't fit into neat little tick boxes. I think it's too easy to blame the Parents but when there is very little actual practical Support it can be frustrating. 8. When it comes to rare disorders I think healthcare professionals need to keep an open mind because we don't know how a deletion or duplication will affect our child. It took some years before my daughters chromosome duplication was linked to her orthopaedic issues. My thoughts... **Data & Measurement**: What do Genetics do with their data upon diagnosis? It is added to an international database? What data is captured and recorded? Think it is really important for our group to know about any/partake in any clinical trials/other research/studies that might add value in knowledge building, support etc. This includes any international studies. **Clinical Health Services**: Think it is really important to link into overseas organisations like Unique (UK) and Chromosome Disorder Outreach (US) who have well established databases, guides etc for Rare Chromosome Disorders that can be useful and guide medical professionals here in their responses to their patients. Keeping up with the latest research. **Disability and whanau support and coordination**: Not just those with Rare Disorders, but also those caring for them - especially when the person with the rare disorder may not have a voice/speak for themselves Look at having supported decision making. **How easy would it be to make progress? Are there any significant barriers?** Currently I often find all the different medical professionals/groups involved in my son's care often work in silos and don't communicate with each other. This needs to be improved dramatically. **What important possibilities are not yet identified?** It would be nice to know how positive it has been for other countries who have put a rare disorders strategy in place. What worked/didn't work. What can we (NZ) learn from them in putting one in place here = not making the same mistakes, or could bo better? Thanks for the opportunity to contribute with feedback.
Ehlers-Danlos Syndromes New Zealand	Thank you for making Rare Disorders in New Zealand a focus point, it's been a long time coming. I have been working in the rare disorders community since 2017, and the majority of all of our support groups have the same concerns and issues in the current systems that we all experience. First and foremost, my primary concern is that the rare disorder community was not involved from the very start of this process. It is essential to adopt the "nothing about us without us" approach when developing strategies that directly impact our community. I strongly believe that co-design is key to the success of this strategy. By actively involving individuals from the rare disorder community in the drafting process, we can ensure that vital elements are not overlooked or misrepresented. Additionally, early involvement can help avoid the challenges associated with making changes once strategies are already in place. In light of the proposed principle of "giving voice," I would like to emphasize that our goal is not simply to have a voice but to be actively included. It is crucial that the strategy acknowledges the value of including individuals from the rare disorder community as equal partners in decision-

	making processes. Our lived experiences, insights, and perspectives are invaluable in shaping the direction and implementation of initiatives. Furthermore, I would like to highlight the importance of including Māori and Pacific peoples in the co-design process. To ensure an appropriate understanding of cultural needs and wants, it is necessary to actively involve diverse cultural groups. By incorporating their perspectives, we can promote cultural safety and tailor strategies to be more effective and respectful of the unique requirements of these communities. The Social Model of Disabilities 1 2 The recent Rare Disorders Survey in 2021 asked the question, "Are you disabled?" This question reflects an individual identity, which is not something the medical system can fully quantify. It is crucial to recognize that some people may not perceive themselves as disabled, while the majority of our community identifies as disabled based on the social model of disability. I noticed that the strategy draft does not mention the social model of disability, despite mentioning disabled individuals. The social model of disability emphasizes the barriers and lack of equitable care faced by disabled individuals due to societal factors rather than their individual impairments. It is essential to address this aspect within the strategy and prioritize a focus on equitable care for individuals with rare disorders. By acknowledging and incorporating the social model of disability, we can shift the narrative from an individual-focused perspective to a more comprehensive understanding of the barriers and challenges faced by the rare disorders community. This shift in perspective will help guide the development of strategies that aim to address the systemic and societal issues that hinder equitable care and support. I strongly recommend including a dedicated focus on the social model of disability within the strategy. By doing so, we can ensure that the needs and experiences of the rare disorders community are accurately reflected and that measures are taken to address the lack of equitable care that many individuals face. 1 https://social.desa.un.org/issues/disability/crpd/convention-on-the-rights-of-persons-with-disabilities-crpd 2 https://social.desa.un.org/issues/disability/crpd/article-1-purpose 3 https://www.raredisorders.org.nz/about-rare-disorders/rare-disorders-in-new-zealand/nz-voice-of-rare-disorders-white-paper-2022/ 4 https://www.hqsc.govt.nz/consumer-hub/engaging-consumers-and-whanau/code-of-expectations-for-health-entities- engagement-with-consumers-and-whanau 5 https://www.archives.govt.nz/discover-our-stories/the-treaty-of-waitangi 2 Why have a rare disorders strategy • Impacts of rare disorders • Unrecognised and undiagnosed conditions • Equity Another critical point that I would like to emphasize in my feedback pertains to the importance of timely and accurate diagnosis within the rare disorders community. While the strategy draft mentions equitable and timely care, it is crucial to recognize that at the forefront of a consumer with a rare disorder is the need for knowledge, compassion from medical professionals, medical access, and accurate diagnosis. Many individuals within our community face significant challenges in obtaining a proper diagnosis, often enduring years of uncertainty and frustration. It is not uncommon for individuals to struggle for 20+ years before gaining a comprehensive understanding of their condition. Without a timely and accurate diagnosis, it becomes exceedingly difficult to provide appropriate care and support to those in need. To address this issue effectively, it is essential to establish better pathways for diagnosis within the healthcare system. This includes improving the accessibility of diagnostic tests, ensuring that healthcare professionals are knowledgeable about rare disorders, and implementing streamlined processes that prioritize early and accurate diagnoses. By providing the necessary resources and support for early diagnosis, we can alleviate the burden placed on individuals and improve their overall healthcare experience. I recommend that the strategy draft explicitly addresses the need for improved pathways to diagnosis for

	individuals with rare disorders that are used and are accessible, as currently the pathways that are available are not being used or are not 'believed'. While common conditions may be easier to understand, some treatment plans, based on a consumer presentation, may be rare, and these treatment plans would be best supported by a rare disorders strategy as well. What is wanted from a rare disorders strategy? & What a strategy will provide? This should not be included here until co design and lived experience expert consultation has been commenced, as without it there is not a true gauge of what's important to our community. What are rare disorders? Terms of reference for the inclusive nature of disorder to disease etc is important to include anywhere that the meaning of rare disorder is noted throughout all systems. We often find that discussions are had to make one seem more 'lesser' than the other, and we want to strengthen the understanding of what's important and not frivolous meanings of words. Also, how is a definition, 'promoting equitable access and support in a health sector?' and 'concentrating effort in improving information and guidance to underpin quality healthcare?'? Key elements of a rare disorders strategy Proposed principles 'giving voice', no we don't want a voice, we want to be included. Doing the work to develop a strategy there doesn't seem to be any clarity in this section at all. Who what where • Identifying key opportunities that could be built on Another item that will need to be addressed by the appropriate parties is around Māori and Pacific peoples, and the 'assumed' medical diagnosis that they face as a hurdle to a diagnosis and understanding. I personally was not happy with the use of 'especially Māori and Pacific peoples' throughout this document; It seems to be once again dividing us, when we should be inclusive of all and not othering any area of our population. 'Including Māori and Pacific peoples' is the way to address this issue. The ability to help coordinate care better, and approach consumers with a more holistic approach with multi-disciplinary teams, is hindered by the fact that the medical systems do not talk to each other as mentioned above. • Capturing voices of the rare disorder's community • Inputs that are underway or planned Under 'doing the work',' Process being made internationally such as' - how is this NZ's work? 1 https://social.desa.un.org/issues/disability/crpd/convention-on-the-rights-of-persons-with-disabilities-crpd 2 https://social.desa.un.org/issues/disability/crpd/article-1-purpose 3 https://www.raredisorders.org.nz/about-rare-disorders/rare-disorders-in-new-zealand/nz-voice-of-rare-disorders-white-paper-2022/ 4 https://www.hqsc.govt.nz/consumer-hub/engaging-consumers-and-whanau/code-of-expectations-for-health-entities- engagement-with-consumers-and-whanau 5 https://www.archives.govt.nz/discover-our-stories/the-treaty-of-waitangi 3 'We need to develop a more detailed picture of key work underway' - I don't think a strategy's job is to understand other strategies, the focus should be on progressing rare disorders in NZ, and doing the work to get us included at the table of all these other discussions. If we are not included in the beginning, or during consultations phases, are voice once again will be missed, and in turn we will become the 'tack on'. We cannot delay action. Identifying focus areas for the strategy We need to be planning for the future and beyond, planning for a centre of excellence is the future of rare disorders in NZ. Not only would they join other countries in being world leading, but they would be able to provide support to consumers, and supporting, guiding, and connecting medical professionals with the correct knowledge, treatment plans, and international

	contacts to allow for overall more successful outcomes for consumers. We have some planned pathways of care in the medical systems, but these are not being used, or even not being believed. A centre of excellence would help strengthen the knowledge and more acceptance of these conditions. There needs to be a bigger understanding that it's not just a delay in treatment that can hinder a consumer and progress of an illness, but that this can have permanent impacts on an individual, and in turn require even more assistance within our healthcare system. This draft strategy does not seem to have this understanding. The statement 'The strategy will not do the work or identify specific actions or commitments in focus areas' - is extremely alarming. No, it should plan to identify actions, and it should commit to better outcomes for people living with rare disorders. • Data & measurement • Digital solutions and enabling Mentioning 'health coaching and wearable tracking'? - this is the least of our worries and very specific. • Critical health services An extremely important point that needs focus in this strategy, is the main issues we all face currently. All of our rare disorder support stems from poor data and information collection. Our government/ministries/ hospitals/education etc systems rely heavily on facts and numbers to get funding, assign workers, and dedicate research to, among a raft of other allocation situations, and if we do not have visibility, we are simply lost in the systems that are supposed to be there to help support us. I have also faced medical professionals complaining to me with the lack of visibility in their current systems. The public health (hospitals), Allied Health, Private care and General Practitioners, all use different systems (And also struggle to access MRI's, CT, and x-rays from other services), and within these systems they use different coding systems and therefore the collection of data is harder to collate. Not only is it hard to collate, but our medical staff are struggling to find time to actually complete coding on their consumers. There is not enough money, time, or staff in their day to do this correctly. Medical staff are, with good reason, focusing more on their growing consumers needs and concerns, and do not have time to deal with 'frivolous' admin work, but if they had the support staff around them to do this because government agencies saw this as a priority, it would immensely help our entire population as there would be true visibility. • Disability and whānau support and coordination It is important to include UNCRPD and social models of disability here. It is also important to note that more carers or whānau are also disabled themselves. • Knowledge building All of Aotearoa, New Zealand • Funding, assessment and prioritisation We also need reassessment on services when they are blocking parts of our community from receiving social services, when this would allow them to participate in society more and have better quality of life. • Research and evaluation New Zealand in the past has been seen as a 'great sample size' for research. Why are we not tapping into this to become worldwide leaders? Why are we not giving medical professionals support to do this vital research? Allowing medical professionals to do this good work, not only impacts our community in positive ways, but also helps spread the knowledge and understanding of situations and conditions. 1 https://social.desa.un.org/issues/disability/crpd/convention-on-the-rights-of-persons-with-disabilities-crpd 2 https://social.desa.un.org/issues/disability/crpd/article-1-purpose 3 https://www.raredisorders.org.nz/about-rare-disorders/rare-disorders-in-new-zealand/nz-voice-of-rare-disorders-white-paper-2022/

	4 https://www.hqsc.govt.nz/consumer-hub/engaging-consumers-and-whanau/code-of-expectations-for-health-entities- engagement-with-consumers-and-whanau 5 https://www.archives.govt.nz/discover-our-stories/the-treaty-of-waitangi 4 Key points: 1) We need more co-design from rare disorder lived experience experts, including Māori and Pacific peoples. 2) The mention of 'support pae ora for people and whānau with rare disorders' is throughout this strategy. From what I can tell, since there is no terms of reference for this word, is that this is related to Pae Ora (Healthy Futures). Healthy futures could be seen as offensive and a kick in the teeth. Pae ora being healthy futures, does not apply to some, if not most of our community, we will not get better, our condition requires appropriate management, not fixing. Equitable outcomes and equity for people and whānau with rare disorders is what needs to be mentioned throughout. 3) Fixing the data and system issues - without a high level of focus on the discrepancies between systems talking to each other, medical staff will not be able to provide timely appropriate care levels for consumers, and will find it harder to connect with other medical staff in a multidisciplinary approach that benefits the consumer. Without fixing our broken data and supporting medical professionals to utilise our coding systems correctly, our government and government agencies will continue to have an incorrect view of people living with rare disorders, and in turn our allocations of funding, staff, and training will continue to go unheard. 4) The importance of including future plans for a Centre of Excellence within the rare disorders strategy. Establishing a Centre of Excellence can serve as a central hub for knowledge, expertise, and resources dedicated to rare disorders. Such a centre would provide a unified platform for collaboration between medical professionals, researchers, and the rare disorders community, thereby strengthening our collective efforts. A Centre of Excellence can play a vital role in fostering innovation, improving diagnostic capabilities, enhancing treatment options, and advancing research in the field of rare disorders. It can serve as a focal point for multidisciplinary expertise, ensuring that medical professionals have access to the latest information and best practices in the diagnosis, treatment, and management of rare disorders. Moreover, it can provide a platform for knowledge-sharing, training, and capacity building within the healthcare system. For individuals and families affected by rare disorders, a Centre of Excellence would serve as a valuable resource and support network. It would offer specialized care, tailored interventions, and comprehensive services to address their unique needs. Additionally, it would facilitate collaboration and engagement between the rare disorders community and medical professionals, ensuring that consumer perspectives are integrated into research, policy development, and decision-making processes. I strongly recommend incorporating plans for a Centre of Excellence within the rare disorders strategy. This strategic approach will provide a clear and tangible goal for strengthening our collective efforts and fostering collaboration between medical professionals, researchers, and the rare disorders community. By establishing a Centre of Excellence, we can consolidate resources, enhance expertise, and ultimately improve the outcomes and quality of life for individuals with rare disorders. 5) Multi-disciplinary teams are extremely important to consumer outcomes. We often have multiple specialists that we have to deal with, and it would be great if this was promoted and there was support to be able to provide this, including more nursing staff to help be key coordinators.

	6) Using our obligations for UNCRPD1, Te Tiriti o Waitangi5 and Code of expectations for health entities4 as key documents to our community, being able to leverage vital information from these documents. Question for your consideration - How are we going to get the initial data from our community as a landing pad to start this work from? Advertising nationally for people with rare disorders to come forward? Advertising to medical professionals (especially GP) to assess patients on their workloads that fit rare criteria (since the data work hasn't been done) etc… This is critical to success. Are we going to support more research to be done in New Zealand, and include this in the strategy as an ongoing, funded, purpose to help understand the lives of people, whānau, and communities living with and supporting rare disorders If you have any questions, comments, or need to discuss any points made, please contact me on kelly@ehlers-danlos.org.nz Noho ora mai, and look forward to connecting soon. Kelly McQuinlan Founder and Chief Visionary Officer Ehlers-Danlos Syndromes New Zealand
DEBRA NZ	Initial responses – Anna Kemble Welch, Director, DEBRA NZ and Martin Hanley, DEBRA NZ volunteer Projects Officer and RDNZ Trustee. The strategy document goes a long way to identifying the problems faced by people living with rare disorders in NZ in a general way. The following points are focused on some details that impact on how that will be applied. 1) 'Rare' is even more rare in a country as small as NZ, where the small and widely dispersed population mean it's even more unlikely that hospitals and medical centres have clinicians with any experience let alone expertise in a condition that is rare or very rare. I'm not sure the strategy really addresses this issue. In many other countries a city hospital is serving a population of millions who all live in that district, so it's more likely there are a group of people with a particular rare condition and it's more realistic to have specialists in a particular rare condition in each different centre. In NZ, for patients to see a healthcare professional who has developed any expertise, they need to be able to travel to a specialist centre in a different district. The Rare Disorders Strategy needs to acknowledge this and include developing expert specialist multi-disciplinary clinical care that is concentrated in a particular centre, not dispersed and diluted. This might require clinicians to come together from different districts for national clinics on a regular basis that patients travel to attend, or for clinicians with expertise in a rare disorder to travel to different districts where they see the patients in that area. 2) Acknowledging that clinical care needs to be based on evidence, it needs to be understood that with rare conditions it is far less likely that articles will have been published in peer reviewed journals referencing evidence based evaluations of treatments and for Rare and Very Rare this is not the only clinical knowledge that should be considered. Rare conditions frequently involve too small a population for statistically valid evaluation of medical trials. As is the case with successfully adopting indigenous medical knowledge, with Rare Disorders a Realist Evaluation Methodology can be a more important basis for confirming best practice treatment and care protocols. With rare conditions it is important to listen to the lived experience and expertise of the patients, and the accumulated shared health care knowledge gathered by patient groups. The usual treatment pathways clinicians are trained in and the protocols developed for treating the 'normal' population can be totally inappropriate and not Best Clinical Practice for a person with a rare condition. The Strategy needs to emphasise that there will be times when the usual treatment for a patient in the 'normal' population can be the wrong approach and dangerously life shortening for those with a Rare Disorder.

