Email from Chris Higgins (CE RDNZ) to [Ministry of Health Principal Policy Advisor working on the Rare Disorders Strategy] 13 November 2023

Kia ora [Ministry of Health Principal Policy Advisor working on the Rare Disorders Strategy]

Following discussion at last Thursday's RDS meeting RDNZ recommends that the following be included as a minimum set of four actionable items in the next draft of the RDS

1. Creation of a rare and undiagnosed disorders centre of expertise, modelled on Rare Disorders New Zealand's Rare Support Centre Aotearoa proposal presented in Appendix X (attached)

2. Recognition of RDNZ as a key enabler for the Strategy's implementation

3. Incorporation of coding of rare disorders in the roll-out of the nationwide Electronic Medical Record (EMR) and other administrative data sets

4. Establishment of a single barrier-free pathway to enable people with rare disorders to access the medicines they need. Access will be consistent with or exceed best practice international norms.

We've elaborated on all of the above points in our intended Briefing to the Incoming Ministers of Health, also as attached.

By way of further elaboration on point 3 we suggest incorporating Orphacoding into EMR and administrative data sets. See here for rationale: https://www.frontiersin.org/articles/10.3389/fped.2023.1283880/full. The New Zealand birth defects (congenital anomaly register) would be one key register to introduce Orphacoding, as has been done in the UK and in WA. Birth defects are a large class of mainly rare diseases, and (aka disorders of embryogenesis) the biggest class of rare diseases (especially in children). We also recommend developing public hospital

outpatient appointments data as a rich but largely untapped source of information for all diseases, including rare disorders.

The four points above have been developed in close collaboration with a list of over 20 RDNZ identified specialists and experts and some 150 rare disorders support group leads. We believe that they were further reinforced during the clinical consultation sessions led by the Ministry's Hamish Gray.

As such they have wide sector support as having the best impact for patients and whanau/families. As part of our ongoing public advocacy campaign (boosted with TVNZ news items yesterday on the 6.00 pm news and this morning on Breakfast), we will during the course of the week make them widely available through our various communication and media channels. This may result in our recommending that they be further strengthened.

We're aware that consultation is occurring on the Strategy with both Te Whatu Ora and Pharmac. While it is important that these agencies have an opportunity to express their perspectives we think it important that the RDS in its final version is an aspirational yet actionable document which is not compromised by provider capture.

We are encouraged to know that there will be other important elements to the RDS which will be or already have been identified by the Manatū Hauora Strategy team, and we look forward to contributing to their development as we see iterations of drafts over the next few weeks.

Ngā mihi

Chris

Chris Higgins

Chief Executive

Rare Disorders NZ

RARE SUPPORT CENTRE AOTEAROA

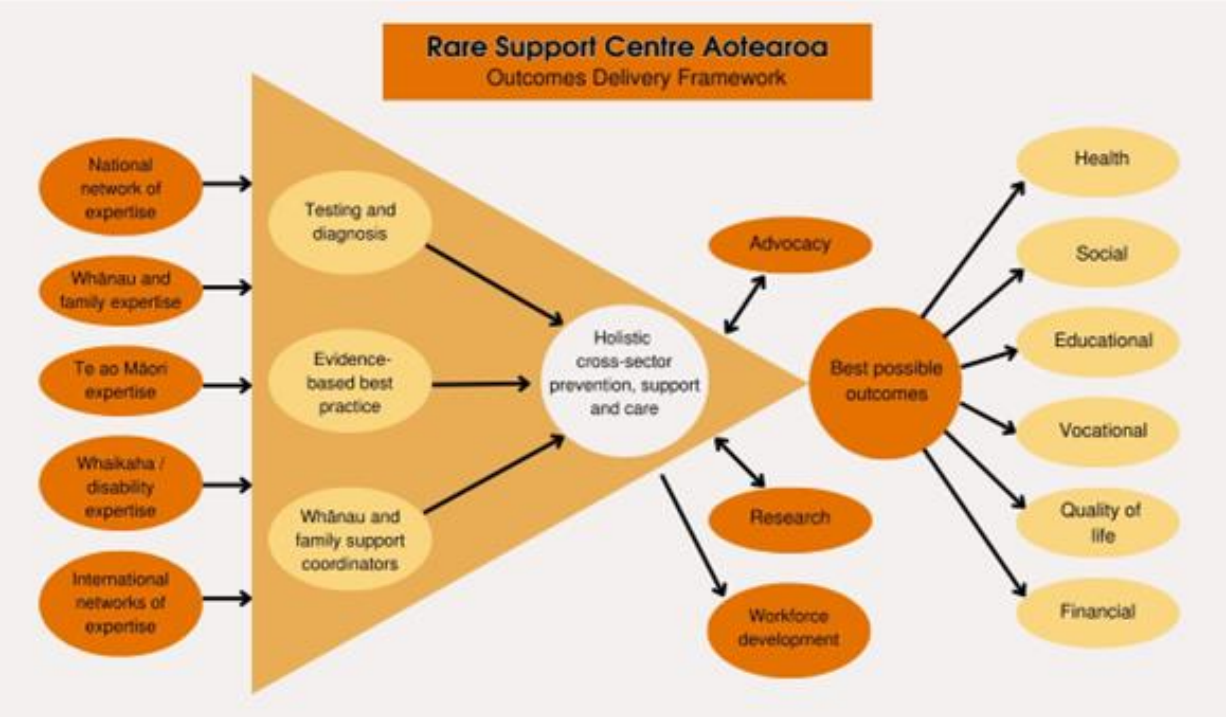
(Rare and Undiagnosed Disorders Centre of Expertise)

