RARE SUPPORT CENTRE AOTEAROA

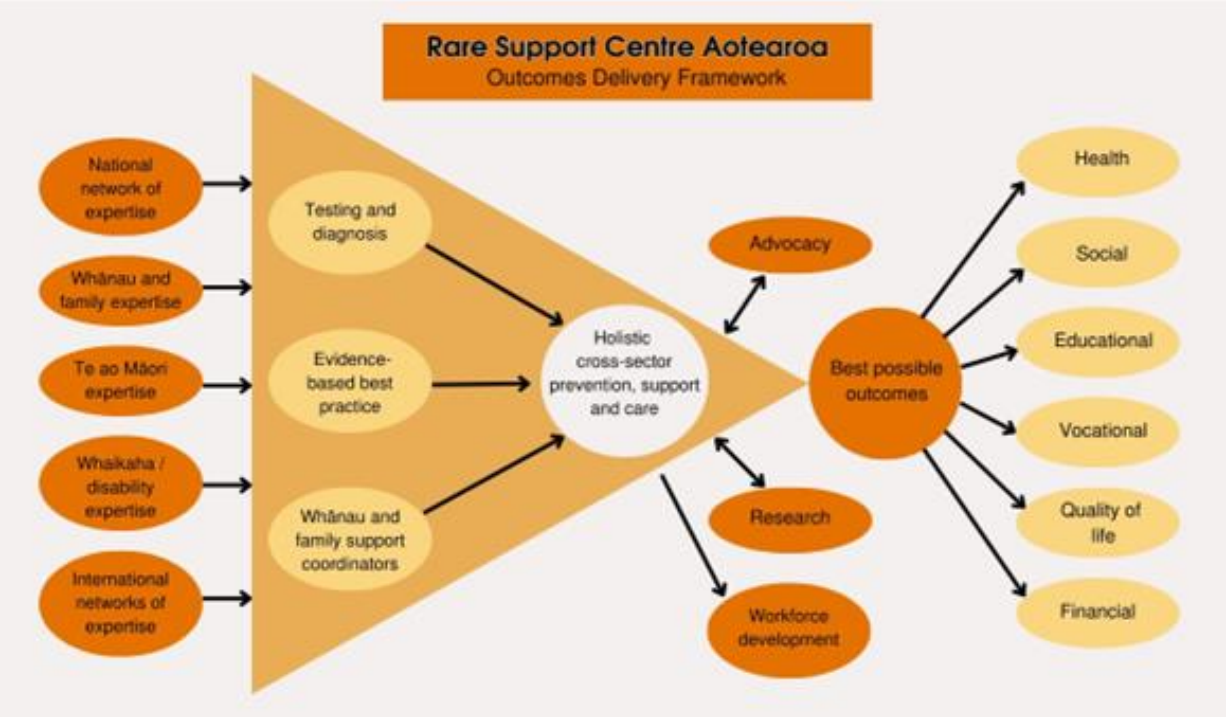
(Rare and Undiagnosed Disorders Centre of Expertise)

"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.