	3) In respecting te Tiriti and Maori expertise, it's important to acknowledge that much indigenous knowledge focused on healthcare will not have been published in journals, and again this shouldn't be excluded as valid evidence for guiding treatment pathways. A Realist Evaluation Methodology can provide the framework for accepting that the different threads of knowledge that will lead to the best health outcomes are not always drawn from the usual western published scientific articles model of what constitutes 'evidence'. 4) The timeframe to develop the strategy that leads to providing co-ordinated care for people with rare disorders, shouldn't be an excuse to not press on rapidly in any quarters where improvements in access to care and treatment could be made right now. Those with Rare Disorders are often confronted with a much, much shorter lifespan than the 'normal' population. The system needs to embrace temporal equity and move promptly through developing to adopting this strategy. With many rare disorders 5 years might be 1/5 of that person's lifespan. The 'normal' population would not tolerate a health strategy that took more than 15 years (1/5 of a normal 75 year lifespan) to devise and implement. Aotearoa's Rare Disorders Strategy must be well resourced so that its implementation can be expedited.
Cystic Fibrosis NZ	Cystic Fibrosis NZ Feedback on Rare Disorders Strategy Document 18 July 2023 As far as we can see, this is a scoping document to attempt to inform the direction and content of the proposed Rare Disorders Strategy rather than a first draft of the strategy itself. It provides a very general background and discussion of rare disorders and key issues, along with proposed principles, structure, and key areas of focus for the strategy. One positive thing is that the document does propose a definition of rare disorders as 1 in 2,000. Obviously, CFNZ strongly supports this definition. General Comments Our main concern is to ensure that we have a strategy that leads to real action and measurable progress on the ground for people with rare disorders – with agencies held accountable for delivery of their aspects of the strategy. This document doesn't give us great confidence that this will be the case. We are particularly concerned by a comment on page 8 "The strategy will not do the work or identify specific actions or commitments in focus areas. However, to successfully point to the directions for maximum leverage and progress, a good overview is needed or what is already planned, what could be possible and how much could be realistically achieved across possible areas to emphasise." The document also gives no indication of process, next steps, or timing for the preparation of the strategy. There is clearly a major amount of work still to be done but it is essential that there is a clear timeline for delivery. Feedback to Ministry of Health Questions: The Ministry of Health has set out some areas under which they have asked for feedback. To make things easier for Rare Disorders NZ to collate feedback, we have attempted to group our comments under the Ministry of Health headings. Are there additional points that are important to capture – in the background (why have a strategy) and in the proposed definition? • There needs to be a section in the strategy that discusses previous reviews that make recommendations in respect of rare disorders, in particular the recent Pharmac review,

	so that these shortcomings are captured and brought through to the focus areas to ensure those issues are being addressed. • The proposed definition for rare disorders of 1 in 2,000 is strongly supported – it is essential that there is a clear, quantitative definition in line with accepted international practice. Consideration may also need to be given to a definition for ultra rare disorders and again this should be in line with international practice. Do you agree with the key elements (aims and principles) proposed for the strategy? • CFNZ supports the intentions expressed by Ministers as to the results of the strategy (page 4), but the strategy needs to make explicit how progress against these results will be measured and the parties responsible for delivery held accountable. • The principles proposed to underpin the strategy also need to include a reference to identifying and supporting the delivery of real improvements to the lives of people living with rare disorders. Are there additional opportunities that could be built on (doing the work to develop a strategy)? • The proposed content of the strategy currently lacks an assessment of the current situation with respect to rare disorders in New Zealand, other than a very general discussion. While current data may not be ideal there are real opportunities to use existing data to paint a picture of the current position e.g., Rare Disorders NZ information, Statistics NZ, Te Whatu Ora databases, MOH's own surveys etc. It is essential that a baseline, however limited, is established not only to provide a base against which to measure progress but also to identify gaps in current knowledge that need to be filled. • It also lacks a comparison of how NZ currently compares with similar overseas countries – hopefully, this is part of MOH's current work programme. Comments on the focus areas proposed for the strategy? • It is premature to confirm the focus areas for the strategy at this stage. The focus areas should fall out from the assessment of the current situation proposed above, the issues already identified by previous reviews e.g., Pharmac review, the issues already identified by Rare Disorders NZ, feedback from the rare disorders' community, and from clinicians with experience of rare disorders. The focus areas identified may well be part of the issues identified but they will not be the only ones. • Once the focus areas have been confirmed following the process above, the next steps would be to compare those areas against the stocktake of work already being undertaken and any gaps identified in the strategy and proposals developed as to how they will be addressed and by whom. What important possibilities are not yet identified? • It is essential that the strategy leads to real action and measurable progress on the ground for people living with rare disorders – with agencies held accountable for delivery of their aspects of the strategy. • While the strategy may not get into the specifics of individual activities to be undertaken by the various agencies, it must provide sufficient detail to enable agencies to be clear about their roles and responsibilities, for progress to be measured, and accountability for delivery to be clear. • This means that the strategy must specify: o The gaps in current programmes and activities o The actions to be taken to address those gaps. o The parties responsible for addressing those gaps and timing for that. o How progress against delivery will be measured and how agencies will be held accountable for delivery • Without the above, the strategy will not lead to real improvements for those living with rare disorders

FARA	Research: needs to prioritise Developing a national strategy to support, promote and disseminate all types of research for rare disorders nationally and internationally Support and promote clinical trials for rare disorders to take place in Aotearoa, enabling NZers with rare disorders to participate in clinical trials and other research activity Support partnerships between researchers, clinicians and people with the rare disorders.
Fabry	• I did not see at the beginning of the document what pae ora is. I soon worked it out and I understand that those in the health system are familiar with the term. but it is new to me and I imagine many others. Use of te reo is fine but there should never be the assumption we all know what the word means. I am learning more te reo daily! • RESEARCH: I presume this is referring to individual rare disorders? Even if we have the capacity to do this in New Zealand there are many rare disorders. Are we duplicating what is being done overseas by countries with institutions likely to be underwritten financially by the pharmaceutical industry and with all the resources just not available in such a small country? Certainly some pharmaceutical companies have lost interest in New Zealand as Pharmac just isn't interested in buying their product. {A double edged sword}. • I would like to see mention of the **cost to the economy** of those with rare disorders not getting equity which is likely to result in delayed diagnosis, treatment etc and therefore may not be able to work. • There cannot be too much emphasis put on educating the many different agencies that will be party to enabling this strategy. I have a particular beef with WINZ from personal experience, who certainly had a high turnover of staff when I dealt with them just over 10 years ago. With rare exceptions, staff were not the highest calibre. Will the education around this strategy filter down to those who encounter the public face to face? • Equity under the tenets of the Treaty of Waitangi for Maori, and for Pacific peoples is fundamental but please do not lose sight that this strategy is for everyone of us with a rare disorder who suffer inequity under our present health system; sometimes this appears to be lost.
NZ Amyloidosis Patients Association	I had read the details of the submission and I am very happy with all the points highlighted. It would be wonderful and a great start if the government can incorporate and enforce even half of what we ask for, starting with the very important rare disorder strategy and registry. I think everything else will follow suit.
Prader-Willi Syndrome Association New Zealand	Feedback on Draft: Rare Disorders Strategy for Aotearoa New Zealand – July 2023 Prader-Willi Syndrome Association New Zealand Thank you for the opportunity to provide feedback. In response to the particular questions for feedback, we have made the following notes: • We agree with the reasons stated for why the strategy is needed and the definition provided of what a rare disorder is. Our only comment on this section is regarding page 4, under the heading 'What is wanted from a rare disorders strategy?'

	The second point states "allow the health sector as a whole to provide much better support for people with rare disorders", but we feel this should state 'support and healthcare...' • We agree with the key elements (aims and principles) - page 6. • Regarding additional opportunities that could be built on (Doing the work to develop a strategy = pages 6 / 7), does bullet point 4 "advances in science, such as through new genetic and pathological testing capability" include pharmacogenomics? It would be good to include this type of testing as a new science to build upon. Pharmacogenomic testing is not currently readily available in NZ but could be invaluable for rare disorders, and in the treatment of mental health conditions where patients sometimes trial lots of medications before finding an appropriate treatment. Pharmacogenomics allows medication and dosage to be tailored to the individual and in PWS for example, this would be very useful because medications are often metabolised differently. An example being the death of a young man aged 20 recently (my own son) from suddenly becoming diabetic and presenting with ketoacidosis - awaiting full postmortem results, but he was on psychotropic medication which could possibly have exacerbated the hyperglycaemia. Pharmacogenomic testing prior to being prescribed this medication may have helped find a more appropriate treatment. This testing is available if a) patients / families / clinicians know about it and the testing can be funded privately, b) a clinician is available to interpret the results for the patient. The following organisations were providing this service in NZ at patient cost: https://easydna.co.nz/ and https://www.mydna.life/en-nz/ but there are sometimes stock distribution issues in NZ (and testing is sent to Australian labs we believe.) • Doing the work to develop a strategy / capturing voices of the rare disorders community - we believe the 7 priorities previously identified and set out by RDNZ are an excellent summary of what our community thinks is important and what is needed from a strategy. We would like to add some detail to priorities 2 and 3 as follows: 2. "Planned pathways for clinical care – coordinated and integrated pathways for cohesive healthcare." In addition to clinical care, we would like to ensure that the planned pathways also include allied healthcare, and care provided by supported living providers / support staff in residential situations that relates to health and wellness. Pathways should not be geographically determined - all patients should be able to access / benefit from expert clinical advice. 3. "Access to disability and social support – implement simple mechanisms to ensure appropriate access to disability and social supports." We would like to add access to educational support where necessary (not relevant for all rare disorders). For example, all pupils with PWS will need some level of learning support at school, but parents and schools often have to apply more than once to be successful at obtaining ORS funding and many applications are declined. A mechanism in their care pathway following diagnosis that ensures an entitlement to some learning support (the level of which would vary per individual pupil) would be very

	helpful and avoid repeated, time consuming applications and the negative consequences of some pupils missing out. As a support organisation, we meet many of our members with PWS and it can be difficult to understand why one pupil may have been successful in receiving ORS funding and another has not. • Identifying Focus Areas for the Strategy (pages 8-10): We were a little uncertain what is meant by this sentence in the 2nd paragraph "The strategy will not do the work or identify specific actions or commitments in focus areas." Data and measurement: Another purpose of data collection would be to identify gaps in order to improve care. Regarding research, not only would we hope to attract researchers to NZ, but we would also hope that with improved data, researchers would be encouraged to more easily include New Zealanders in studies initiated overseas, for example research partnerships could be established with Australia. (Return flights to a clinical site visit in Sydney, for example, would not be too dissimilar to return flights to Auckland from some parts of NZ.) Data collection needs to include information gathering from Te Whatu Ora, Whaikaha, WINZ, NASC organisations and Education services. It's also important that relevant data should be shared with support organisations such as ourselves (DIAS provider) and other relevant groups, in order that we can also provide better targeted education, support and advocacy. Digital solutions & monitoring: The possibility of accessible diagnostic and clinical standards, guidance and pathways for health practitioners integrated with patient health records is extremely important to us. This has the potential to not only improve the health of those living with PWS, but to save lives. Clinical expertise in PWS is limited in NZ and as such, our organisation does not have a team of medical advisors / advisory board. There are one or two experienced individuals we consult at times, but we have mainly built relationships with experts overseas through international conferences and the ability to communicate so easily via technology. As far as we are aware, there are no care standards / guidelines currently in use for PWS in NZ, and we agree with the suggestion that these may need to be sourced internationally. These are urgently needed and as an Association, we could provide an outline of what care standards should include for PWS, but to develop medically detailed standards from scratch would be a very lengthy process. We are aware that the PWSA of Victoria Australia and the Prader-Willi Research Foundation Australia have both been working on developing standards of care documents and they would likely share this work with us. We would like to be involved in the development process of care standards for NZ alongside medical professionals due to our knowledge of patient experience and international practice and contacts. We have found that medical organisations in NZ are not proactive in reaching out to us, for example endocrine, paediatric, obstetric associations. It would be beneficial to improve these relationships so that we are invited to attend relevant national conferences, establish useful contacts, or to easily contribute information for review or international information that could be shared / presented. This could also lead to groups like ourselves having medical advisory boards, which in turn could be advantageous for medical practitioners in terms of professional development / career development etc. Critical health services: We agree with all the comments and suggestions in this section to improve system responsiveness. However, there is concern that focus may be overly weighted toward paediatric services: "...some specialties and service areas will be critical in improving this overall responsiveness.....service areas like paediatric and especially developmental paediatric and paediatric cancer services, genetic, metabolic, screening and pathology." Whilst early diagnosis and intervention are vitally important in PWS, we are aware that services tend to drop off as children get older and once they reach adulthood, healthcare monitoring is much reduced. Healthcare pathways / care standards should ensure that patients have

	access to necessary services at the appropriate age or stage, and data collection should be able to tell us where the highest risk factors exist. Our current knowledge and experience tells us that patients with PWS have greater health risks during the transition phase of adolescence to adulthood. Currently, once PWS patients exit paediatric services and 3 monthly endocrine monitoring, they may see an endocrinologist annually, if at all, under adult services. Monitoring of a rare and complex health disorder often falls on a local GP for adult patients. When intellectually disabled adults are living in residential facilities, visits to a GP or health practitioner are also often reliant on there being proactive and knowledgeable support workers or involved parents with guardianship orders in place. The cost of GP visits also has the potential to be a barrier for beneficiaries. The adolescent and young adulthood years are also the period when families typically struggle the most with supporting their family member, but community supports tend to reduce. Disability and whanau support & coordination: We agree with the point that support can be hard to access for people living in rural or remote communities and/or with particular language or cultural needs. In PWS this might be a lack of vocational or employment opportunities for school leavers, or a lack of therapist services etc. Our experience seems to be that it is nationally difficult to access mental health services, education support and respite services. However, we would like to note that it is our experience that our larger cities are also experiencing problems of their own, in particular an increased wait time for NASC assessments and a very long wait time for residential care places. We are also aware that some of our families privately fund medication for their adult children once they no longer qualify which creates an issue of equitable access to treatment. Knowledge building: Care standards linked to patient health records will greatly improve knowledge and care provision of health practitioners, but we need to think about how others (who don't have access to health records) will access information / build knowledge to effectively support and respond to people with rare disorders. As a DIAS provider we provide information to families or organisations who contact us and we reach out to neonatal units to provide newly diagnosed families with information packs, but not all families get in touch with us, and not all service providers or community organisations reach out to us or are receptive to receiving our training programmes. There needs to be clear advice or recommendations on where these groups can go for information. Perhaps clinical care pathways can include checklist notes for medical practitioners to ensure that other agencies involved in their patient's life have been informed of where to go for information to receive basic training on health risks to be monitored or managed. We feel it may need to be made a compulsory requirement for residential service provider organisations who have full duty of care to receive training in a rare disorder such as PWS, and to repeat this frequently due to rapid staff turnover. Standards of care documents for supported living providers that cover other elements of support provision in addition to healthcare monitoring requirements would be useful, but this would be a lengthy task to develop these. Such documents do already exist (developed by the International PWS Organisation's Professional Care Provider's Board) but they are extremely lengthy and detailed documents that are not very user friendly for provider organisations. Funding, assessment & prioritisation: We do not feel that the assessment and prioritisation of products such as medicines and medical devices in NZ can currently be described as 'good'. We need to build on the processes we already have to enable new medicines to be evaluated more easily. There are inexpensive medicines used overseas in PWS, mainly for mental health or behaviour, which are not available here yet, perhaps only because they haven't been put forward for funding consideration due to lack of medical expertise and awareness about them. Often these medications are used 'off-label' so their use for PWS wouldn't necessarily be

	routinely considered. A lack of knowledge and awareness of the latest research in rare disorders often contributes to a lack of assessment, or to prioritisation decisions that our patient and family members might not agree with. Research & evaluation: There is a wide variety of research being undertaken into PWS globally, including metabolic, appetite and obesity drugs, hormone therapies, gene therapies, therapeutic devices, and investigations into therapeutic intervention programmes. Most of the larger scale studies take place in the USA due to larger patient numbers, relationships with pharmaceutical companies and funding, but studies are widespread internationally and there are several multi-site international studies that take place. It would be unlikely that new drug studies would ever be solely run in NZ due to our small patient population, so it is important that collaborations are encouraged in order to allow both participation of New Zealanders in clinical trials and the growth in knowledge of our clinicians. There are well established research organisations who fundraise for research and work with expert clinicians (scientific advisory board, clinical trial consortium) to prioritise the research projects they fund and to grant funds. In PWS, this organisation is FPWR (Foundation for Prader-Willi Research) which is based in the USA but has 'satellite' organisations in the UK and Canada. Australia has a similar research foundation. Establishing links with these organisations would be very worthwhile for our clinicians to access knowledge, promote trial participation and to enable evaluation of effectiveness for Māori and Pacific populations. Over the years, there has been keen interest from our members in attending international conferences, both the international (IPWSO) conferences and the Asia-Pacific conferences. The Asia-Pacific conference has historically been jointly run by the Australian and New Zealand PWS Associations, although held in an Australian city due to having a greater number of registered participants, so travel costs are reduced for attendees. Attendance by health practitioners from NZ has always been extremely low. The last Asia-Pacific conference was jointly hosted by Malaysia, Australia, NZ and Thailand and was held virtually. It was advertised across our medical associations, hospitals and universities and we hoped for a greater number of professional attendees, but there were still very few from NZ. Malaysia had a high number of professional attendees and there appeared to be a professional development accreditation programme which encouraged participation. It was also much easier for Malaysia to source and attract national experts to present at the conference because the Malaysian PWS patient association has strong connections with the medical community in Malaysia. We would like to see more collaborations like this in NZ with patient and medical groups working together, and encouragement toward involvement in conferences. Technology could also allow us to connect via virtual networks and both patients and professionals could benefit through information sharing via webinars. Many of our family members are members of international discussion forums where they are often able to learn from others and it would be good if our medical community also had access to international virtual networks for health professionals. Data collection is key to improving research and evaluation opportunities. Relevant confidential data needs to be accessible to researchers. Patient groups like us can possibly assist in data collection via patient surveys. Responses to surveys do not capture a full picture as they require time commitment and rely on technology use, but large-scale patient survey programmes are used overseas, for example for The Global PWS Registry and the PATH for PWS surveys. Some of our members already participate in these international survey programmes and if our clinicians connect with these programmes, survey results could provide useful information

EGPA/Churg Strauss New Zealand	The draft strategy sets good intent in raising the profile and quality of care for people with rare disorders. There is one additional area of concern not explicitly addressed and that is a recognition within the health system overall that people on treatment for a rare disorder are often immune-compromised. I don't believe that risk is managed adequately right now as I experienced with an admission to hospital and put in a ward where 50% of patients had the flu. They were very aware that I was seriously immune compromised and that was putting me at further risk. Some sort of marker needs to go on the health system database to better manage patients with that risk.
Tuberous Sclerosis Complex New Zealand	• Are there additional points that are important to capture? o In the background (why have a rare disorders strategy?) *The limited research is also partly due to heavy workload of clinicians.* o In the proposed definition (what are rare disorders?) *Looks good to us.* • Do you agree with the key elements (aims and principles) proposed for a strategy? *Yes* • Are there additional opportunities that could be built on? (doing the work to develop a strategy) • For each the focus areas being explored on pages 8-10: o Do you know of work underway that could address this? o How important is it to achieving pae ora for people and whānau with rare disorders? *Very important. It would be amazing to have a strategy like this guiding care for people and their families living with rare disorders.* o How easy would it be to make progress? Are there significant barriers? *Re Critical Health Services: One of the barriers to workforce is the cost of training to be a genetic counsellor. The training courses are based in Sydney or Melbourne and although the Sydney course can be done mostly remotely, it costs over $50,000 per year. This is not attractive for training a diverse workforce.* *Having the clinical guidelines easily accessible would be fantastic.* Reviewing genetic testing at birth. Some countries are looking at bringing in testing for TSC soon after birth especially as research is showing very early intervention to stop seizures beginning is helping avoid some of the long term disability in people with TSC. *Investigating effective models of co-ordinated care is important to us. Our members would love to be cared for by health professionals who have knowledge of their condition before they meet them.* o How long would it take? *We have expert clinicians in NZ already. It might not take very long to bring them together to create a centre of excellence whether in person or virtually.* • What important possibilities are not yet identified? *Screening tools for surveillance of rare diseases. People with TSC need regular MRIs and blood tests to monitor for changes in their condition so that treatment can be begun in a timely manner. We would like reminders such as the cervical screening register provides to help patients and their clinicians keep on top of the recommended surveillance.*