"Delivering equitable outcomes and best possible health and wellbeing for people and whānau living with rare and undiagnosed conditions in New Zealand/Aotearoa through world class and world leading health, disability, education, social and other support."

Inequity of outcomes for people with rare disorders is represented by the health and other outcome differences experienced by people who receive a full suite of planned and coordinated evidence-based services and therapies compared with those who don't. These outcome inequities are exacerbated for people with rare disorders who are Māori and/or disabled.

The figure below sets out a framework for achieving improved outcomes for all New Zealanders who have a rare or undiagnosed disorder.

Figure 1: Rare Support Centre Aotearoa Outcomes Delivery Framework



"The absence of a holistic approach made the whole family's devastation unnecessarily more profound and led to 10 years of feeling let down" (Jaime Christmas, Chief Executive, New Zealand Amyloidosis Patients Association)

The Rare Support Centre Aotearoa (RSCA) will comprise networked holistic[1] cross-sector multidisciplinary prevention, support and care services which seek to eliminate these inequities by addressing the service and support gaps in New Zealand's health and other systems. Health will be nested in a larger support centre which addresses a wide set of important life outcomes.

[1] Miller, N. Where is the wraparound care? Interview with Jaime Christmas, Chief Executive, New Zealand Amyloidosis Patients Association. RARE Revolution Magazine, p34. October 2023. Rare Revolution (pagesuite-professional.co.uk)

Drawing on and extending the WHO definition of health[2] the RSCA takes its inspiration from comparable initiatives globally[3] [4] [5], including the Government of Western Australia's Clinical Centre of Expertise for Rare and Undiagnosed Diseases[6] [7] [8], Undiagnosed Diseases Programs[9] and the Undiagnosed Diseaseas Network International[10].

Coordinated from [city] the Centre will be a member of the Global Network for RD[11] and comprise a diverse multidisciplinary virtual team of internationally networked experts distributed across various Aotearoa/New Zealand locations. Typically their contribution to the work of the Centre will be complementary to and an extension of roles they hold with locality based health and other service providers. Clinical experts will be affiliated with Te Whatu Ora's properly resourced Rare Disorders National Clinical Network[12], rather than be fragmented across multiple clinical networks as is the case currently.

Prior to the enactment of the Pae Ora (Healthy Futures) legislation people with rare disorders often reported that they faced obstacles to receiving support because of the "postcode lottery" effects of living in a location where the services they required were unavailable. People are now reporting that services that have since become (together with those that already were) available nationally rather than just locally or regionally are not able to meet the resulting increases in demand, resulting in long waiting times to be seen.

It will be necessary for the RSCA, the extended roles of its networked experts, and allied and complementary services to be appropriately resourced. They will also need to be technologically connected with each other and with patients. Sufficient resources supported by appropriate technology will ensure that people are assessed and supported in a timely manner, especially Māori, people with disabilities, and those who live in rural locations.

Testing and diagnostic service[13]

For those without a definitive diagnosis the RSCA's testing and diagnostic service will be the portal to the RSCA itself and available as a referral centre to any health or other professional with clients or patients presenting with unusual co-presenting symptoms, with or without a suspected diagnosis.

A definitive diagnosis is often an essential component of understanding how to best provide services and therapies to a person with a rare disorder. A definitive diagnosis can be obtained

