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Kia ora koutou,

Welcome to the latest edition of the **Rare Disorders Research Network Newsletter.**

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Three-year plan for rare disorders research in New Zealand

The Rare Disorders Research Network (RDRN) Leadership Group met recently to develop a three-year plan, along with the project team set up to promote the now established New Zealand ERDERA (European Rare Disease Research Alliance) National Mirror Group.

The plan will set out the impact that rare disorders research should be having on the lives of New Zealanders living with rare disorders, and what steps we can take in the short to medium term to start to bring that impact about.

From the outset there's been a commitment to ensuring that the RDRN's mahi reflects an authentic commitment to being an effective Tiriti partner, and one of the early actions will be to seek advice from Rare Disorders NZ's newly established Māori Advisory Group as to an appropriate te reo identity.

The consultation draft refers to the need to:

- identify areas for future rare disorders research development;
- advocate for this to be prioritised by New Zealand's science and health research systems;
- advocate for increased financial investment;
- strengthen international collaboration and networking (particularly with other countries committed to ensuring that the priorities of indigenous populations are reflected in rare diseases research planning and advocacy);
- ensure that implementation of the New Zealand's Rare Disorders Strategy includes rare disorders data and research, particularly with respect to clinical trials and consumer participation;
- promote the translation of rare disorders research into policy and practice.

RDRN members are invited to give feedback on the consultation draft by emailing chris.higgins@raredisorders.org.nz by Thursday 19 November

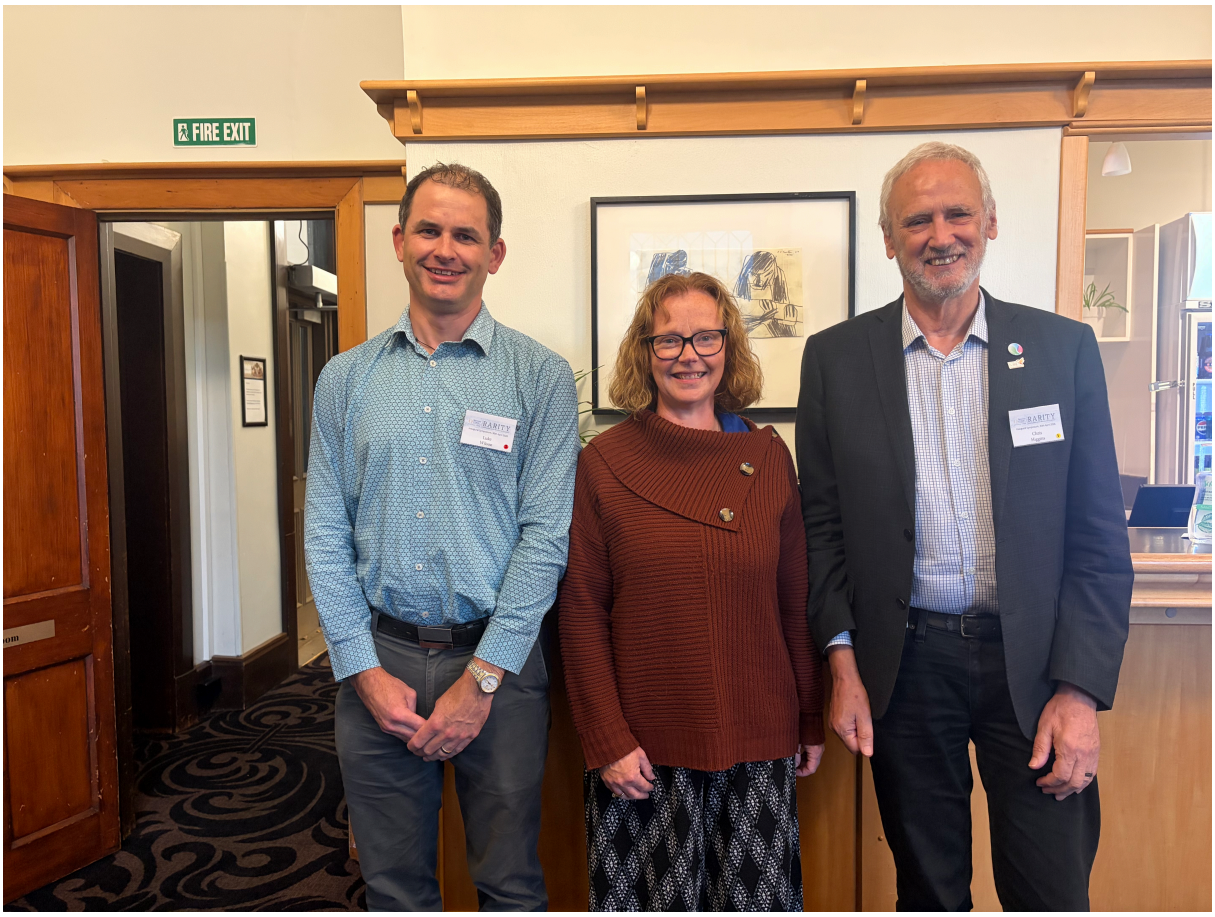
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Rare Disorders Research Forum