Cure Our Ovarian Cancer	Jane had a read through of the strategy and her feedback was 'they've done a great job - it seems quite general and nothing specific jumps out at me.'
Arthrogryposis TAGNZ	Arthrogryposis Multiplex congenita is an umbrella term that describes up to 400+ different conditions. The name describes the presence of multiple joint contractures at birth. The contracture is limited In the range of motion of a joint And there's a non-progressive disorder, meaning children do not get worse over time. People who live with AMC can be affected mildly to those whose lives are affected severely and require high support levels. I won't comment on all the key priorities below but will pull out the most relevant for our members • Diagnosis – early and accurate diagnosis of rare diseases o We have seen this too often within our group where the parent/s has been given poor information or left feeling very despondent through poor or no information. These people are then left to try and find out themselves or discover a support group. Better education of medical staff is vital, particularly in the early stages. • Planned pathways for clinical care – coordinated and integrated pathways for cohesive healthcare • Again TAGNZ hears time and time again that it is a nightmare to navigate the health system. Hopefully, the new system, disabilities ministry, and your efforts will help develop simple clearly defined pathways. • Access to disability and social support – implement simple mechanisms to ensure appropriate access to disability and social supports • With the advent of enabling good lives / Min. disabilities we are hopeful the future looks brighter for our members and members of the disability / rare disorders communities. • Rare disorder medicines – equitable access to modern rare disorder medicines through a specific assessment pathway • Research – coordinated and funded programme of research for rare disorders • National rare disease registry – capture relevant data on rare disorders in New Zealand • Again, vital to establishing support and programs around these communities. One of the biggest issues we have is getting the word out that we are here to provide support if needed. AMC cover 400+ conditions so it is very hard to adequately describe what a typical AMVer looks like. • Workforce development – planned training on rare disorders for health professionals and support staff. • As above Again thank you for allowing us to voice our views. Although not adding anything new to this document, we agree that the strategic direction proposed covers the areas that concern us most and trust that the funding/resource can be allocated to achieve the best outcomes.
Pompe	Thank you for giving the NZPN the opportunity to read the draft rare disorder strategy and provide any feedback. After reading the report, the only points I will query or comment on would be; I think in the paragraph where we talk about what an effective rare disorder strategy could do, we should also include that by setting up a rare disorder strategy we can help

	to avoid the high-cost rare disorder patients are to the health system in the long term if there are not equitable and easy processes for rare disorders. That the socio-economic impact is not always measured. By not receiving the right treatments whether that be access to medicine, or physio to prolong mobility, patients are less productive in society, and their carers have to spend more time at home, removing their contribution to society. I note it makes reference to clear links to the Pae Ora strategies - we don't know what this is referencing if it hasn't been published yet, do we? As in, how rare fits into the Pae Ora strategies. Especially as I thought that the recommendations from RDNZ on that bill had not been fully included. I know it will take a while to implement but it makes me nervous when they say; "A rare disorder strategy will provide a clear direction of travel for the health sector on how improvements for people and whānua with rare disorders will be made progressively over the next decade." This does not sound like a priority if it is to be planned out over the next decade. I note that the draft does not specifically reference access to medicine or pathways for this. It does state that RDNZ has seven priorities and that these specific action areas will be considered in developing the strategy. Under Funding, assessment and prioritisation it does state that; "Even the wealthiest countries, or those with the greatest research and innovation outputs, cannot afford to fund all treatments,"...It goes on to say, "In NZ we have good assessment and prioritisation of individual products, (such as medicine..)". Where are they getting this information from? When I last checked we still woefully underfund medicine, with NZ ranking last for market access to modern medicine. I like the fact that we are finally tackling these issues, but I fear that it is all still too vague, but understand it is a draft. They reference a few times that the Pae Ora Bill or other work already underway might already address some of our strategies. But isn't the point of the rare disorder strategy that we look at better ways to have our own pathway within the MoH? Lumping us in with general practices may not be the best solution. I like the fact that they have mentioned a few times that strategy progression can be measured against international practices. I think this is important. We are already so far behind international practices, but I hope that we can now start to research, evidence review overseas rare disorder strategies, and practices, and see how we can implement them here.
KIWI CRPS	I read through the email regarding the rare disorder's strategies. It's very well written and comprehensive. It covers a lot of the difficulties that people with CRPS have, ie; delays in diagnosis, lack of knowledge from medical professionals, not being referred to appropriate treatment providers, receiving wrap around care, or continuity of care. Lack of social services input, and knowing what is available to us for assistance and where to get help and finally receiving equitable treatment around the country- some receive appropriate treatment and some receive nothing at all. A large stumbling block is that a good portion of CRPS patients attain CRPS via an injury so our treatment is driven via Acc.

	Therefore, your care, treatment, and whole journey can depend on who your case manager is, if they even accept your claim, how much they fight your claim and treatments, and this is once you have finally received your diagnosis. This leads to further inequality than what you receive via the normal healthcare system with a normal condition. Often, we receive the cheapest, non-specific treatment, which is for the injury and not CRPS, as opposed to what we actually require due to Acc's standard approach. This invariably wastes funds, causes appropriate treatment delays and irreparable damage - which they do not understand as they choose not to understand a rare disorder. With appropriate strategies and pathways in place, it would hopefully mean that it would put an end to this, as regardless of provider, they would have to follow the rare strategy and pathway - and then fairness in all systems would be addressed. These are the thoughts I had anyway in relation to CRPS when going through it. I think it is vital to improve what we have all struggled with and what you have developed is bang on.
Dystonia	Diagnosis; We regularly receive messages about either no diagnosis or misdiagnosis for dystonia, resulting in extreme stress and delay in treatment. Lately there have been requests from allied health workers on treatment pathways for patients, which is a concern. Our group focuses on supporting those with dystonia, we cannot be expected to also guide health professionals in the diagnosis/ referral/ treatment process. Access to disability & support services is not good. An example is me needing another mobility walker as the old one was last assessed in 2015. My GP was not sure who to refer me to. After checking with a colleague I was referred to NS hospital for an up-to-date assessment. Two days later I received a decline letter as I was not considered eligible. Finding the 2015 invoice I contacted the company requesting a re-assessment. Within the week a new walker was delivered to my home. So I got a new walker and am now safely getting around because I was proactive. My GP was amazed that I did this, but still does not know what to do for the next person who needs assessment for MOH mobility aids. Unfortunately, there is low or no confidence in the health system at this time from our members. Most folk can access the botox injections that are the main treatment for dystonia. It is those in the smaller regions of NZ that are sometimes struggling to get this treatment. Australian research is uncovering other options that would be most beneficial to some Dystonia patients. This includes surgery, medications and integrated health interventions. At present in New Zealand there are botox injections with no associated neuro-physio support; and Deep brain surgery (DBS), that is only helpful for certain types of Dystonia. Unlike some other chronic disorders there are no annual checks (except for DBS) with a movement disorder specialist or neurologist or any health professional.

	Quality of life is important for those of us with Rare Disorders; and yet this is becoming more and more elusive as the health system becomes more and more fragmented. Hope is a precious commodity and is in short supply, amongst those contacting our group. Keywords are isolation, frustration, loneliness, confusion, hopelessness, sadness, depression, grief, despair ... My hope is that those of us with RD will be listened to and that the changing health system will indeed change the way we experience our healthcare.
The Angelman Network	Dear Chris, Thank you for giving us the opportunity to provide feedback on the draft in-confidence "Rare Disorders Strategy for Aotearoa New Zealand" The Angelman Network is a family/whānau driven charitable trust based in New Zealand that connects and supports those impacted by Angelman syndrome. We are the only organisation in New Zealand dedicated to providing up-to-date information on Angelman Syndrome and this is 100% volunteer-based. We share this information on our website, social media sites and newsletters, and keep families connected and supported through social media, family gatherings and fundraising activities in NZ. This is a massive undertaking for parents to manage on top of their own stressful lives. Our organisation has established a proactive international network of Angelman support organisations, using social media as the tool for capturing family engagement on a global scale. The Angelman network is also credited for establishing International Angelman Day – an awareness day celebrated globally for the past decade. A brief outline of Angelman syndrome: • Angelman syndrome (AS) is a rare condition that occurs in approximately 1:15,000 • It affects the 15th chromosome resulting in a severe reduction of expression of the gene UBE3A in the brain. UBE3A (Ubiquitin Protein Ligase E3A) is a Protein Coding gene. • AS is not a degenerative condition. With a healthy and well managed lifestyle, individuals with AS have a normal life expectancy but will require life-long care. • AS affects males and females of all racial/ethnic groups, equally. • It was only in the early 1990s that genetic testing was developed for what is now known as Angelman syndrome. • Angelman syndrome is a neuro-genetic disorder that affects the nervous system causing physical and intellectual impairments. • AS is caused by a severe reduction of expression of a single gene UBE3a in the brain. UBE3A is a ubiquitin ligase whose function and targets relevant to AS are still unknown. • AS occurs through 4 different genetic mechanisms, all involving chromosome 15. • In about 80% of cases clinical diagnosis can be confirmed by laboratory testing. Most cases are sporadic but some genetic mechanisms have implications for other family members. Our responses to the questions posed, are in BLUE below: • Are there additional points that are important to capture? 2 1. In the background (why have a rare disorders strategy?) We fully support a rare disorder strategy for NZ 2. In the proposed definition (what are rare disorders?) We agree with the terminology, Rare Disorders We are interested in an additional definition of ultra-rare disorders of 1/50,000. Angelman syndrome is caused by Deletion, Mutation (UBE3A), Uniparental Disomy (UPD), or Imprinting Defect (ICD). The latter 3 are much rarer genotypes that could potentially be viewed as separate rarer disorders in the future. • Do you agree with the key elements (aims and principles) proposed for a strategy? Yes, we agree. • Are there additional opportunities that could be built on? (Doing the work to develop a strategy) See below _____ Capturing voices of the Rare Disorders community Rare Disorders New Zealand (RDNZ) have set out seven priorities their community has identified to improve health and wellbeing for people living with a rare disorder: We agree with these 7 priorities, and have been

	working on some of them independently already. • Diagnosis – early and accurate diagnosis of rare diseases We hope this will include a focus on newborn screening unit to identify rare disorders, as soon as any new testing becomes available. Many people with AS are misdiagnosed as having Autism, CP or simply 'global developmental delay' due to no specific genetic testing being carried out. Testing for newborn screening is currently being researched. • Planned pathways for clinical care – coordinated and integrated pathways for cohesive healthcare We hope this will include: • the coordination of specialist Clinics for specific disorders in the 3 main centres – Auckland, Wellington, Christchurch. • Early access to AAC – communication devices, and ongoing parent training • Equity of access to, and outcomes from, specialist clinical services • Consistent data collection through all parts of the health, disability, social services and education systems, to establish a natural history study of AS in NZ 3 We are currently looking at setting up Angelman clinics in NZ and have recently been communicating with larger AS Organisations in Australia and the USA, as well as Cystic Fibrosis NZ, to plan how to best go about this. This is an important priority for our families in NZ yet it is almost impossible to set this up from scratch from the 'outside'. Ideally this should be initiated by a key coordinator based at one of the main Hospitals/medical schools/Universities. • Access to disability and social support – implement simple mechanisms to ensure appropriate access to disability and social supports • Disability and social support mechanisms are complex and time-consuming to coordinate, and do not take into account the acute stress that families caring for someone with AS, endure. • A recent FB poll indicated that our families are most desperate for more respite opportunities. • Sleep deprivation is a massive burden in our community, yet very little overnight opportunities are provided to help resolve this. • There is still no crisis call centre to support families in crisis. • Rare disorder medicines – equitable access to modern rare disorder medicines through a specific assessment pathway It is still unclear to us, how quickly Angelman patients will be able to access any new therapeutics (FDA approved), here in NZ. With several trials underway overseas, we wish to be better informed of the process to gain access, should any new therapeutic become available overseas. • Research – coordinated and funded programme of research for rare disorders If we had a NZ Angelman registry and AS clinics in the main centres in NZ, these clinics have the potential to become trial sites for research. This would create more interest for clinicians, plus pharma companies are looking for sites outside the USA • National rare disease registry – capture relevant data on rare disorders in New Zealand It would be good to know how long it would take to implement this. It seems to be virtually impossible to set up an Angelman registry in NZ without an 'insider' in the NZ hospital system/Universities, so we are currently in discussions with FAST AU, to use the Global Angelman Registry as a platform for our own NZ Angelman registry. • Workforce development – planned training on rare disorders for health professionals and support staff. Once we establish our own Angelman clinics with the support of the Angelman Syndrome Foundation in the USA, participating specialists could join the online ASF Learning Network for Clinicians (80 - 90 clinicians attend these group calls each month). Recordings are also available. • For each the focus areas being explored on pages 8-10: Data and Measurement 4 We will coordinate our Angelman data to match that of our overseas counterparts. Digital Solutions and enabling We will adapt Angelman syndrome guidelines/standards of care, sourced internationally Critical Health Services We will need to build capacity and capability in these service areas Disability and whanau support and coordination Access to adequate supports is particularly difficult for our families, especially for respite. It will be essential to have a social worker attend the Angelman clinics, to ensure all families are given the information they need and are able to access the supports they are eligible for.

	Knowledge Building This is vital and should be more consistent across the country. Based on the population concentration, Auckland should be acknowledged as 4 separate zones (North, South, Centre/East, and West) so that services can reach communities in all 4 zones. Funding, assessment and prioritisation For people with Angelman syndrome, access to regular, ongoing therapeutic services and equipment is inconsistent across the country and also poorly coordinated, yet these services are potentially more important to support pae ora, than any other health services. There is also no good reason why coordinated therapeutic services should end, when the person with AS leaves school. Research and Evaluation The global Angelman community is very aggressive in its research programmes. We (The Angelman Network) are a member of the Angelman Alliance – a research collective based in Europe. We are also closely affiliated with the two main groups in Australia, and in the USA. We would like to be able to participate in clinical trials, but first need our own registry and clinics in NZ. _____ o Do you know of work underway that could address this? The global Angelman community is very proactive and supportive, and The Angelman Network is a well-known entity in this global community. We can connect with larger overseas organisations for advice, to collaborate, to share resources, replicate strategies, and even to source funding. We are already working on setting up our own registry and clinics, as we are aware of how long it could take for NZ to get this strategy in place. o How important is it to achieving pae ora for people and whānau with rare disorders? Of utmost importance. Our total NZ population is such a manageable size - it seems somewhat disappointing that we still don't have efficient systems in place. 5 o How easy would it be to make progress? Are there significant barriers? The only significant barrier is the time taken to adopt the strategy and then get it into motion. We can accelerate the implementation process by coordinating collaborations and proactive networking within the Rare Disorders groups and the Health system. Cultural barriers do play a role, but these can be overcome by working directly with local communities. o How long would it take? Once approved, it should be relatively quick and easy to replicate the implementation process of the Rare Disorders strategies, as are currently in place in Australia, Canada, UK. • What important possibilities are not yet identified? Having a national strategy is one thing; implementing it is another. The role of seeking out and coordinating families with a rare disorder, and encouraging them to actively participate in this new national framework, should not fall on the shoulders of volunteer parents who manage small support organisations. This duty should be covered in paid roles within the Health sector. Informal FB poll for mothers caring for a child with Angelman syndrome - July 2023 This feedback has been provided by Ms Ursula Christel Founder and Chairperson, The Angelman Network
AusEE Inc (Eosinophilic disease)	Thanks for reaching out and apologies for my delay in responding. We have our National Eosinophilic Week coming up 6-12 August and things have been extra busy with preparations! I have reviewed the document and it looks good. I've also virtually met with Sarah Vane from NZ (included in this email) to discuss it and she has reviewed it as well. In Australia, I was part of some working groups for the development of The National Strategic Action Plan for Rare Diseases (https://rarevoices.org.au/action-plan/) which I'm sure you are well aware of the contents of it and how Australia continues to use this resource in collaborative efforts to have our voices heard.

	I look forward to following the progress of the Rare Disorders Strategy for Aotearoa New Zealand. From NZ group lead: *Needed a glossary of terms eg what is pae ora – is it just overarching healthy future?* *Whaikaha – ministry of disabled people* What a strategy will provide - *The strategy is being developed in partnership with health entities and Whaikaha, as well as with RDNZ and experts from clinical, science, cultural and lived experience viewpoints. The aim is to bring all capability together and ensure that future possibilities inform our efforts to improve health system responsiveness and work towards pae ora for people and whanau with rare disorders.* This is of particular interest as you may well find / know that clinicians & agencies have very little knowledge, interest or understanding of a rare disorder they come across. Here may find best leads within the consumer community who may have good links internationally. This is not an unusual situation but health providers are not necessarily open to listening, 'sharing the table' so to speak or still think they know who best to talk to – it may not be that buddy in the US or who met at conferences – then again it might – that needs to be an open discussion. This has greater implication for Pharmac – when looking at medicines that NZ apply for initially via specialist – look locally to comment – spec ref, pharma distributors or patient knowledge? For a condition that requires treatment that is unsubsidised or effective – how is that defined? Careful not to be too pedantic about 1 in 2000 especially since there can be a sliding scale within a condition – one end could be managed reasonably well (whilst remaining possibly incurable & requiring life long treatment) or severe & refractory, almost placing that group of individuals in a subset of rare of rare. Plus there's the ol' was it there already & testing improved vs there's been some real growth. Maybe even numbers grew but due to 'new' nature of conditions coming onto scene & diagnostic tools then the same care needs to be taken due to lack of know / expert / medications regardless if suddenly get >1 in 2000. Doing the work to develop a strategy Identifying key opportunities that could be built on

	Totally agree, esp with more co-op & collab – less silo, less admin, less geog boundaries -really only way NZ can move forward & reflect our economy of scale (health economics) but we therefore must be aware how we work in that environment, what we offer. This also translates to maybe considering more resource to centres of excellence rather than spread across the nation – as long as all post codes have access (ie not turned away or denied as is regularly & currently done). Even with protocols, I have seen them roll out & not followed, guidelines written are cherry picked to avoid having to deliver a suitable internationally recognised minimum – some real accountability has to go behind 'best practice' & clinical ref groups who design these need to have consumer representation (that's part of co-design a bit as well) rather than hide (or maintain arrogant / power play) to make effective & possibly simpler / common sense steps forward – 'don't be frightened & closed off'. <u>Capturing voices of the rare disorders community</u> Access to disability and social support – this impacts who whanau, recognising hauora impact impt as this has multiple long term costs – either emotionally, financially, physically on other carer/family members. <u>Inputs that are underway or planned</u> *capturing Māori perspectives on life for whānau with rare disorders, how whānau ora and pae ora can be promoted for whānau with rare disorders, what is most important and what the health system can do better* - Maybe special qualities to look at here but plenty within general rare community also not addressed (some may be different, many will potentially overlap) An area that I am noticing more is the transition from paediatric to adult – compounded by maybe more & new conditions moving across ages where services can be less frequent, personal & seem to require 'adult' only without understanding complexity requires family continued involvement (which is strange, considering the specialist may have little knowledge themselves but expect young adult to cope on own). I think rethinking a more open approach to families transitioning is required (maybe there is some work in the disability field on this that could be seen as comparable?) There is also something to be said for continuity of care of personnel as well if possible. Maybe more family/consumer presentations at under grad level could help or spots within their medical conferences or regional presentations (some have satellite medical school summary multiple topic nights)?

	This also links to the primary to secondary care gap in both understanding & referral. There needs to be an acknowledgement re: ability & care that can be provided realistically by both, not to use this as a delay tactic or simply ignore – a heightened duty of care might be applicable. This leads onto public vs private, I have noticed more are being forced into private care specialist appointments to see the very same people. They can see them more quickly & for longer. It could end up with mix public & private. Not only is this a financial & access barrier it is messy & dilutes the possibility of holding centralised information required to get an accurate picture of the rare community & its needs. Health navigators / co-ordinators have always been a gap in service for many, many years, as have social workers or councillors. Surely, we could simplify? If we see cancer support, transplant etc – why doesn't this cross over in some even generic form esp when we might be talking very low numbers with no condition specific organisational support, mentoring or funding. Preferably without it taking too long to dev & get through. https://rarevoices.org.au/action-plan/ There was quite a bit under this, I haven't been able to read it all. I imagine those involved will read numerous overseas approaches & strategy docs. or the UK (framework, action plan)/ Canada where looking at more similar health funding models, then the more progressive nations … etc

Appendix C

Rare Disorder NZ's Support Group Leads' summarised comments on what is going well, what the barriers are, and what they wish to change for rare disorder care in New Zealand, grouped by Rare Disorder NZ priority area.