[2] Health is a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity. https://www.who.int/about/accountability/governance/constitution
[3] Centres of Excellence for Rare Diseases. Rare Diseases UK. 2013. centres-of-excellence.pdf (raredisease.org.uk)
[4] European Reference Networks. https://health.ec.europa.eu/european-reference-networks/overview_en#work
[5] National Expertise Centres for Rare Disorders. Leiden University Medical Centre, Netherlands. https://www.lumc.nl/en/patient-care/polyclinics-nursing-wards-and-expertise-centers/expertisecentra/expertise-center-for-rare-disorders-ecza/
[6] https://pch.health.wa.gov.au/Our-services/Rare-Care-Centre
[7] Rare Care Centre. First Year Impact Report. Feb 2022-2023. Rare-Care-Centre-Impact-Report-Y22-23.pdf (health.wa.gov.au)
[8] Rare Care Clinical Centre of Expertise for Rare and Undiagnosed Diseases Strategic Framework 2022-2023.
[9] Baynam G et al. Initiating an undiagnosed diseases program in the Western Australian public health system. Orphanet Journal of Rare Diseases. 2017.https://ojrd.biomedcentral.com/articles/10.1186/s13023-017-0619-z
[10] Taruscio D et al. The Undiagnosed Diseases Network International: Five years and more! Molecular Genetics and Metabolism Volume 129, Issue 4 (https://pubmed.ncbi.nlm.nih.gov/32033911/)
[11] Baynam G. Rare Care Centre: global needs, local leadership. Sept 2022. Medical Forum. https://mforum.com.au/rare-care-centre-global-needs-local-leadership/
[12] National Clinical Networks – Te Whatu Ora- Health New Zealand *"Joined-up clinical leadership comprised of diverse expert voices to drive system shifts through development of national standards and models of care".*
[13] The importance and increasing ability to achieve a diagnosis is discussed in Baynam G et al. Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. M. Posada de la Paz et al. (eds.), Rare Diseases Epidemiology: Update and Overview, Advances in Experimental Medicine and Biology 1031, chapter 4. 2017. https://doi.org/10.1007/978-3-319-67144-4_4

via non-genetic testing, genetic testing, expert observation or sometimes requires a combination of the three.

The RSCA will offer a culturally appropriate and safe diagnostic counselling and coordination service which will fully inform and directly assist clients and patients to access tests which are consistent with their values, actual and potential life choices, and symptoms, having regard to the results of any previous inconclusive or negative tests. This service will be available until a definitive diagnosis is achieved or until the client or patient withdraws consent to continue, and will be supported by use of AI[14] and other emerging technologies.

Unless there are compelling clinical reasons to do otherwise, and in any continuing absence of a definitive diagnosis, clients and patients who have been accepted into the service will be presumed to have a diagnosis of a rare disorder. Suspecting or believing that a client or patient is imagining or making up their symptoms does not constitute a "compelling clinical reason".

Care and support

The RSCA's cross-sector and multidisciplinary care and support service will:

- Accept rare disorder referrals from the RSCA's testing and diagnostic service, and other credentialed diagnostic and screening services (such as the national newborn screening service)
- Develop and maintain evidence-based world class, world leading and globally connected standards of holistic best practice cross-sector care and support for **specific and identifiable** rare disorders
- Develop and maintain evidence-based world class, and world leading globally connected standards of holistic best practice cross-sector care and support for **undiagnosed** rare disorders[15] [16]
- Provide expertise and guidance for general practitioners, specialists, other clinicians and other professionals in how to support their patients and clients in accordance with best practice standards
- Develop and implement measurement of appropriate outcome measures for patients and clients focusing on patient experience measures and disease-appropriate outcome measures
- Facilitate belonging to relevant national and international rare disease support groups
- Support workforce development through:
    - provision of curriculum development and rare disorders training to clinical and other training schools and entities, continuing professional development providers, other professional development agencies and service providers
    - identification of workforce gaps including in the supply of skilled staff to meet demand and requirements for new subject matter experts to harness and implement new knowledge and technologies, and facilitate closing these gaps.

[14] Eg Cliniface. https://cliniface.org/
[15] Baynam G et al. Initiating an undiagnosed diseases program in the Western Australian public health system. Orphanet Journal of Rare Diseases. 2017. https://ojrd.biomedcentral.com/articles/10.1186/s13023-017-0619-z
[16] Taruscio D et al. The Undiagnosed Diseases Network International: Five years and more! Molecular Genetics and Metabolism Volume 129, Issue 4, April 2020, Pages 243-254 (https://pubmed.ncbi.nlm.nih.gov/32033911/)

Service coordination

The RSCA will engage a team of professionally trained service coordinators who will acknowledge whanau and family as their own best experts, and support them as personal advocates to navigate the health and other systems in a timely way, ensuring that all appropriate services are accessed, and appointments are made and kept, in accordance with best practice individual care and support plans

Research and clinical trials

The RSCA will identify gaps in knowledge and expertise and engage with health and wellbeing researchers, research funders, service professionals and international rare disorders research networks[17] to have those gaps filled, and the resulting knowledge translated into best practice standards of support. For 95% of rare disorders clinical trials are the only hope of a disorder specific prognosis altering drug therapy[18]

Advocacy and awareness

The RSCA will identify gaps in service provision and support, both internally and externally in the health and other sectors, and will partner with rare disorders support groups through the Rare Disorders New Zealand peak body to advocate both for required system changes and professional and public awareness of the RSCA's services.