In August Rare Disorders NZ (RDNZ) hosted a rare disorders research forum in Christchurch which featured an array of local and international presenters who spoke to themes including: improving outcomes for people living with rare disorders by translating rare disorders research into policy, practice and new therapies; information, coding and data for rare disorders research; connecting globally (with the UK, Australia and Europe), and NZ rare disorders research networks.

Thanks to all the presenters who were happy to share their presentations. These can be viewed together with the programme on RDNZ's website here.



RARITY

RARITY (Research Alliance for Rare Illness Translational Pathways) is a University of Otago initiative led by Professor Stephanie Hughes and Dr Luke Wilson to bridge the gap between laboratory research and real-world clinical and community impact for rare disorders. RARITY and the RDRN are mutually affiliated with each other.

Two recent RARITY-hosted events were an inaugural symposium held in April 2026 and a follow up meeting in October to discuss next steps.

The events brought together a diverse group of members, including researchers, clinicians and patient and consumer advocates, to examine the current state of rare disorders research and care in Aotearoa New Zealand, and to identify opportunities to accelerate translation from discovery to meaningful clinical impact.

The proposed next step is to develop the major themes and recommendations into a national viewpoint on the current rare-disorders research and translation landscape in Aotearoa New Zealand.

To find out more and sign up to the RARITY newsletter email rarity@otago.ac.nz



ERDERA 3rd General Assembly Meeting

The third Annual General Assembly Meeting for ERDERA was held in beautiful Coimbra, Portugal – home to one of Europe's oldest universities, and a UNESCO heritage site. The three-day meeting brought together ERDERA work package leaders and the ERDERA Governing Board, of which Professor Louise Bicknell is the current representative for New Zealand.

It was a fantastic few days, meeting with now-familiar ERDERA colleagues. Louise was particularly able to progress plans for our commonwealth / Indigenous focus for our National Mirror Groups.

Our Australian colleague Professor Hugh Dawkins, who chairs the ERDERA Governing Board, led an excellent session on considering frameworks to help ensure activity and accountability in the Governance Board, with core messages reflecting our own efforts as we get the National Mirror Group operational.

Two documents mentioned that are due to be realised soon that could be helpful to us all – The Lancet Commission Report of Rare Diseases, and the World Health Organisation Global Action Plans for Rare Diseases.

Aligning ERDERA National Mirror Group plans across the Tasman



With the support of Catalyst funding to promote a collaborative approach to both Australia and New Zealand establishing ERDERA National Mirror Groups (NMGs), RDRN Leadership Group members Chris Higgins and Professors Phillip Wicox and Louise Bicknell met recently in Perth WA with Professor Gareth Baynham, Dr Zoë Healey and team members of the wider Rare Care Clinical Centre of Expertise for Rare and Undiagnosed Diseases.