Priorities	Going well	Barriers	Wishlist
Diagnosis	• Introduction of newborn screening (SCID) • Nearly all children with PWS are diagnosed in infancy now.	Access to testing: • No newborn screening for the disorder • Poor availability of genetic counselling and testing for families. Misdiagnosis/slow diagnosis delaying treatment and causing morbidity and mortality. • Delays/wait time in the public system, declined referrals • Difficult to access a specialist for diagnosis outside of main centres • Lack of awareness of early signs and symptoms particularly in primary care • Delayed diagnosis leads to uncertainty, frustration, and stress.	• Resources to help the medical team recognise the disorder • An easier diagnosis process in the regions. • Promotion of early warning signs to GPs and the general public to facilitate early diagnosis and treatment • Clear pathways to early diagnosis and for the first steps after diagnosis- referrals, medicines, distribution of information for people with a rare disorder and their care team.
Planned pathways for clinical care	Access to a coordinator and/or specialist: • Community based specialist nurses (DEBRA) • Nurse specialist to help with care co-ordination (Adults with glioblastoma in Auckland and Wellington) • Contact details to access specialist support outside of appointments	Variable care across the country: • Lack of regional availability of treatment administration (e.g. Botox for dystonia) • Inconsistent treatment and standards of care, with those outside the main centres being particularly negatively impacted.	Coordination of care: • Specialised MDT clinics and a MDT approach to treatment and long-term care, as well as ongoing monitoring • Better systems for interdisciplinary communication • Transition from Paediatric to Adult services is planned for and supported

	(e.g email to paediatrician, nurse specialist) • Paediatric liver transplant service is a national service and works well for people with a diagnosis • TS Australia has a nurse who provides advice on the phone or online. US, Australia and UK have TSC clinics in some areas. Dedicated clinics and care coordination: • Dedicated clinics in the main centres (CF) • A high level of wrap around care from specialist teams (Children with high-grade brain tumours at Starship and CHOG) • Early intervention centres in Christchurch and Wellington (The Champion Centre and WEIT), enable families to attend multiple therapy appointments in one visit, as well as make connections with others • PWS clinic days where peoples can see their endocrinologist, developmental paediatrician and sometimes a dietitian in one visit (Keneperu) • Starship and Christchurch Hospitals have knowledgeable teams who are treating children with TSC according to international best practice	No specialist oversight of the management of the disorder: • No standard of care document/national protocols for the disorder or only guidelines for children but not adults • No systems in place to ensure surveillance and management guidelines are implemented. • Best practice not known about/followed. Some planned pathways of care exist, but these are not being used, or even not being believed • Lack of expertise overseeing care, parents remain the main health advocate for their adult children • Unwillingness of/lack of interest from the medical team to work with overseas specialists and/or consult and follow international best practice guidelines • Available treatment not started promptly or at appropriate doses following diagnosis • Transition from paediatric to adult services is not supported and at times the only ongoing care and monitoring is from the person's GP. No pathways or care coordination for rare disorders:	• A dedicated health professional/point of contact and coordination for the person/family. • A rare disorder card for the person to carry/Disorder fact sheets supplied to GPs and ambulance. • Health system navigators/co-ordinators • Inclusion of social workers and/or councillors • accessible diagnostic and clinical standards, guidance and pathways for health practitioners integrated with patient health records. • reminders such as the cervical screening register provides to help patients and their clinicians keep on top of the recommended surveillance. A rare disorder Centre of Excellence or central hub of information and oversight: • Organisation that provides people with a rare disorder and doctors with information about the disorder • Development of comprehensive standards of care and treatment guidelines reflecting international evidence to apply consistently across NZ, including for allied health, that recognise and mitigate challenges faced at different stages of the disorder • Standards of care accessible in emergency departments and fast track protocols for some presentations

	guidelines with neurologists or paediatricians managing care • MDT pain program (CRPS) • Emergency management (at Starship) by a team familiar with the condition and history. Access to clinical trials and new advances in best practice: • Access to a clinical trials through international collaborations (children with DIPG/DMG at Starship). • Access to advanced molecular diagnostics (such as NGS) through international collaborations (Children with high-grade brain tumours) • An international network of specialists, researchers and people with the disorder groups who share their expertise and experience (TSC).	• Unwillingness of the medical team to recognise a different approach to standard practice may be needed for rare disorders • Being slotted into a health system people don't feel they fit in/being lost in systems • Lack of allied health support (e.g. SLT for school aged children with PWS) • Difficult to access mental health services, especially with expertise in the disorder • Medical professionals work in silos and don't communicate with each other.	• Annual check up with specialist/s clinic up to date with research and trials and clear pathways for coordinated care • Engagement in medical advisory teams, recognition of the person with a rare disorder as an expert • Nationwide ordering and direct distribution process for specialised products necessary to manage the disorder (e.g. specialised bandages) • facilitation of social connections for the parents and children at specialised clinics/allied health appointments • Collaboration between countries, utilise work done and expertise overseas, medical teams who participate in networking with, and training from, international experts • A complaints system that is timely and appropriate • A mobile clinic made up of a specialised team of experts who can travel throughout the country (including rural) to diagnose affected people • Provides a platform for knowledge-sharing, training, and capacity building within the healthcare system.
Access to disability and social supports	• Individualised funding is working well • Community events • Active support groups.	Services not designed for the rare disorder population: • People not being believed by government agencies providing support due to the hidden nature of the disorder	Information: • More information about what support is available and where to access it • Information about what you should be able to apply for with a given disorder and when to do it.

		• Lack of understanding of the disorder leading to having to repeatedly explain the disorder, justify and apply for everything • Long waitlists for support, then support provided is generic advice not tailored to the disorder (e.g. behavioural support) • Lack of support at times of transition like leaving school. Administrative challenges: • Multiple forms to fill in and repeated visits required to access financial support • Inequity in accessing financial support because of varied health literacy • Annual renewals of funding requiring doctor's certificates when the disorder is life-long • Referral pathways to get support and equipment are poorly understood including by medical professionals. ACC: • Treatment injury claims rejected • Lack of funding from ACC due to lack of understanding of disorder. Schooling:	Workforce education/development: • Recognise invisible disabilities in those with rare disorders • More awareness of rare disorders • Specialised behavioural support for children with disabilities (not through CAMHS) from people with knowledge of evidence based approaches for the disorder • Education around the strategy filtering down to those who encounter the public face to face. A simplified and more coordinated pathway for people with a rare disorder to access disability and social supports: • More recognition and support • Support to attend appointments • Streamlined process for access to financial support for those with rare disorders. Once diagnosed with an incurable rare condition, people should not have to keep struggling to access funding and support • Better educational and community supports that are coordinated with stages of the syndrome and health needs - a holistic approach to care • A nationwide, simplified pathway to access disability support (including mobility equipment, home modifications etc) • Clinical care pathways include a checklist for medical practitioners to ensure that

		• School does not understand the disorder, hard to find information to support teachers and student to enable learning. • Lack of learning support funding (High Health Needs funding does not include learning support). • More access to special residential schools/special education units. Disability Support Services: • Long NASC wait times for needs assessments (particularly Auckland and Wellington) • Lack of respite options especially longer than one night and of high quality • When using IF, difficulties finding and employing support workers for older children/adolescents with health and behavioural challenges • Funding for disability supports insufficient to live a close to normal life • No professional support for siblings. Residential care/supported living • Long waitlists to be assessed for supported living / out of home placements and then waitlists to find a place once funding awarded • Lack of access to hospice beds, dementia level care.	other agencies involved in their patient's life have been informed of where to go for information to receive basic training on health risks to be monitored or managed • A mechanism in their care pathway following diagnosis that ensures an entitlement to some learning support (the level of which would vary per individual pupil). • Early access to AAC – communication devices, and ongoing parent training.

		• When intellectually disabled adults are living in residential facilities, visits to a GP or health practitioner are often reliant on there being proactive and knowledgeable support workers or involved parents with guardianship orders in place.	
Rare disorder Medicines	• New medicine funded.	• Treatment used internationally not accessible here or not funded by Pharmac, or very limited criteria, or not funded for that disorder • Difficulty accessing newer medicines with lower side effect profile • Pharmac determines that clinical trials are necessary to fund treatment. These will never be done because of the rare nature of the disorder • pharmacogenomic availability exists in New Zealand but not in the public health system • Pharmaceutical companies not applying for Medsafe approval for a drugs for rare diseases. • insufficient and inefficient processes and timelines at Pharmac.	• Improvements in access and funding for rare disorder medicines. • Development of an NZ medicines strategy including how Pharmac will address funding for modern medicines given the increasing importance of these drugs. • Standard of care guidelines that incorporate the use of modern medicines. • Different criteria to be considered when making decisions about funding medicines for rare disorders • A review of the current practices, strengths, weaknesses and opportunities within Pharmac in relation to medicine access for rare disorders. • Experts in the relevant rare disorder (whether local or international) are given a seat at the table for all rare disorder Pharmac applications • Easier, faster and proactive evaluation of medicines by Pharmac and Medsafe • Consideration of interim approval for rare disorder medicines while further data is awaited

			• Accessible information about, and transparency around, the process to gain access, to new therapeutics as they become available overseas • An interim solution for people to access rare disorder medicines while further work is undertaken to improve processes.
Research	• The Centre for Brain Research in Auckland runs research clinics for people who have Freidreich's Ataxia • An international group who conduct research to improve the lives of those living with TSC.	• No registry makes it difficult to get research participants • Overseas research not having an impact in NZ • Clinical trials not happening in NZ. • Not having a registry or specialist clinic in NZ is a barrier to NZers being included in clinical trials.	• More research • A coordinated and funded programme of research for rare disorders • International collaborations are encouraged in order to allow clinicians to access knowledge, promote trial participation and to enable evaluation of effectiveness for Māori and Pacific populations • With improved data, researchers would be encouraged to more easily include New Zealanders in studies initiated overseas • Research findings are disseminated.
National rare disorder registry	• Unique database (overseas) allows connection with others • FACSNZ worked with Manata Hauora to get FVSD on ICD-11-AM (used in NZ). FACS and FVSD are on SNOMED and SNOMED international. This allows data collection.	• Lack of a register of people with the disorder e.g. Lack of a brain tumour registry, which is essential for research and resource planning.	• A comprehensive national register for all rare disorders allowing participation in clinical trials and the capture of relevant data for global research • Relevant confidential data is accessible to researchers • Consistent data collection through all parts of the health, disability, social services and education systems • data collection would be to identify gaps in order to improve care and assist in

			identifying stages/ages where where the highest risk factors exist.
Workforce development	• Rett NZ biennial conferences (support group led) • National/international awareness days for disorders.	Workforce knowledge: • Little known about the disorder • Medical team not up to date. Workforce capacity: • No or few specialists in NZ • Insufficient imaging capacity across the country (MRI, PET scans) • Lack of advanced molecular diagnostics capability (NGS) • Insufficient number of specialists.	• Increase the engagement with people with a rare disorder e.g. conferences and medical students • Planned training on rare disorders for health professionals and support staff • Specialists in leadership positions aid dissemination of up-to-date best practice • Utilisation of technology to connect via virtual networks, clinicians seek and learn from international expertise, and have the resources required to do so • A compulsory requirement for residential service provider organisations who have full duty of care to receive training in a rare disorder such as Prada Willi Syndrome, and to repeat this frequently due to rapid staff turnover.

Notes:

1. Support group leads' responses are sourced from a survey carried out by Rare Disorders NZ in May 2023 (noted in black), and support group leads' July 2023 responses to the Ministry of Health's consultation document titled, 'A rare disorders strategy for Aotearoa New Zealand - Initial proposed content and points to consider and provide feedback on' (noted in blue).
2. Some responses have been summarised and grouped to highlight the key insights and themes.

Appendix D

Rare Disorders NZ

Priorities for improving the health and well-being for people living with a rare disorder.

The rare disorder community has collectively identified seven strategic priorities to improve the health and well-being for people living with a rare disorder.

1. DIAGNOSIS

Early and accurate diagnosis of rare disorders

- Ensure diagnosis of a rare disorder is timely and accurate.
- Equitable access to a range of diagnostic tools/tests, supported by policy.
- Ensure the newborn screening program is robust and funded to keep up with international best practices.
- Expanding newborn screening program
- Ensure a pathway for carrier testing. This involves testing people who are known to be at increased risk of being carriers of a specific inherited disorder. This may be because a relative is known to be a carrier or has the condition or certain genetic conditions might be more prevalent in their community.
- Ensure a pathway for genetic testing once a patent presents with undiagnosed symptoms.
- Putting protocols in place to identify people with no diagnosis, ensuring that a lack of diagnosis does not create a barrier to treatment.
- Making high-quality diagnostic tests accessible through common, clinically agreed systems or pathways.
- Develop policy that supports timely and equitable access to new and emerging health technologies.
- Those with an increased chance of being carriers of rare disorders have equitable access to peri-conception genetic testing and counselling and provide women with certain chronic conditions, thus an increased chance of having babies with rare congenital anomalies, with access to pre-conception and peri-natal care.

2. PLANNED PATHWAYS FOR CLINICAL CARE

Coordinated and integrated pathways for cohesive healthcare

- Develop a care pathway for rare disorders including diagnosis and genetic testing for people with no diagnosis.
- Develop standards of care documents for specific rare disorders to be implemented within the health system.
- Services support people living with a rare disorder through life-stage transitions. Develop seamless pathways for transition, from childhood to adolescence, and on to adulthood and older age – access criteria and measures of quality and outcomes.
- Targeted awareness and education for people in their preparation for conception and pregnancy.
- People with an undiagnosed rare disorder are identified and have priority access to a specialised diagnostic response.
- Coordinated rare disorder care and support that is integrated, while being person and family-centred.

- Clear coordinated pathways throughout health, disability and other systems.
- Services respond to the changing needs of people living with a rare disorder and their families.
- Rare disorder care and support systems address mental health and wellbeing.
- Guidelines that address the specific needs of people living with an undiagnosed rare disorder.

3. ACCESS TO DISABILITY AND SOCIAL SUPPORTS

Implement simple mechanisms to ensure appropriate access to disability and social supports

- Ensure those with an undiagnosed disorder are included.
- Develop an easily accessible pathway to information on support services available to those with a rare disorder.
- Develop a pathway for those with a rare disorder within the education system.
- Develop a pathway for those with a rare disorder leaving the education system.
- Remove the administrative burden of proving the disorder is ongoing for conditions that are lifelong.
- Ensure that people with rare disorders that include both health conditions and disabilities and/or hidden disabilities are able to access support that is comparable to people who have an isolated or more visible disability.

4. RARE DISORDER MEDICINES

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

- Future-proofing a pathway for new and innovative modern medicine for those with a rare disorder for example gene therapy.
- Update the factors considered in cost-benefit analysis to include the wider health and social support system cost.
- Make sure PHARMAC is funded adequately to be able to fund new and innovative modern medicine.
- Develop a medicines strategy to include rare disorders, gene therapy and innovative modern medicine.
- Develop policy that supports timely and equitable access to new and emerging health technologies.
- Ensure people living with a rare disorder have equitable access to medicines with demonstrated clinical benefit for a rare disorder,
- Equitable access to the best available health technology.

5. RESEARCH

Coordinated and funded programme of research for rare disorders

- Develop a national research strategy for rare disorders
- Develop a pathway for those with a rare disorder to participate in both national and international research.
- Invest in rare disorder research in New Zealand
- Translate research and innovation into clinical care; clinical care informs research and innovation.

- Support partnerships between researchers and clinicians in research into rare disorders.
- Support clinical teams to collect and input data, contributing to research and evidence-building.
- Ensure that Māori and Pacific peoples are involved at every level of development, implementation and governance of genomic research as per *Te Mata Ira* guidelines.
- Develop policy which maintains sovereignty for Māori over their health data to ensure that Māori and iwi aspirations are realised.

6. NATIONAL RARE DISORDER REGISTRY (DATA COLLECTION)

- Capture relevant data on rare disorders in New Zealand this could include.
- Get **Orphanet Coding (ORPHACODES)** into the health system. This can be done in a way that is compatible with other key coding systems (e.g. SNOMED, ICD-10/11)
- Where appropriate, support the availability of computerised prompts to help GPs diagnose a rare disorder when a rare disorder has not previously been considered.
- Ensure that the registry mirrors the ethnic demographics of Aotearoa so that it is generalisable and can aid in the diagnosis of Māori, Asian and Pacific peoples.

7. WORKFORCE DEVELOPMENT

Planned training on rare disorders for health professionals and support staff.

- Create effective rare disorder clinical networks that connect to international research and best practice.
- Develop a pathway to work with international partners wherever possible.
- Identify existing gaps in the rare disorder workforce and develop a national rare disorder workforce strategy.
- Equip and encourage frontline health professionals to consider, investigate and refer for a potential rare disorder diagnosis.
- Embed rare disorders in medical training, including diagnosis pathways and undiagnosed ones.
- Promote healthcare professionals to 'think rare' when presenting with symptoms.
- Promote healthcare professionals to take the 'whole picture of a condition into account when seeing different specialists
- Strengthen healthcare professionals understanding of tikanga Māori especially pertaining to whakapapa, human tissues and genetic material.

Increase awareness of rare disorders within the health system and to the public

- Increase every New Zealander's awareness of rare disorders.
- Awareness of relevant prevention measures.

- Targeted awareness and education for people in their preparation for conception and pregnancy.
- Awareness and education about testing and screening opportunities.
- Awareness about rare disorder services.
- A multi-purpose digital repository to detail available services and provide information.
- Strengthen genetic literacy in Māori communities.

Action Plans - implementation

Appendix E

A New Zealand Rare Disorder Centre of Excellence – moving the Strategy forward

As we look ahead to the implementation of the National Rare Disorders Strategy and how resources available across the health sector can be used in efforts to improve health system responsiveness for people and whanau with rare disorders as soon as possible, Rare Disorders NZ has identified a key opportunity for maximum leverage and progress for rare disorders through the establishment of a Rare Disorder Centre of Excellence in New Zealand.

In a number of other countries 'Rare Disease Centres of Excellence' have been established that act as hubs of expertise and promote best practice, bringing together experts from multiple specialities to reduce the time to diagnosis and improve the availability and coordination of multi-specialty clinical care.

Strong support for a Rare Disorder Centre of Excellence was identified in the previously supplied May 2023 survey of RDNZ support group leads regarding what they would love to see change in New Zealand for people with rare disorders, as well as what the biggest barriers people with the disorder and/or their carers are facing.

Staged implementation

Rare Disorders NZ would support a step-wise approach to the establishment of such a Centre of Excellence in New Zealand that can begin to be implemented now, and is also aspirational for the future. This approach recognises there is an urgent unmet need in the rare disorder community that requires immediate action and would not only be a huge relief to rare disorder patients that have waited so long for a responsive approach to their healthcare, but would also relieve the overburdened health system of many of these cases taking up unnecessary time and resources.