[17] Eg International Rare Diseases Research Consortium (IRDiRC) (Who we are – IRDiRC) and The European Joint Programme on Rare Diseases (EJP RD) (EJP RD – European Joint Programme on Rare Diseases)
[18] Baynam G. Personal communication. 7 November 2023.

Briefing
Rare Disorders in Aotearoa 2023

Overview

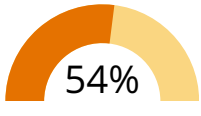
An estimated 300,000 New Zealanders live with at least one of over 7,000 known rare disorders.

A disorder is considered rare if it affects fewer than or equal to 1 in 2,000 persons in New Zealand. Often complex, debilitating and life threatening, these conditions require a broad range of services.

Regardless of the specific disorder, we know that in New Zealand those affected largely share the same challenges and systemic barriers to enjoy the best possible health and quality of life outcomes - lack of timely diagnosis, limited treatment options, lack of support for coordinated care, isolation, significant carer impact and for many, being lost in the system. The common experience of being bounced around from specialist to specialist takes a significant toll on patients' mental and physical health, and puts unnecessary pressure and additional costs onto the health system.

Lack of timely diagnosis
For one in five, it took over 10 years to get a diagnosis.

Poor coordination of care
Over 50% felt that communication between different service providers was poor.



Financial strain
54% felt the costs associated with managing their rare disorder were hard to manage.

Source: Impact of Living with a Rare Disorder in Aotearoa New Zealand, February 2022

Improving the health and wellbeing of people with rare disorders is possible, beginning with a purposeful strategy, and a commitment to implement key changes in the health system to improve health system responsiveness for people living with a rare disorder.

New Zealand's first Rare Disorders Strategy

In 2022, the Minister of Health instructed Manatū Hauora to develop New Zealand's first Rare Disorders Strategy. The intention of the strategy is that it will lead to better, more timely services and more equitable support and outcomes for people and whānau with rare disorders. A draft strategy for the Minister of Health is expected to be delivered before the end of 2023.

Rare Disorders New Zealand, as the only umbrella organisation supporting all New Zealanders who live with a rare disorder, has been involved both as a member of the RDS reference group and as a co-designing partner.

Delivering on the Strategy

For the Rare Disorders Strategy to deliver on its intentions to ensure the health system responds and relates equitably to rare disorder patients and improves their quality of life, it will need to include a plan of action. Rare Disorders NZ, in consultation with domestic and international experts, has identified four essential implementable actions to prioritise beyond the completion of the strategy:

Action # 1

Establishment of a Rare and Undiagnosed Disorders Centre of Expertise

Action # 2

Establishment of a single barrier-free pathway to rare disorder medicines

Action # 3

Incorporation of coding of rare disorders in the roll-out of the nationwide Electronic Medical Record

Action # 4

Recognition of RDNZ as a key enabler for the Strategy's implementation

#1 Rare and Undiagnosed Disorders Centre of Expertise

The Centre will identify gaps in the delivery of service and support in New Zealand's health and other systems for people living with rare disorders, similar to other initiatives globally. The centre will comprise a multidisciplinary team of internationally-networked experts within New Zealand, who will be available to provide expertise and guidance for clinicians and professionals in how to support rare patients and clients in accordance with best practice standards. This work will be an extension of roles they hold with locality-based health and other service providers.

#2 Single barrier-free pathway to rare disorder medicines

The current one-size-fits-all model under Pharmac's pharmaceutical schedule does not work for low volume, high-cost medicines for rare disorders, and there are too many inconsistencies under the only other avenue – the exceptional circumstances framework. A separate assessment pathway for rare medicines with criteria separate to common conditions (as is implemented in Europe, Australia and Japan) will ensure equitable access to medicines for those with rare disorders.

#3 Incorporating coding of rare disorders in the classification system of diseases

The classification system for diseases that New Zealand uses does not include most rare disorders. This lack of data is problematic for funding and resource allocation, as these patients tend to be high-need, high-cost patients. With the implementation of the nationwide Electronic Medical Record (EMR) using SNOMED terminology, there is an opportunity to incorporate coding of rare disorders using Orphanet Coding (ORPHACODES) in this system, as 6,500 Orphacodes, have been mapped to SNOMED CT codes. Orphanet is the most comprehensive database of rare disorders.

#4 Rare Disorders NZ is a key enabler for the Strategy's implementation

As the only national organisation supporting all New Zealanders living with a rare disorder and their carers, Rare Disorders NZ has played an integral role in supporting the development of the Rare Disorders Strategy, by advising, providing essential information and establishing connections between the Ministry and stakeholders, including patients and whānau. The organisation is well-placed to continue to support the implementation stage of the strategy, and should be acknowledged and compensated accordingly.