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

- [redacted] 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.

- 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.