Our proposed first stage of a Centre of Excellence would align with current workstreams. Te Pae Tata Interim NZ Health Plan 2022 identified the establishment of regional and national networks as a key step in removing unwarranted variations in access to care, waiting times and clinical practice. Te Whatu Ora have advised these networks will focus on developing national standards and models of care, identifying ways to address variation in service quality and outcomes, addressing equity, and developing innovative, efficient, and evidence-based solutions that will inform investments and workforce planning and be applied nationally[i].

The first stage for a Centre of Excellence would be for it to serve as a national hub that develops and holds resources and pathways related to the diagnosis and management of rare disorders, including an information bank of specialists for clinicians to refer to while the centre becomes established.

A large number of rare disorders have existing standards of care or clinical guidelines internationally, and the initial role of the Centre of Excellence would include identifying and adapting these documents for the NZ setting. This work would result in the development of comprehensive standards of care and treatment guidelines linked to people's electronic health record that are applied consistently across NZ, including for allied health, that recognise and mitigate challenges faced at different stages of the disorder.

Other early steps for the Centre of Excellence would include collaboration with other countries, utilising work done and expertise overseas, and supporting NZ specialists to participate in networking with, and international training from, experts.

Filling gaps and meeting needs

Our aspirations for the Centre of Excellence are that it will eventually bridge gaps identified in our current systems by delivering expertise, ensuring care coordination, and assisting with navigation and/or providing the right services at the right time for people with rare disorders.

It would take learnings from the United Kingdom approach to rare disorders, the European Reference Networks, and Australia's Rare Care Centre to drive improved healthcare standards and better patient experiences for the rare disorder community.

---

[i] www.tewhatuora.govt.nz/whats-happening/what-to-expect/national-clinical-networks/#what-the-networks-will-do

Rare Disorder Centre of Excellence

At a glance

There are estimated to be around 300,000 New Zealanders living with a rare disorder.

Rare disorders can be complex, debilitating and life threatening, and often require a broad range of services.

Key stats

Rare Disease UK's (2018) *Rate your rare disease care* survey results found

> 50%
of people who had access to a specialist centre rated their care as 'good' or 'excellent'

For those respondents who had none of their care provided at a specialist centre

< 25%
rated their care as 'good' or 'excellent'

RARE DISORDERS NZ

PO Box 14-313, Kilbirnie,
Wellington 6241, NZ

www.raredisorders.org.nz

CHALLENGES

People living with a rare disorder often report being lost in the health system, with a long pathway to diagnosis, and difficulties accessing information, treatments, specialists and healthcare services. Challenges surrounding coordination of care are common.

SOLUTIONS

Other countries have addressed these challenges by establishing Rare Disease Centres of Excellence that act as hubs of expertise, bringing together experts from multiple specialties to improve time to diagnosis, create effective referral pathways and coordinate multi-specialty clinical care.

Effective referral pathways

Network of expert healthcare professionals

A central hub for coordinating multi-specialty clinical care

BENEFITS

1

Benefits One

Time to diagnosis is reduced and patient experience, well-being and overall quality of life is vastly improved.

2

Benefits Two

Patients will access appropriate care and treatment faster, lowering risk of becoming high-need, high-cost patients, and reducing the demand on health and social services.

3

Benefits Three

New Zealand delivers an equitable healthcare system that is able to keep pace with advances in genomics and precision medicine.

Centres of Excellence and reference networks for rare disorders around the world

"A Centre of Excellence is a specialist clinic where expert healthcare professionals come together to provide the very best care and treatment for rare disease patients. Our survey on rare disease care found that over half of people who had access to a specialist centre rated their care as 'good' or 'excellent'. For those respondents who had none of their care provided at a specialist centre less than a quarter rated their care as 'good' or 'excellent'"[1]

A Centre of Excellence can be 'virtual' with a network of experts across different hospitals or 'physical' with their own building[2].

This document outlines the different approaches that have been taken in Australia, the United Kingdom, Europe, and being adopted by the United Nations and Rare Diseases International, to address the need for Centres of Excellence for rare disorders.

Australia

The following information is sourced directly from a published interview with Dr Gareth Baynam, the Medical Director of the Rare Care Centre[3], and the Government of Western Australia Child and Adolescent Health Service website[4], and collated for the purposes of this document.

On 25 February 2022, a new Rare Care Centre was launched at Perth Children's Hospital, Australia.

Rare Care – the Clinical Centre of Expertise for Rare and Undiagnosed Diseases (Rare Care Centre) aims to help children with rare and undiagnosed diseases (RUD), and their families, live the best lives possible. The Rare Care Centre provides a cross-sector care coordination service which aims to holistically address the concerns or challenges experienced by children with RUD and their families across Western Australia.

The cross-sector team is designed to help bridge gaps through care coordination, navigation to the right service, integration of services and advocacy when needed, without duplicating any of the services already being received.

The Rare Care team includes:

- Perth Children's Hospital Doctors and Nurses
- General Practitioner (GP)
- Genetics Counsellor
- Mental Health Nurse
- Aboriginal Health Worker
- School Educator
- Welfare Support Worker
- NDIS Support Coordinator

- Department of Communities Coordinator.

Alongside its care coordination service, the Rare Care Centre also focuses on:

- Education and training
- Digital technologies
- Workforce capacity building
- Advocacy
- Peer to peer support
- Connection to global expertise, research and clinical trials.

The rare care centre takes a cross-sector approach that employs people from all the different sectors, within and beyond the health system and from all parts of the social and community sectors.

The co-design process included over 400 stakeholder meetings with people from all sectors - education, employment, community, disability, primary care, Aboriginal health, mental health and welfare sector. People from all parts of the community were engaged- young people, indigenous communities and remote regions in the Youth Reference Group, Community Reference Group and Non-Government Organizations. WA is the world's single largest public health jurisdiction, so it is critical to have a focus on remote and regional health.

Alongside its clinical service, the Rare Care Centre also has streams of work to build multi-stakeholder capacity, support health systems, and enhance equity and scale, including Education and Workforce Capacity Building; Digital Technologies and Devices; Peer-to-Peer Mentoring and Systems Advocacy; Global Partnerships and Connection to Global Expertise; and Research and Evaluation.

The Rare Care Centre has recently released its inaugural impact report[5]. The report highlights the key milestones, accomplishments, partnerships and new initiatives led by the Centre in the first year.

United Kingdom

In the UK there are Rare Disease Collaborative Networks, Specialised Services and Highly Specialised Services, as well as Reference Networks.

The Consultation on the UK Plan for Rare Diseases[6] was launched in February 2012, and a summary of the responses was published in November 2012. The consultation identified a need for Centres of Excellence. It found consensus among experts and people with rare conditions that expert centres caring for people with rare and complex disorders must have the following characteristics:

- co-ordinated care;
- adequate caseload for expertise;

- not dependent on a single clinician;
- arrangements for transition from children's to adults' services;
- engaged with people with rare conditions;
- research active.

Following this, Rare Disease UK (RDUK) collected data through surveys, interviews, and workshops, capturing a wide range of views from the rare disease community, including both patients and carers, as well as multiple stakeholders (clinicians, academic researchers, patient organisations and industry representatives). From this RDUK delivered a report[7], titled Centres of Excellence for Rare Diseases, which develops a broader understanding of what a Centre of Excellence should be and establishes criteria under which a centre can be classed as a Centre of Excellence.
In this report RDUK also added the following two points to the characteristics that expert centres caring for people with rare and complex disorders must have:

- Education and training for medical professionals;
- Membership of international networks of excellence.

The report's recommendations regarding a Centre of Excellence were the following:

- Centres of Excellence should play a role in undergraduate medical education, specialist training and in continued professional development.
- Workforce planning must ensure continuity of services.
- Centres of Excellence should ensure communication to local healthcare providers is of a high standard.
- Existing best practice in collaboration between Centres of Excellence in the UK and the rest of the world should be adopted by all centres in the UK.
- Resources should be allocated to support the valuable role of patient organisations in supporting and representing patients.
- Research at Centres of Excellence should develop all aspects of the patient pathway including diagnosis and treatment.
- All Centres of Excellence should either maintain registries for the diseases they treat or contribute to national and international registries as necessary.
- Centres of Excellence must ensure patients are fully informed about their condition and provide facilities for patients to access information on their condition.
- Every patient with a rare condition should have a named care coordinator within a Centre of Excellence to ensure they can access information and support as needed.
- Centres of Excellence should facilitate peer support for people living with a rare condition.
- Centres of Excellence should consider means by which they can lower the cost of a family attending an appointment.

Rare Disease Collaborative Networks (UK) - information presented is directly copied from the NHS website[8,9] and collated for the purposes of this document.

Rare disease collaborative networks (RDCNs) are an important part of the NHS architecture initiated by NHS England and NHS Improvement to improve care and support for patients with rare diseases. 18 RDCNs had been established to date across a range of specialties and disease groups.

RDCNs are expected to operate under the following principles:

- RDCNs are made up of groups of providers (Rare disease collaborative centres (RDCCs)) who have an interest in developing understanding of a particular rare disease and are committed to working together to progress research, increase knowledge and improve patient experience
- RDCNs should include providers that are research-active in a particular rare/very rare disease
- They are based on the principle that, when it is practical, 'the knowledge moves rather than the patient'
- They must be focused on rare/very rare diseases where expertise is likely to be scarce and where there may also be benefits in national and international collaboration
- RDCNs should describe and demonstrate a positive impact on patient experience and provide good geographical coverage
- RDCNs provide one of a number of tools for NHS England and NHS Improvement to take forward its rare disease agenda

The priority areas of existing RDCNs include:

- Raising awareness of the rare disease
- Improving co-ordination of care
- Sharing of expertise and best practice
- Establishing a disease registry to improve the understanding of the epidemiology of the rare disease
- Research on treatment options and diagnostics
- Establishing a support network for patients and families, including co-ordinated transition from paediatric to adult services.

Specialised Services UK - information presented is directly copied from the NHS website[10,11] and collated for the purposes of this document.

Specialised services support people with a range of rare and complex conditions. They often involve treatments provided to patients with rare cancers, genetic disorders or complex medical or surgical conditions. They deliver cutting-edge care and are a catalyst for innovation, supporting pioneering clinical practice in the NHS.

Specialised services are not available in every local hospital because they have to be delivered by specialist teams of doctors, nurses and other health professionals who have the necessary skills and experience. Unlike most healthcare, which is planned and arranged locally, specialised services are planned nationally and regionally by NHS England.

In total, there are 149 specialised services currently directly commissioned by NHS England. Three factors determine whether NHS England commissions a service as a prescribed specialised service. These are:

- The number of individuals who require the service;
- The cost of providing the service or facility;
- The number of people able to provide the service or facility.

The specialised services commissioned by NHS England have been grouped into six national programmes of care (NPoC). Each has an NPoC that coordinates work across the services in that programme of care.

The six NPoCs have been identified as:

- Cancer
- Mental health

The following four NPoCs are grouped together as acute programmes of care:

- Blood and infection– infection, immunity and haematology
- Internal medicine– digestion, renal, hepatobiliary and circulatory system
- Trauma – traumatic injury, orthopaedics, head and neck and rehabilitation
- Women and children– women and children, congenital and inherited diseases.

Each NPoC brings together clinical and commissioning leadership, an empowered patient and public voice, and policy expertise to:

- Contribute to the development and delivery of strategy and policy objectives, such as the NHS Long Term Plan.
- Support regions to commission specialised services which meet population needs, provide consistently high-quality care and excellent patient experience, as part of an integrated care system and patient pathway transformation.

The NPoCs principally operate through a network of affiliated clinical reference groups and task and finish groups.

There are three different models of Clinical Reference Groups (CRG), aligned to the programme of work within each service area. The models are allocated to each service area to ensure that commissioning support provided to CRGs is aligned to prioritised work programmes. Programme of Care teams agree which CRGs within their programme will operate inside each model and this is kept flexible, in line with the commissioning agenda related to that service.

Highly Specialised Services UK- information presented is directly copied from the NHS website[12] and collated for the purposes of this document.

Highly specialised services are provided to a smaller number of patients compared to specialised services; usually no more than 500 patients per year. For this reason they are typically best delivered nationally through a very small number of centres of excellence.

For example Alström syndrome service (adults and children) Alström is a rare genetic syndrome that usually presents with blindness in childhood. Patients go on to develop insulin-resistant diabetes, fibrosing cardiomyopathy (where abnormal tissue growing in the heart stops it working effectively) and renal failure. They may also become Deaf. Fewer than 100 people are thought to be affected by Alström syndrome in England. Both the adult and paediatric services run two-day clinics that undertake assessment of all patients in a multidisciplinary structure. Patients are assessed and reviewed by all the specialties appropriate to their needs during the clinic. A management plan is agreed and communicated to local care providers to allow their healthcare professionals to implement the recommendations and monitor patients' progress. Alström Syndrome UK support workers attend the clinic to provide advocacy and guidance on the social care aspects of living with the condition.

A full list of the highly specialised services and a brief outline of each is available here: www.england.nhs.uk/commissioning/wp-content/uploads/sites/12/2022/06/B1769_Highly-specialised-services-2019-20_June-2022.pdf

The Rare Diseases Advisory Group (RDAG) is responsible for making recommendations to NHS England and the devolved administrations of NHS Scotland, NHS Wales and NHS Northern Ireland on the development of services for people with rare diseases and on highly specialised services. RDAG makes recommendations to the Clinical Priorities Advisory Group (CPAG) about how highly specialised services should be commissioned, including providing advice on which services or technologies should be prioritised for investment. In addition, RDAG recommends the most appropriate model of provision for the service and which expert centres may (or may no longer) be nominated to deliver highly specialised services.

RDAG receives outcome information on the services and makes recommendations on any action required as a consequence of poor outcomes as well as ensuring proper provision of services commissioned, with equal access opportunities for patients across different geographies. RDAG makes recommendations to NHS England and the devolved administrations on developing and implementing strategy for highly specialised services including making recommendations on how the UK Strategy for Rare Diseases should be implemented.

The central ethos of commissioning highly specialised services is to concentrate expertise in a small number of expert centres. The trade-off implied by this ethos is that access may be difficult for patients who need to travel long distances to access care from the expert centres. Hence it is incumbent on the HSCT to monitor the geographical access to highly specialised services.

Europe

European Reference Networks - information presented is directly copied from the European Union website[13] and collated for the purposes of this document.

European Reference Networks are virtual, Europe-wide healthcare networks working to treat those with rare and complex conditions. They link hospitals and clinicians, allowing for cross border sharing of expertise and knowledge and helping to provide diagnosis and treatment for patients. They also help to facilitate European wide research projects and the development of new medical technologies and medicines. There are 24 different ERNs organised around different disease groups.

To review a patient's diagnosis and treatment, ERN coordinators convene 'virtual' advisory panels of medical specialists across different disciplines, using a dedicated IT platform and telemedicine tools.

The process and criteria for establishing an ERN and for selecting its members are set in EU legislation.

The first ERNs were launched in March 2017, involving more than 900 highly-specialised healthcare units from over 300 hospitals in 26 EU countries. 24 ERNs are working on a range of thematic issues including bone disorders, childhood cancer and immunodeficiency.

Resources regarding development of clinical practice guidelines and clinical decision support tools are available here: https://health.ec.europa.eu/publications/european-reference-network-clinical-practice-guidelines-and-clinical-decision-support-tools_en.

WHO and Rare Diseases International (RDI)

Information presented is directly copied from the RDI website[14] and collated for the purposes of this document.

The UN Resolution on Persons Living with a Rare Disease and their Families, 2021[15], Encourages Member States to foster the creation of networks of experts and multidisciplinary specialized expert hubs.

The Global Network for Rare Disease (GNRD) follows this Resolution. It is a joint initiative between the WHO and RDI engaging and supporting the rare disease community to develop a person-centred global network of care and expertise for all Persons Living with a Rare Disease (PLWRD) worldwide

The complex nature of rare diseases and their infrequency require an evolution in the model of care, from a multi-disciplinary approach to a "networked care" model, through which expertise can be accessed from across a national, regional and global network of experts to inform care locally.

The Global Network for Rare Disease aims to harness advances in digital technologies to foster the development of an international network to drive peer-to-peer learning and the sharing of knowledge, resources and capacities to strengthen health systems for rare diseases.

The components of the proposed network are:

- National Hubs – nationally endorsed centres connecting under a national hub-and-spoke model
- Regional Hubs – virtual multi-centre collaborative hubs of expert centres and patient organisations
- Global Network – a global learning system and knowledge-sharing network, informing global public health action and leveraging technology to extend care for rare diseases around the world.

References

[1] Illuminating the Rare Reality, Genetic Alliance UK, February 2019
[2] Centres of Excellence for Rare Diseases, Rare Disease UK (RDUK), October 2013, available here: www.raredisease.org.uk/media/1601/centres-of-excellence.pdf
[3] Rare Care Centre – A new holistic and cross-sector model of care, Global Expert Series, Rare Diseases International, January 2023, available here: www.rarediseasesinternational.org/wp-content/uploads/2023/01/January-2023-Gareth-Bynam-Experts-Series-Article.pdf
[4] https://pch.health.wa.gov.au/Our-services/Rare-Care-Centre
[5] Rare Care Centre – First year Impact Report – Update for Philanthropic Funders, Feb 2022-2023, acccessible here: https://pch.health.wa.gov.au/~/media/Files/Hospitals/PCH/General-documents/Our-Services/Rare-Care-Centre-Impact-Report-Y22-23.pdf
[6] Consultation on the UK Plan for Rare Diseases', Department of Health, February 2012, available here: https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/215141/dh_132883.pdf
[7] Centres of Excellence for Rare Diseases, Rare Disease UK (RDUK), October 2013, available here: www.raredisease.org.uk/media/1601/centres-of-excellence.pdf
[8] www.england.nhs.uk/commissioning/spec-services/highly-spec-services/rare-disease-collaborative-networks/
[9] www.gov.uk/government/publications/england-rare-diseases-action-plan-2022/england-rare-diseases-action-plan-2022
[10] https://www.england.nhs.uk/commissioning/spec-services/
[11] https://www.england.nhs.uk/commissioning/spec-services/npc-crg/
[12] https://www.england.nhs.uk/commissioning/spec-services/highly-spec-services/
[13] https://health.ec.europa.eu/european-reference-networks/overview_en#work
[14] https://www.rarediseasesinternational.org/collaborative-global-network/
[15]Addressing the challenges of persons living with a rare disease and their families, Resoluton adopted by the United Nations General Assembly on 16 December 2021, accessible here: https://digitallibrary.un.org/record/3953765?ln=en

Encouraging Inclusive Policy for Rare Disorders in New Zealand

A summary of discussions from a multi-stakeholder roundtable meeting, hosted by Rare Disorders NZ on 26th October 2022

People living with rare disorders are estimated to make up 6% of New Zealand's population.[1] However, this large community experiences substantial **unmet need** and **inequitable access to healthcare**.[2]

New Zealand is currently undergoing large-scale health reform. At this critical time, **New Zealand has the opportunity to become a model for how to address the unmet needs of the rare disorder community on a national level**. By combining learnings from other countries and capitalising on the nation's unique culture of valuing and prioritising equity in public policy, New Zealand has the chance to **transform the national health system in a sustainable way to improve the wellbeing** of people living with a rare disorder and their whānau.

This multi-stakeholder meeting brought together 19 international stakeholders to discuss challenges and solutions to achieving inclusive health policy for rare disorders in New Zealand. The attendees represented:

Healthcare professionals

Policymakers

Rare disorder advocates

Researchers

Patients & caregivers

Three key challenges were identified by the roundtable participants:

1. Poor visibility & awareness
2. Inequitable access to healthcare
3. Lack of a national rare disorder strategy

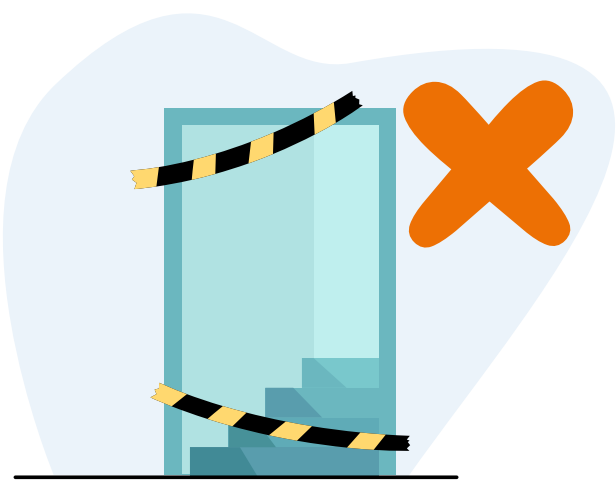
1. Poor visibility & awareness

There is a lack of awareness and understanding of rare disorders amongst healthcare professionals, health system leadership, government agencies and the general public in the New Zealand.

- **Inadequate research funding**
- **Poor care coordination**
- **Delayed diagnosis**
- **Lived experiences of people with rare disorders being questioned and undermined**

To improve the visibility and awareness of rare disorders in New Zealand, the roundtable participants recommended:

- Increasing high-quality data collection on rare disorders, and employing a national registry
- Utilising the ORPHAcode system[3] to encourage precise and accurate coding within electronic medical records
- Considering the needs of the rare disorder community in the context of other areas of national focus (e.g., mental health, chronic diseases, transitional care, indigenous health)
- Incorporating education and training on rare disorders into medical school curricula



2. Inequitable access to healthcare

People living with rare disorders in New Zealand do not have equitable access to healthcare. The roundtable attendees noted that comparison with other nations highlights just how poor access to care in New Zealand is for the rare disorder community. International comparison also emphasises how access differs between areas of New Zealand, which results in a "postcode lottery" of care.

To make access to healthcare more equitable for people living with rare disorders, participants recommended:

- Developing an effective interface between primary, secondary and tertiary care services, supported by online resources[4, 5]
- Setting up a national, Rare Disorder Centre of Excellence, to act as a hub of expertise and promote best practice
- Building partnerships within the rare disorder community and with Māori, Pasifika and other disadvantaged groups to strengthen advocacy for equitable access to healthcare
- Advocating for increased access to and investment into rare disorder clinical trials in New Zealand

3. Lack of a national rare disorder strategy

Despite advocacy efforts to increase recognition of unmet needs in the rare disorder community, New Zealand still does not have a national strategy for rare disorders. National strategies, supported by strategic frameworks, are essential for promoting positive change and ensuring a sustainable health system.

The roundtable attendees urged for concerted action by the New Zealand government to follow through with the development of a national rare disorder strategy. Throughout strategy development, they recommended:

- Looking to international best practices, using templates for Centres of Excellence abroad, and considering regional partnerships
- While international models are useful guides, a local approach should be developed including an understanding of Te Tiriti and cultural partnership
- Encouraging inter-agency and cross-sector collaboration, with the voice of the rare disorder community at the centre of the strategy development process
- Framing rare disorders as a collective group, focussing on the common challenges and experiences

New Zealand has the great opportunity of a new starting point; the country can learn from what has been done internationally, and do it better.

Gareth Baynam

Medical Director, Rare Care Centre, Perth Children's Hospital

Clinical Genomics Policy Advisor, Western Australia Health

Rare Disorders NZ, the only umbrella group for rare disorders in New Zealand, has a mission to amplify the community's collective voice to improve healthcare and wellbeing for people and their whānau living with a rare disorder. Their vision is to see New Zealand become a country where people living with rare disorders are fully recognised and supported, with equitable access to health and social care.

Developed in partnership with Costello Medical on a Pro Bono basis: www.costellomedical.com

References

1 Rare Disorders NZ. 2022. Available from: https://www.raredisorders.org.nz/about-us/news/largest-ever-survey-on-new-zealanders-living-with-rare-disorders-confirms-huge-health-inequities-for-this-population-group/. Accessed 29 November 2022.

2 HealthiNZ. 2022. Available from: https://www.raredisorders.org.nz/assets/VOICE-OF-RARE-DISORDERS-White-Paper-February-2021-FINAL.pdf. Accessed 29 November 2022.

3 Orphanet. 2022. Available from: https://api.orphacode.org/. Accessed 30 November 2022.

4 WA Primary Health Alliance. 2022. Available from: https://www.wapha.org.au/health-professionals/healthpathways/. Accessed 30 November 2022.

5 Rare Voices Australia. 2022. Available from: https://rarevoices.org.au/rare-disease-project-echo/. Accessed 30 November 2022.

Appendix F

Thank you for the opportunity to comment on "A rare disorders strategy for Aotearoa New Zealand – initial content and points to consider.

I found the document a rather diffuse and hesitant piece of work. It does not give a sense of awareness of similar activity across the world (even in Australia) and seemed to set a tone that was non-aspirational (e.g. predicting an inability to code, capture and count individuals with rare disorders was a particularly depressing prediction based on no evidence). There is every reason to expect that we can achieve this. Every rare disorder has a diagnostic code.
Most remarkably Rare Disorders New Zealand have laid out in stark detail 7 components of a strategy which they consider a well-considered framework. This is the current voice of individuals with Rare Disorders in New Zealand but this framework sits within the middle of document, prefaced by a statement that says these features **may be** considered in the work that will go into the formulation of this strategy. I am amazed at this framing. Elsewhere in the document much is made of listening to the community who live with these conditions and then this document says accounting for this perspective is somehow discretionary.

The document is particularly nebulous in what it promises to emerge with. The Aims are Health for All. This goes without saying. Some granularity about these Aims is required and needs to relate to specific, measurable and accountable outcomes in this strategy. This needs to be a document that speaks to those requirements.

Specific Responses to statements made:

- Are there additional points that are important to capture?

In the background (why have a rare disorders strategy?), the statement that "limited clinical knowledge can lead to misdiagnosis" is a truism but it is key to recognise that good clinical knowledge frequently exists – it is a case of organising a system to that the right patients get to those specialist people early in their journey. Access (through a rare diseases network for clinical diagnosis, care and management) to expertise is a direct consequential of this statement and it is illustrative of this document that rather vague impediments to the wellbeing of those with RD are alluded to generally but there is no specific Aim listed to address the problem. The Document needs to align such problems with a specific workstream that will emerge with solutions, educated by Māori perspectives and also experience offshore. For instance with regards to "limited knowledge" It is not necessarily increasing knowledge that is the response it is about improving patient flow and access to those that have it. We can set up a national clinical network for rare focused for instance on Rare Disease Diagnostic programmes overseas.

- Impacts

Borrowing numbers from overseas points to our lack of knowledge here. A body of work to obtain local figures is urgent for equity and planning purposes. I see no aim or ambition to address this data deficit except the point alluded to above which seems to speak to a failure to deliver in these respects even before we start.

The passage on "Unrecognised and Undiagnosed Conditions "relates to problems of generalists being the chief managers – referral pathways are absent and an undiagnosed diseases framework as exists in other countries does not exist. It is easy to frame GPs as ignorant, but this is hardly a solution.

The statement on "What is wanted from a rare disorders strategy?" Talks recurrently about support. This word is too general and does not provide focus or refer to an enunciated need in the RDNZ framework. We need to lay out the components that the strategy should contain including access to diagnostic services, clinical evaluation, therapeutic planning, and on-going management plans, all underpinned by a system

that enumerates the components of the system – the number of patients and the diversity of their conditions (including dissections via ethnicity).

- The section on "What a strategy will provide" is incomplete.

It states the areas of focus or action towards those aims (how it will be achieved) but stops short of citing milestones within a temporal window. A strategy speaks to action and measuring progress towards those aims. This document lacks a plan to evolve that as a Terms of Reference for this piece of work.

The proposed definition (what are rare disorders?) of Rare I think is quite good. We need to align with WHO. The Māori element is important and is untouched. Who are the Māori around your Table (I am not talking about salaried intramural Māori, I am talking about Māori with Rare). I have been involved in conversations on a WHO expert working group on Rare Diseases and their emphasis is on harnessing trans-national expertise. This is not mentioned here. It needs to be in the strategy.

- Do you agree with the key elements (aims and principles) proposed for a strategy?

As noted above I think that this is poorly formulated. Align the RDNZ framework with actions to speak to them and evolve a workplan to achieve that. Currently the Aims are too high flown and lack this granularity. For someone involved in the care of those with Rare Disease the action "give voice to" perspectives is rather galling. This is a starting point, not an aim and certainly not an endpoint. Action needs to follow these perspectives and there need to be several underpinning substeps to achieve this. These include – the formulation and implementation of:

  - Informatic strategies to count disorders and people with them. They exist internationally and our next generation plan for health data handling in this country is capable of incorporating it. Intersecting this workplan with those workstreams in the Ministry is essential
  - Create funded structures that can sponsor referral pathways and identified centres of excellence for RD diagnosis and management
  - Resource rare disease diagnosis, evaluation and management, particularly the data heavy and laboratory infrastructural requirements that underpin genomics and metabolic medicine.
  - ……all of the 7 points from the RDNZ statement that "should" form part of the strategy. Otherwise you are not acting on or valuing the voice of the rare community.

- The "Doing the Work" to develop the strategy looks good but it does not refer back to an architecture of what they are meant to be building.

- Are there additional opportunities that could be built on? (doing the work to develop a strategy)
Seeking international expertise, linking with cross sector workplans on data and technology.

- Do you know of work underway that could address this?

Undiagnosed disease networks across Australia, Australian Genomics, Genomics England. Speak to your experts in this country. They are aware of them all.

- How important is it to achieving pae ora for people and whānau with rare disorders? This is a galling question to ask those with Rare. The answer is a self-obvious yes. Please don't voice this sort of thing, because they have been waiting for action for years. To even intimate that their right to healthcare could be secondary or not a priority is really insulting.

- How easy would it be to make progress? Are there significant barriers?

That depends on Manatu Hauora!! The major inhibition is the reluctance to engage the voice of those with Rare Conditions, to reach out to experts at the clinical interface across New Zealand and to be brave enough to outline strategic Aims that will cost resources and time.

- How long would it take? With determination and the right mix of people around the table a year to formulate this Strategic Plan will be ample but only if the Rare Disorders Community are around the Table together with those providing clinical diagnosis and care. This timeline should also be enough to include consultation.

- What important possibilities are not yet identified? There is very little here that explicitly speaks to ensuring that resources to facilitate Rare Diseases clinical care is adequate. Infrastructure for genomics for instance requires considerable investment in data systems as well as laboratory equipment and human capability.

- There is substantial risk of re-inventing the wheel here when Australians and other similar nations to use have done a lot of this work already. Call upon their expertise and borrow extensively from their resources and documents.

Stephen Robertson FRACP FRSNZ
Professor of Paediatric Genetics
University of Otago
22 July 2023

A rare disorders strategy for Aotearoa New Zealand

Initial proposed content and points to consider and provide feedback on

We are interested in your feedback on any points in this draft paper.

Some particular questions we have are:

- *Are there additional points that are important to capture?*
  - *In the background (why have a rare disorders strategy?)*
  - *In the proposed definition (what are rare disorders?)*
- *Do you agree with the key elements (aims and principles) proposed for a strategy?*
- *Are there additional opportunities that could be built on? (doing the work to develop a strategy)*
- *For each the focus areas being explored on pages 8-10:*
  - *Do you know of work underway that could address this?*
  - *How important is it to achieving pae ora for people and whānau with rare disorders?*
  - *How easy would it be to make progress? Are there significant barriers?*
  - *How long would it take?*
- *What important possibilities are not yet identified?*

Why have a rare disorders strategy?

Our newly reformed health system is set up to work towards pae ora for all New Zealanders. This requires system responsiveness to and inclusion of many communities. A rare disorders strategy will improve system responsiveness and support pae ora for people and whānau with rare disorders.

People and whānau living with rare disorders often face significant challenges in getting timely, effective and equitable health care because their condition is rare. Limited clinical knowledge can lead to misdiagnosis, long wait times for diagnosis, and missed opportunities for early treatment and care. Lack of understanding of disorder progression and effective care and support can lead to inappropriate treatment and management. Limited research, with lack of economic incentives for research and development and high treatment costs, further restricts health system responses.

These significant challenges are barriers to pae ora for people and whānau, and can profoundly affect quality and length of life.

Impacts of rare disorders

Rare disorders have significant impacts for people, communities and the health system. For example, a 2010 study in Western Australia found that a cohort of people with rare disorders, representing 2% of the Western Australian population, accounted for 9.9% of hospital discharges and 10.5% of state inpatient hospital costs[1]. Many estimates of rare disorder prevalence are around 6% to 8% in populations.

In New Zealand, people with rare disorders responding to surveys report[2] rates of unhappiness, depression and anxiety that are disproportionately high in comparison with populations of disabled people as well as the general population. Income, education and other socioeconomic wellbeing indicators are lower and/or become lower over time. While health service utilisation is high and good experiences with health services are reported, many people report problems accessing sufficient or the right support, practitioners and services.

Awareness of rare disorders is considered by people with a rare disorder to be low among health practitioners as well as the wider community. Many people say they simply want to be believed when they tell practitioners about their concerns. Patients frequently face long delays before diagnosis that may mean the disease progresses before treatment can start; treatments may not be available in New Zealand; supportive or symptomatic relief is often limited; and coordinating care across specialties and carers is a very large barrier, especially for whānau who live far from tertiary hospitals or urban centres.

Unrecognised and undiagnosed conditions

Many of the challenges that come with rare disorders are also experienced by people with unrecognised or undiagnosed conditions – even though in some cases those conditions are not rare. Relatively common conditions, like foetal alcohol spectrum disorder or many neurodiverse conditions, have not been well understood by health practitioners nor been a high focus of research or service improvements until recently. Even well-recognised conditions can be difficult to diagnose and/or manage. A rare disorders strategy is not intended to further dilute focus on the high health needs that may come with less rare conditions. Rather, a rare disorders strategy will address the clear difficulties with providing healthcare for very many rare conditions that individual practitioners may see once in their career if that often.

An effective rare disorders strategy would support the system to better share and access knowledge, improve understanding and effective responses, and direct research and development efforts to areas of high human impact. In so doing, over time a rare disorders strategy should benefit all New Zealanders – especially people and whānau with rare disorders and others who, like them, do not yet get equitable health care or outcomes.

Te Tiriti o Waitangi responsibilities

As Tiriti o Waitangi partners we have a particular responsibility to ensure Māori aspirations for pae ora are honoured, that Māori determine what is important to them and that Māori whānau living with rare disorders have services and supports that meet their needs.

---

[1] The collective impact of rare diseases in Western Australia: an estimate using a population-based cohort
[2] 2021 Voice of Rare Disorders Survey

In order to give effect to te Tiriti, the following principles, recommended by the Waitangi Tribunal for primary health care[3], underpin Manatū Hauora work for the wider health and disability system:

- *Tino rangatiratanga:* The guarantee of tino rangatiratanga, which provides for Māori self-determination and mana motuhake in the design, delivery, and monitoring of health and disability services.
- *Equity:* The principle of equity, which requires the Crown to commit to achieving equitable health outcomes for Māori.
- *Active protection:* The principle of active protection, which requires the Crown to act, to the fullest extent practicable, to achieve equitable health outcomes for Māori. This includes ensuring that it, its agents, and its Treaty partner are well informed on the extent, and nature, of both Māori health outcomes and efforts to achieve Māori health equity.
- *Options:* The principle of options, which requires the Crown to provide for and properly resource kaupapa Māori health and disability services. Furthermore, the Crown is obliged to ensure that all health and disability services are provided in a culturally appropriate way that recognises and supports the expression of hauora Māori models of care.
- *Partnership:* The principle of partnership, which requires the Crown and Māori to work in partnership in the governance, design, delivery, and monitoring of health and disability services. Māori must be co-designers, with the Crown, of the primary health system for Māori.

Equity

Many people with rare disorders and undiagnosed health issues also have disabilities[4]. Equity issues for the rare disorders community are likely, on the whole, to align with those prevalent in disabled communities. Though there may be wide individual variation, in the rare disorders community health needs are very high. While health service utilisation may also be high compared with the general population, it may not be high enough in proportion to need.

Also similar to disabled communities, disproportionately high rates of unhappiness, depression and anxiety are reported[5]; and income, education and other socioeconomic indicators are lower and/or become lower over time, compounding access issues.

The rare disorders population have significantly poorer health outcomes than the whole population. This is further exaggerated among Māori and Pacific peoples for whom culturally informed services may be lacking, and for people including Māori and Pacific peoples who live in remote and high-deprivation communities[6].

A rare disorders strategy is expected to improve equity and health outcomes for disabled people (since a large number of people with rare disorders live with disability) – and similarly, the new Pae

[3] Waitangi Tribunal. 2019. Hauora: Report on Stage One of the Health Services and Outcomes Kaupapa Inquiry. Wellington. Waitangi Tribunal. pp. 163–164

[4] 2021 Voice of Rare Disorders Survey

[5] 2021 Voice of Rare Disorders Survey

[6] Manatū Hauora engagements for Pae Ora strategy development, including Ngā Wānanga Pae Ora and Te Mana Ola engagements

Ora Health of Disabled People Strategy will be expected to lead to improvements for many people with rare disorders who live with disability, whether as a result of a rare disorder or incidentally. A rare disorders strategy is expected to have a high focus on improving equity of outcomes within the rare disorders community, especially for Māori and Pacific people with rare disorders.

What is wanted from a rare disorders strategy?

In asking for a strategy to be developed, Ministers have expressed their intentions that its results would:

- improve the lives of people and whānau living with rare disorders
- allow the health sector as a whole to provide much better support for people with rare disorders
- make it easier for people, practitioners and organisations to get the information and support that would help
- lead to better, more timely services and more equitable support and outcomes for people and whānau with rare disorders.

What a strategy will provide

A rare disorders strategy will provide a clear direction of travel for the health sector on how improvements for people and whānau with rare disorders will be made progressively over the next decade. It will set out strategic aims (what is to be achieved) and describe the areas of focus or action towards those aims (how it will be achieved) using the resources and inputs that are available across the health system.

The strategy is being developed in partnership with health entities and Whaikaha, as well as with RDNZ and experts from clinical, science, cultural and lived experience viewpoints. The aim is bring all capability together and ensure that future possibilities inform our efforts to improve health system responsiveness and work towards pae ora for people and whanau with rare disorders.

What are rare disorders? (a proposed definition as a starting point)

> *A rare disorder is a medical condition with a specific pattern of clinical signs, symptoms and findings that affects fewer than or equal to 1 in 2000 persons.*
>
> *Rare disorders include, but are not limited to, rare genetic disorders, rare cancers, rare infectious disorders, rare poisonings, rare immune-related disorders, rare idiopathic disorders, and rare undetermined conditions.*

Background to a proposed definition

This definition is a summarised version of that proposed to WHO by Rare Diseases International (RDI), 'a global alliance of patient-driven organisations working together to advance equity for all Persons Living with a Rare Disease'. Globally, rare 'disease' and 'disorder' are often used interchangeably; 'disorder' has become the preferred terminology for RDNZ and in New Zealand.

Over the last 15 years, many countries have developed rare disorders strategies or actions. Most have struggled to define rare disorders and definitions vary. Definitions commonly include a prevalence threshold (such as disorders that are present in less than 1 person in 2000) or thresholds (such as for rare and ultra-rare disorders) and many countries have adopted a specific definition for orphan drug regulation purposes.

WHO and RDI agreed in 2019 to develop an operational definition of what a rare disease is as a global reference point. The definition proposed by RDI, still being refined, is:

> *People living with rare diseases face distinct and significant challenges that arise from the infrequency of their medical conditions, such as a long diagnostic journey, inadequate clinical management, and limited access to effective treatments. The burden of rare diseases on patients, their carers and families, healthcare systems, and society overall, merits greater visibility and recognition. A rare disease is a medical condition with a specific pattern of clinical signs, symptoms, and findings that affects fewer than or equal to 1 in 2000 persons living in any WHO-defined region of the world. Rare diseases include, but are not limited to, rare genetic diseases. They can also be rare cancers, rare infectious diseases, rare poisonings, rare immune-related diseases, rare idiopathic diseases, and rare undetermined conditions. While the frequency of most rare diseases can be described by prevalence, some rare diseases, such as rare cancers and rare infectious diseases, can be more precisely described by incidence.*

The proposed definition is a starting point that is not yet informed by Māori perceptions and aspirations. It will be crucial that Māori understandings of pae ora and rare disorders are a basis for the strategy and how rare disorders are described.

Intentions in creating a definition

Defining rare disorders is intended to promote aligned efforts for the rare disorders community and provide a focus for the strategy. Definition will:

- raise awareness and highlight the diverse range of disorders under the umbrella of rare and unidentified
- validate the experience of people and whānau who experience similar circumstances when accessing services or participating in community life
- promote equitable access and support in the health sector for those with rare disorders and their whānau
- concentrate effort in improving information and guidance to underpin quality healthcare and support
- align New Zealand efforts with international standards to facilitate better coordination and collaboration.

Defining rare disorders is not intended to create barriers or boundaries for people in accessing services. For example:

- health services will continue to be accessible according to needs and benefits, rather than having access limited for those with rare disorders in the boundaries of the definition

- changes in how rare a condition is will not result in supports being removed, such as when rare complications become more common (eg, rheumatic heart disease, or multisystem inflammatory disease which has increased in prevalence with COVID-19)
- there will always be a need to prioritise work to improve healthcare and to make available intensive or costly care; this work will be focused to promote equity across needs and benefits, and at times the focus will be narrower than on all rare disorders or all impacts
- overcoming particular barriers posed by rarity may by necessity focus on sub-groups or ultra-rare disorders; for example, Pharmac's special processes for rare disorders therapeutics funding applications, designed to overcome economic and process barriers for suppliers, may continue to focus on therapeutics for ultra-rare conditions while access to broader rare disorders treatments continues through Pharmac's standard assessment processes.

Key elements of a rare disorders strategy

Proposed aims of a rare disorders strategy

- Pae ora for people and whānau living with rare disorders
- Pae ora for Māori, Pacific peoples and disabled people living with rare disorders and for those living in rural and other diverse communities

Proposed principles to underpin a rare disorders strategy

- Stands on the shoulders of the Pae Ora strategies, the New Zealand Disability Strategy and the Child and Youth Wellbeing Strategy
- Upholds te Tiriti o Waitangi
- Gives voice to people and whānau with rare disorders
- Supports health practitioners and providers to provide quality care
- Is informed by and seeks out evidence
- Is collaborative and built on partnerships
- Supports pae ora for all while focusing on the health system challenges that come with rarity

In further refining the key elements, clear links through to the principles and goals of the Pae Ora strategies (soon to be published) will be drawn.

Doing the work to develop a strategy

Identifying key opportunities that could be built on

- the newly reformed health system with its focus on becoming more unified and consistent and accessible to all, including improving access to highly specialist services
- the system's heightened focus on equity of outcomes, especially for Māori, Pacific peoples and disabled people, including gathering evidence to inform equity improvement

- a growing te Tiriti focus and greater ability to draw on Māori expertise and governance
- advances in science, such as through new genetic and pathological testing capability
- advances in data and digital capability, such as through personal health records and virtual methods for liaison, consultation and care coordination and harnessing AI technologies
- growing knowledge of culturally informed care models to improve choice, accessibility and outcomes
- progress being made internationally, such as through clinical and research collaborations and virtual specialist networks
- potential for joining in cross-jurisdictional approaches, exploring potential for collaboration and harmonisation in issues like information governance or system interoperability
- themes and trends that will be highlighted in the long-term insights briefing on precision health (being completed in August).

Capturing voices of the rare disorders community

Rare Disorders New Zealand (RDNZ[7]) have set out seven priorities their community has identified to improve health and wellbeing for people living with a rare disorder:

- Diagnosis – early and accurate diagnosis of rare diseases
- Planned pathways for clinical care – coordinated and integrated pathways for cohesive healthcare
- Access to disability and social support – implement simple mechanisms to ensure appropriate access to disability and social supports
- Rare disorder medicines – equitable access to modern rare disorder medicines through a specific assessment pathway
- Research – coordinated and funded programme of research for rare disorders
- National rare disease registry – capture relevant data on rare disorders in New Zealand
- Workforce development – planned training on rare disorders for health professionals and support staff.

These priorities include both aims and specific action areas that will be considered in developing the strategy and in programmes or action plans that may follow it.

Inputs that are underway or planned

- an evidence review of international approaches to rare disorders
- capturing Māori perspectives on life for whānau with rare disorders, how whānau ora and pae ora can be promoted for whānau with rare disorders, what is most important and what the health system can do better
- ensuring Māori partnership in development and oversight of the strategy
- capturing Pacific peoples' perspectives and those of other diverse communities
- examining quantitative evidence on impacts of rare disorders, and how this may differ for Māori, such as through the Global Burden of Disease

[7] https://www.raredisorders.org.nz/

- further analysing input from rare disorders communities such as through detailed responses provided to RDNZ in the 2021 survey[8].

Identifying focus areas for the strategy

We need to develop a more detailed picture of key work underway that is aligned with areas of focus as the strategy develops. For example, much work is planned by new health entities to streamline and improve quality, accessibility, navigability and coordination of health care and provide digital backup and enablement.

The strategy will not do the work or identify specific actions or commitments in focus areas. However, to successfully point to the directions for maximum leverage and progress, a good overview is needed of what is already planned, what could be possible and how much could realistically be achieved across possible areas to emphasise.

Data & measurement

The likelihood of capturing all rare disorders, even with the best tools, coding and classification systems, will remain limited. However, we do have some advantages in New Zealand of a unique health identifier and progress towards universal and accessible health records as well as disability identifiers.

At this stage we have limited information on which to base evaluation or progress monitoring of a strategy. To know how well it is progressing is likely to require development of better data capture as well as creative 'work-arounds' to make the most of more limited or subjective information sets.

Data and measurement is important not only for assessing system responsiveness or performance. For many rare disorders, treatment and management is not well understood and being able to access clinical trials or other research may be important for individual care. Harmonisation with international datasets will be important.

Digital solutions and enabling

There are many possibilities to support people and health practitioners in best support and carer for rare disorders through digital means. Examples include:

- diagnostic and clinical standards, guidance and pathways, which can be accessed more effectively when integrated with health records or patient management systems
- health coaching and wearable tracking or monitoring, which can help people optimise daily wellness
- integrated care coordination apps, which can help facilitate communication and access across multiple services and supporters.

Digital enabling is important to realise best practice when knowledge may not be held widely or even in New Zealand at all. Components such as care standards or guidelines may be able to be sourced internationally and would require adaptation for New Zealand and integration with patient management or other systems to be most easily accessed.

[8] 2021 Voice of Rare Disorders Survey

Critical health services

Rare disorders require input from a very broad range of specialist and generalist health services. The ability of all to respond to the needs of people and whānau with rare disorders will be one focus of a strategy. However, some specialties and service areas will be critical in improving this overall responsiveness. These are those most likely to see people with rare disorders along their diagnostic and treatment journey, and those most likely to advise others on rare disorders and their management – service areas like paediatric and especially developmental paediatric and paediatric cancer services, genetic, metabolic, screening and pathology.

Improving system responsiveness will require examining how we can build capacity and capability in these service areas. Examining capacity and capability may include:

- reviewing workforce numbers and arrangements across many contributing workforces (medical, nursing, therapies, imaging, counselling, social, and other)
- reviewing connectedness of services with peers internationally and uptake of effective models of service provision, both for rare disorders and across all disorders, including virtual and distributed models
- looking into infrastructure that supports service provision, such as imaging, laboratory, genomic medicine (including access to genetic databases or biobanks), treatment infrastructure (medical, surgical and other), data infrastructure, analytics and management, and potentially use of AI.

Disability and whānau support and coordination

Some people and whanau living with rare disorders have high needs for a range of supports, sometimes required at very high intensity. Support with travel, respite, finance, mental health, education or care co-ordination may be needed because of disability or in order to access health services such as diagnostic tests or treatment. Supports can be particularly hard to access for people living in rural or remote communities and/or with particular language or cultural needs.

Knowledge building

Responsiveness to and inclusion for people with rare disorders requires knowledge building about the diversity of people's needs and capabilities and effective ways to support pae ora for all, including in all aspects of community life. Being able to easily access good information about and help with effective responses to people with rare disorders is important. Having the knowledge and ability to support people to live what is a good life for them is important for:

- health practitioners and service providers
- people & whanau themselves
- members of their wider communities.

Funding, assessment and prioritisation

Even the wealthiest countries, or those with the greatest research and innovation outputs, cannot afford to fund all treatments, supports, products or services that could benefit citizens, including those with rare disorders. All countries have to prioritise what will be funded or available for whom or in what circumstances.

In New Zealand, we have good assessment and prioritisation of individual products (such as medicines and medical devices) and certain services (such as surgical points systems), and some examples of integrated assessment of treatments, supports, products and services (such as for child cancer).

Building on the newly joined-up health system and the evidence-informed processes we already have provides opportunities to develop methodology to prioritise across different types of investments such as therapeutic products, services and workforces, infrastructure and systems.

Research and evaluation

Research into identifying and managing rare disorders individually is burgeoning internationally. Evaluation of programmes and policies for responding to rare disorders collectively is also increasing (for example, an evaluation is underway of the European Reference Networks (ERNs)[9] are virtual networks of health professional specialists in the diagnosis and treatment of rare/very rare illnesses in almost all fields of medicine including rare cancers. Their objective is to tackle complex or rare diseases and conditions that require highly specialised treatment and a concentration of knowledge and resources.)

To support a strategy, ways to access knowledge from and translate research for New Zealand will be needed. Equally important will be undertaking evaluation and research locally, especially for responsiveness to Māori and Pacific populations and for knowing effectiveness of practices, service models or programmes in our context.

Research collaboration across countries is important to grow and access knowledge. It can also provide opportunities for people and whānau with rare disorders to contribute to others by helping to increase knowledge of their disorders, such as through participation in larger international studies including clinical trials.

[9] European Reference Networks

ADAM'S STORY

LIVING WITH A RARE DISORDER

By Michelle, Adam's mum

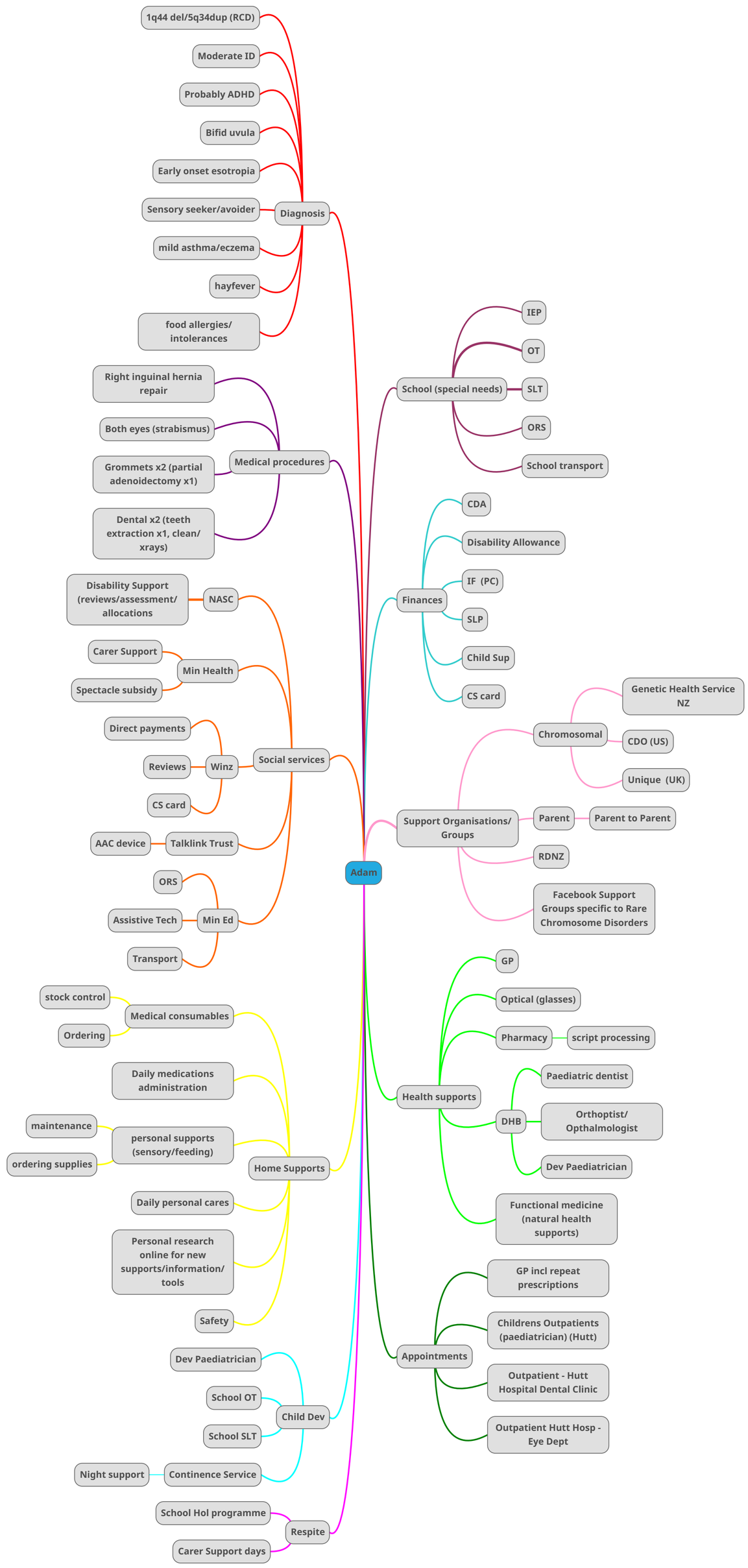
This is a snapshot in time of the current care and supports in place for Adam. Adam is 15 and has a rare chromosome disorder known as 1q44 deletion, along with 5q34q35 duplication. Adam's specific rare chromosome disorder is ultra-rare (<1 in a million) and possibly unique.

It is not a fixed/static snapshot, as things can change often. With the onset of puberty changes have come about, including a recent diagnosis of probable ADHD (high hyperactivity/impulsivity). We are currently trialling medicine to see if it will help to allow Adam to be able to focus on things better and help with some of his behaviours.

Adam has high needs, so this does require constant supervision, as well as assistance/ support for daily tasks such as personal cares. His moderate intellectual disability impacts on his daily life and can present a lot of challenges.

My biggest support in terms of his rare chromosome disorder is through Facebook. I belong to a worldwide group specifically for his deletion where we share information, ask questions and support each other. From there I can gain relevant information that I can then bring up when Adam has a paediatrician appointment for example.

Juggling the coordination of care for Adam is time consuming. The attention needed for Adam does have a big impact on your life – both physically and mentally. You become someone who wears many hats to help Adam navigate the world.



VISIT WWW.RAREDISORDERS.ORG.NZ

Advice received by Rare Disorders NZ regarding incorporating coding for rare disorders in the health system

Advice provided collegially by Garath Baynam (Rare Care Centre) and Dylan Gration (Manager of Data and Digital Linkages, Western Australian Register of Developmental Anomalies), Western Australian Department of Health, in December 2022.

Basically the solution is to get **Orphanet Coding (ORPHACODES)** into the health system. This can be done in a way that is compatible with other key coding systems (e.g. SNOMED, ICD-10/11)

ICD-11 alone is not the solution, it is better than ICD-10 for RD no doubt, but still has significant limitations.

Our WA Health Minister requested us to introduce Orphanet Coding into our Health System, using WARDA as the place to start. Birth defects are a large group of mainly rare diseases and we also now know that about 25% of Cerebral Palsy has an underlying rare genetic disease cause

I mention the NZ Cerebral Palsy Registry as a registry that has done very well in setting up a data ascertainment system. This has been in partnership with the Australian Cerebral Palsy Registry (ACPR) who are probably leaders in terms of how well organised they have been in setting up a national registry despite all the jurisdictional differences between states.

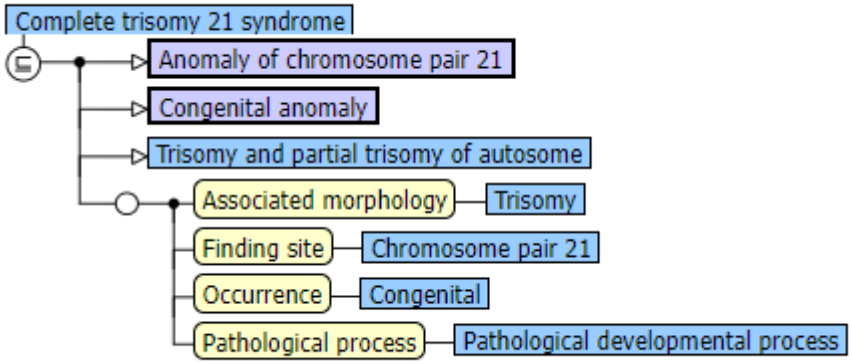
Also the NZ Birth Defects Registry might be a good start to get in contact with to begin looking at how they are capturing congenital anomalies which will be (mostly) rare.
https://www.ehinz.ac.nz/projects/new-zealand-congenital-anomalies-registry/

In terms of coding - NZ, like Australia, has a national terminology service that will provide access to certain types of codes for your hospital systems.
https://www.tewhatuora.govt.nz/our-health-system/digital-health/terminology-service/
One of these is SNOMED CT which is actually a very useful resource. This is because ~6,500 Orphanet rare disease codes, aka Orphacodes, have been mapped to SNOMED CT codes.

The key question will be, WHERE and HOW are they using SNOMED CT codes in NZ? For example, in Western Australia they are only using SNOMED CT codes for coding inpatient admissions. However, this will most likely change in future once we have an Electronic Medical Record since that is the Australian Digital Health Authority standard plugin terminology used in such systems. Coding systems like ICD-10 and ICD-11 will of course still have a role to play for coding for activity reimbursement purposes, but these do not provide the same level of information as codes like SNOMED CT – they are not as 'granular' and do not have underlying logic like SNOMED CT.

I can see that a nation wide EMR is on the cards for NZ and that SNOMED is the selected terminology for the system. This is very good news! https://www.snomed.org/snomed-ct/case-studies/new-zealand-electronic-health-record

You can see an example of this logic below, whereby this SNOMED CT code has logic explaining it is a congenital condition. This is very useful for coding rare diseases since most congenital anomalies are rare, and as a result they are the largest class of rare diseases.



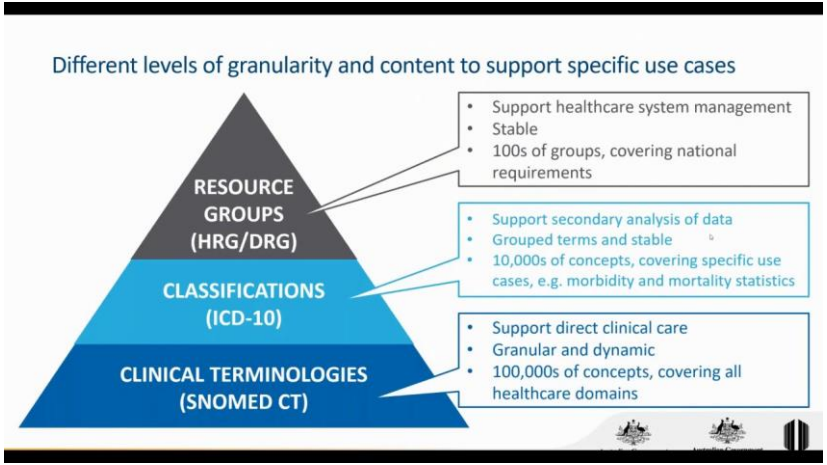
Unfortunately ICD-11 has not yet incorporated as nearly as many RD codes (yet), with ~1500 Orphacodes having an ICD-11 code last time I checked.

I've attached a very brief slide set of I did that might help give some perspective on our AU challenges.

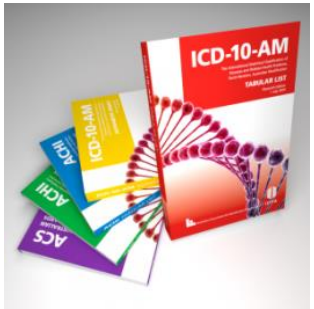
SNOMED vs ICD10AM

Australian Government
Australian Digital Health Agency

SNOMED International



IHPA

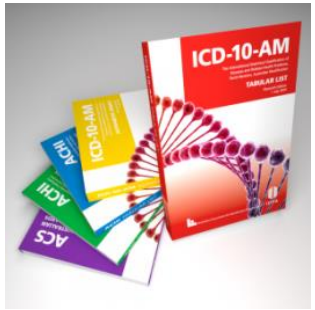


SNOMED CT-AU

ICD10AM

ICD10AM: activity & finance focused

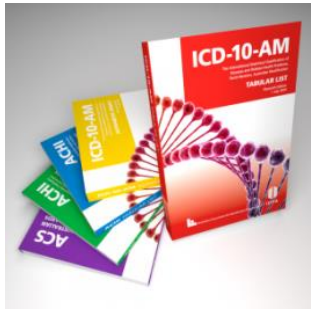
IHPA



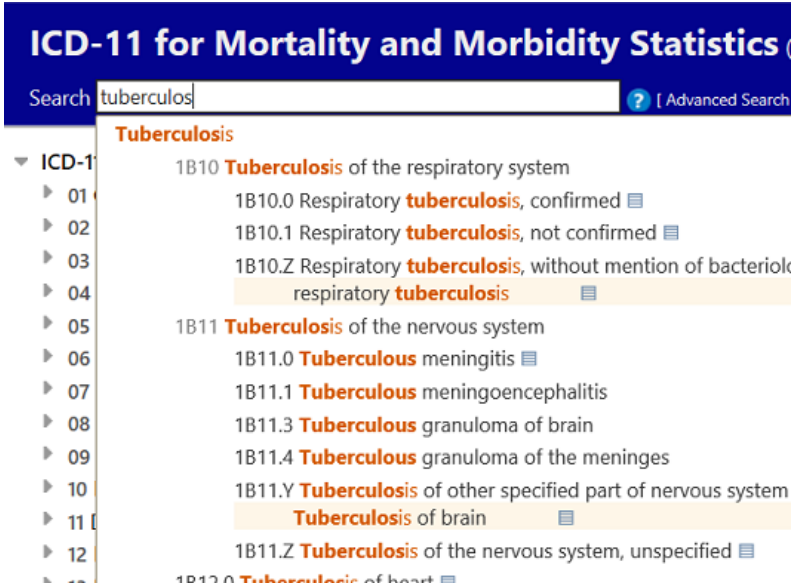
ICD10AM

ICD10AM to ICD11

IHPA



Transition will create a breakpoint in ICD10AM coded data collections. Unable to link back for longitudinal data for exact concept matching



ICD10AM → **Narrow one-to-many** → **ICD11(AM)**

SNOMED CT-AU: more open & interoperable

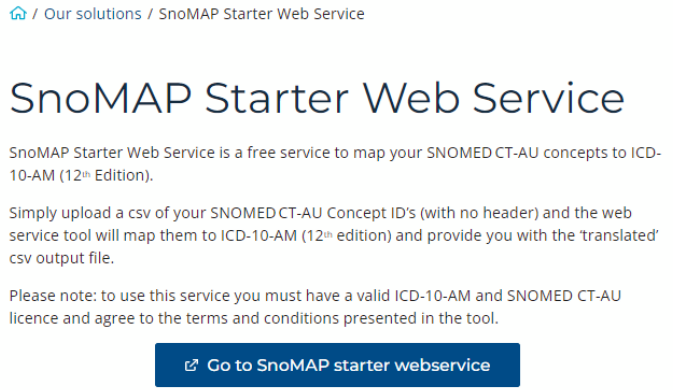
Australian Government
Australian Digital Health Agency

CSIRO

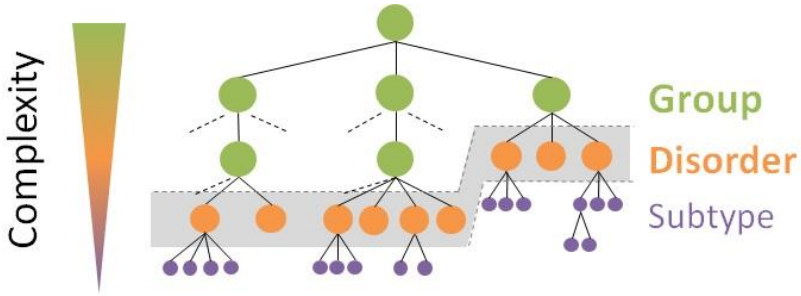
ONTOSERVER

SNOMED International

SNOMED CT-AU



SNOMED CT to Orphanet map



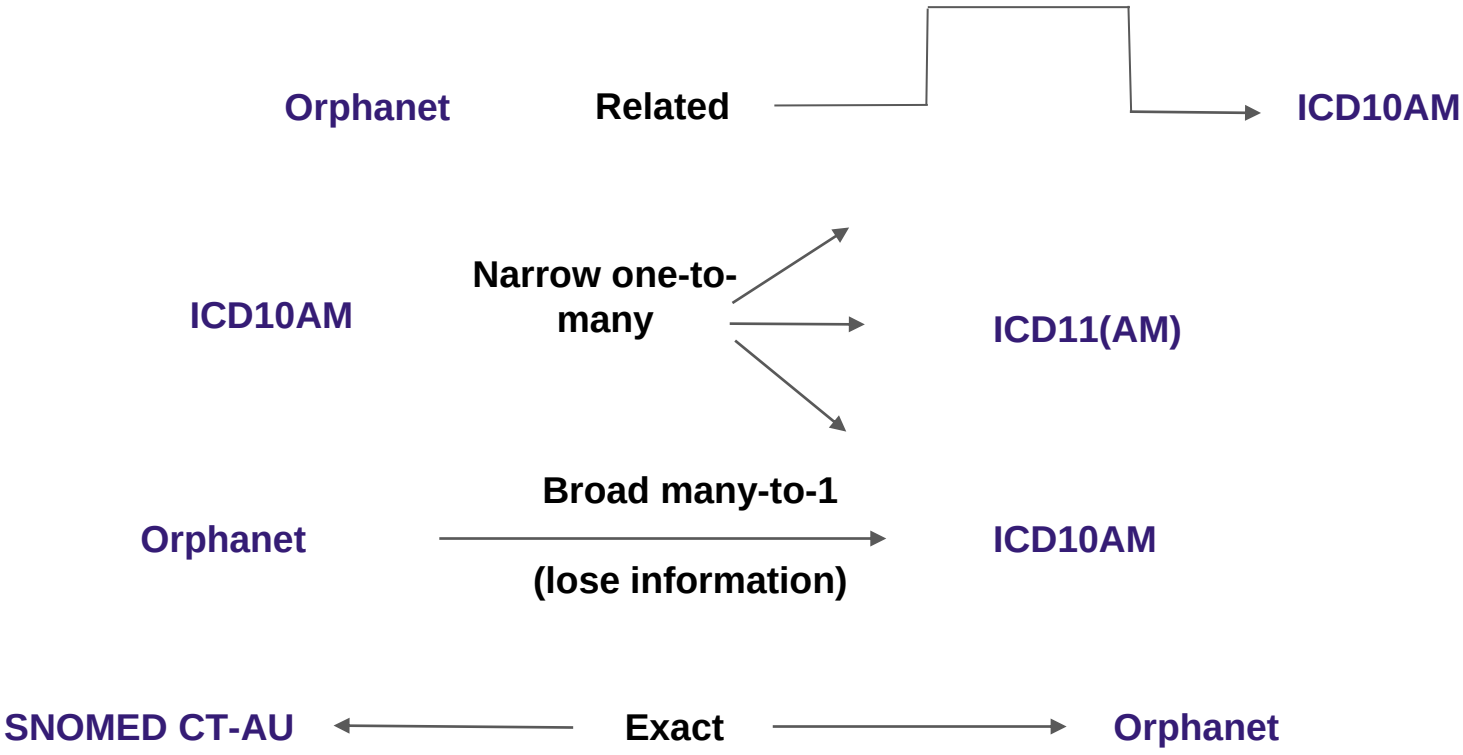
SNOMED International

Exact

~6,500 RD codes

orphanet

Types of mappings for clinical codes



The need for data on rare disorders in NZ

At a glance

There are estimated to be around 300,000 New Zealanders living with a rare disorder.

This estimate is based on international data, because in the New Zealand health system data on rare disorders is not being captured.

Key stats

A 2023 insights report on the prevalence of rare disorders in New Zealand found

The quantity of data available on rare disorders in New Zealand is strikingly low.

The classification system for diseases that New Zealand uses does not include most rare disorders.

Rare Disorders Insights Report: Pathways Towards Better Health Outcomes
https://www.raredisorders.org.nz/about-rare-disorders/rare-disorders-in-new-zealand/berl-insights/

RARE DISORDERS NZ

PO Box 14-313, Kilbirnie,
Wellington 6241, NZ

www.raredisorders.org.nz

CHALLENGES

New Zealand has no official process for collecting data on rare disorders. This means this sizeable population group and their health needs are invisible and not being factored into decisions by policy-makers, nor receiving adequate Government funding and resource allocation, despite often being high-cost and high-needs patients.

SOLUTIONS

An integrated, centralised and accessible data patient registry which uses the Orphanet classification system would improve data collection and accuracy, and benefit international collaboration on research.

Develop a rare disorder patient registry

Establish an official definition of what is considered a rare disorder in NZ

Adopt the Orphanet classification system for rare disorders

BENEFITS

1

Benefits One

Better communication between care providers, improving diagnosis and treatment.

2

Benefits Two

Government funding and resources more adequately allocated to meet the needs of rare disorder patients.

3

Benefits Three

Valuable information for researchers and policymakers to inform inclusive policy and improve the lives of people living with a rare disorder.

OVERVIEW

This report summarises the findings from both the *State of Caring* and *Respite and Time Out* surveys undertaken by Carers NZ and the Carers Alliance in 2021. Over 1600 carers responded to the surveys.

For the purposes of this report and the surveys it summarises, carers are defined as being "individuals, family, whānau and āiga providing care for someone close to them who needs additional assistance with their everyday living because of a disability, health condition, illness or injury".[1]

Carers across New Zealand responded. They were of all ages, ethnicities and walks of life. These are their themes, their findings and their recommendations.

Key themes

The key themes summarised below are the result of analysing the 1600+ survey responses received. The richness and depth of the responses is significant. The following themes came through strongly in the responses. Additional evidence to support the themes is provided in the body of this report.

Caring significantly impacts wellbeing

Carers have rates of depression and/or anxiety that are much higher than in the general population, and many are not seeking support. Much of this anxiety and depression is due to the financial stresses that can come with caring.

Carers have very high levels of loneliness, very low life satisfaction, and low levels of physical health compared to the general population. Carers aged under 35 reported the lowest levels of physical health and the highest levels of depression and anxiety.

"DURING THE COVID LOCKDOWNS IT FELT LIKE THE GOVERNMENT DID NOT THINK ABOUT CARERS' CONCERNS AND SITUATION AT ALL AND WE WERE NEGLECTED. CARERS WHO ARE MORE VULNERABLE WERE NOT GIVEN EXTRA SUPPORTS, EVEN WHEN THEY ASKED."

[1] https://www.health.govt.nz/our-work/carers-in-new-zealand#who

Formal support for carers is lacking

The survey results highlight carers' common struggle to know what support is available and how to access it, with particular challenges around needs assessments and entitlements. Many carers indicated they are missing out or have missed out on supports that they should know about, which then negatively affects their wellbeing, relationships or finances. This can contribute to carers not feeling valued by the government and results in increased financial burden as they are paying out of pocket to meet care-related costs.

The internet is a key avenue for carers to access information and advice. There is varied use of other technologies such as remote monitoring alerts, medication management and telehealth.

Many carers are not planning for their own future because they are focused on caring or are unable to plan more generally (for retirement, for example). Only 7% of carer respondents have plans for their own future care needs.

Carers are not being included in existing processes for needs assessments and service coordination, and 86% of *State of Caring* respondents think that family, whānau and āiga carers should have their own targeted needs assessment. Any needs assessment or similar targeted support allocation process for carers needs to deliver supports that are available, accessible and well promoted to this population.

The challenges of juggling work, finances and caring are huge

Carers have lower rates of employment than the general population, with only 21% in full-time work and 23% in part-time work.

More telling is that 30% of carers indicated they are unable to undertake any paid work due to their caring responsibilities. Fifty percent of *State of Caring* respondents had to give up paid work to care, reduced their hours of paid work, or retained their hours of paid employment but with negative job impacts due to caring. Flexibility at work supports carers to juggle both work and caring.

Many carers (59%) said they cannot pay their bills without struggling financially, and 15% have been in debt because of caring, which makes New Zealand carers worse off financially than UK carers but under similar financial stress as Australian carers. Carers make ends meet in a variety of ways; it is a concern that 30% are cutting back on essential items such as food and heating for their whānau to best manage money and caregiving realities.

This financial stress is having a long-term impact, with 20% of carers not being able to save at all for retirement, 34% having their retirement plans negatively affected, and only 14% unaffected by financial strains.

Limited access to respite and time out options that work

Access to opportunities for breaks from caring (often called 'respite') has varied during the pandemic, with some carers being able to have breaks weekly or whenever they need them; 32% having breaks only a few times a year and 13% only being able to take breaks in emergencies. Access to quality options for respite was already an issue for many carers pre-pandemic, and COVID-19 has made this worse by reducing access to respite support and services. Carers' wellbeing is being impacted by inadequate respite opportunities, with 48% struggling, really struggling, or needing emergency respite. Almost 90% of carers indicated they have had less or much less respite since the start of the pandemic in March 2020.

Suggestions for improvement included providing more flexibility with how funding can be used, more guidance on how funding works, and a competent, reliable workforce that can alleviate carers' stress. Carers also indicated that increasing the availability of respite options and funding is important – especially over time as the 'long-tail' of pressure builds, leading to carer breakdown which in turn will cause significant financial burden for the government.

Recommendations

The survey feedback highlights the issues and challenges facing carers across New Zealand. In reviewing the feedback, there are potentially many recommendations that could be made – however, to provide focus and attention, the following 12 recommendations are made:

1. Fully implement the *Mahi Aroha Carers Strategy Action Plan's* outcomes with vigour and use the data in this report and future evidence to identify useful actions for the next Action Plan.
2. Increase existing supports and provide new ones for carers – including making respite funding more flexible across the diverse population of carers and investing more into respite to improve carer wellbeing as a single vital way to safeguard and improve the wellbeing of this large population.
3. Provide more effective navigation support (particularly around respite) and improve the promotion of information available to carers about financial supports (including being paid to provide care), respite and wider

government assistance.

4. Improve and simplify financial supports to reduce barriers to financial assistance, such as changing spouses' inability to access the Supported Living Payment or family carers' inability to be fairly paid for the significant efforts they make in areas such as Individualised Funding (often at the expense of other paid work opportunities).
5. Formally recognise the role of carers and the value they provide to their whānau and the system by directing government departments to ensure plans and strategies specifically include focus and actions that relate to carers.
6. Prioritise the implementation of an approach to the appropriate consideration of carer needs in their own right and a process to ensure supports meet identified needs., with a specific focus on advance care and emergency planning.
7. Find a place in Government for carers, who too easily 'fall through the cracks' of existing structures, frameworks, and Ministries; this could take the form of a Minister/Ministry for Family Whānau and Aiga Carers and/or a Commissioner who has responsibilities for this large population of New Zealanders. We note that a similar approach is being mooted in Australia for the same reasons.
8. Ensure the evolving structures of government (Te Whatu Ora – Health NZ, Te Aka Whai Ora - Māori Health Authority, Whaikaha - Ministry of Disabled People, ACC etc) are meaningfully 'carer friendly' in important areas such as respite, information, financial support, wellbeing and support for carers who are Māori, Pacific, young, etc.
9. Identify measurable ways to support and improve carer wellbeing in areas such as employment, retirement planning, loneliness and social isolation, finances, and mental health; ensure programmes for wellbeing are adequately resourced and promoted.
10. Identify and implement supports for carers aged under 35 as a direct response to concerns highlighted in this report of the impact caring has on younger carers.
11. Support specific initiatives and programmes that assist working age carers, particularly women and young carers, whose earnings, life success, retirement savings, and financial wellbeing impact their ability to work, earn, save, and thrive.
12. Implement specific supports for carers impacted by the COVID-19 pandemic. These impacts were visible in the comments and data gathered for this report in 2021 and will have deepened since in areas such as respite, wellbeing, employment, finances, and access to support and services.