We agreed that it would be important for the two NMGs to collaborate on the content of our respective three-year plans.

The Australian NMG team presented a strategic framework to be one of the foundations of their three-year plan, whereby all of their research is designed to answer the questions asked by people living with rare and undiagnosed disorders, and to connect with the care provided to them, so that they benefit from the research the Rare Care team are doing.

The New Zealand representatives agreed that the same strategic framework should be reflected in the NZ three-year plan, with the elements comprising early diagnosis, access to treatment and therapies, care delivery, support and well-being, and the wider ecosystem which impacts on the efficacy of the other four elements.



We also agreed that both plans should include as a key element our commitments to responding to the priorities, needs and requirements of First Nations people and Māori respectively.

We were privileged and impressed to have been joined for most of the Thursday afternoon session by the convenor of the Western Australia Parliamentary Friends of Rare Diseases, Hon Klasey Hirst MLC (pictured at left with Chris).

Special thanks to Gareth for convening the meetings and introducing us to so many clever and inspiring people, and to Zoe who did a magnificent job of corralling everybody's ideas and contributions into a cohesive whole, with clear next steps and actions.



MALAGHAN INSTITUTE OF MEDICAL RESEARCH

Making every sample count in rare disorders research

The Malaghan Institute of Medical Research is Aotearoa New Zealand's world-class independent biomedical research charity, focused on harnessing the power of the immune system to save lives.

The institute is home to the Hugh Green Technology Centre, a national centre for advanced research technologies that supports researchers across the country with specialist equipment, expertise and training.

Its Bioanalytical Services team processes whole blood to an IANZ-accredited standard (ISO/IEC 17025), with fast turnaround from collection sites nationwide.

For rare disorder research, where participants are few and spread across the motu, this means every sample can deliver reliable insights. Accredited immunoprofiling by flow cytometry is coming in early 2027.

For researchers planning new grant proposals, considering sample processing early can make studies easier to deliver and strengthen the quality of the results.

More information is available on the Hugh Green Technology Centre website, or contact bioanalytical@malaghan.org.nz.

Digital biomarkers show promise for rare disorders monitoring

A significant milestone has been achieved through the BiomarkR collaboration, of which Gareth Baynam and colleagues are included. A recently published study demonstrates the potential of 3D facial imaging and digital phenotyping technologies to identify digital biomarkers in hereditary angioedema.

The research found distinctive facial changes associated with disease flare-ups and provides early evidence supporting the use of artificial intelligence-driven imaging technologies for disease monitoring. While further validation is required, this work contributes to the growing field of digital biomarkers and precision medicine in rare diseases.

Publication: Jamuar S, Palmer R, Lee HY, Chia FL, Goh CB, Lee S, Helmholz P, Chan S and Baynam G (2026) Utility of 3D facial analysis as a biomarker in rare diseases exploration with hereditary angioedema. Front. Med. Technol. 8:1857918. Frontiers | Utility of 3D facial analysis as a biomarker in rare diseases exploration with hereditary angioedema

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Recent achievements for RDRN leadership members





Professor Phillip Wilcox

Huge congratulations to Professor Phillip Wilcox (Ngāti Rakaipaaka, Rongomaiwahine, Ngāti Kahungunu ki te Wairoa), of the University of Otago, Ōtākou Whakaihu Waka, and co-chair of the Rare Disorders Research Network, who's won the Thomson Medal for outstanding leadership to build capability for genomic science in Aotearoa New Zealand.

Phillip cofounded a national genomics network, led the Virtual Institute of Statistical Genetics, and co-led an internship programme for Māori that is part of an international network that builds indigenous capability in genomics.

He has also fostered engagement between scientists and Māori communities, integrated Māori perspectives into research, and co-led initiatives to improve Māori health through genomics, while upholding principles of data sovereignty.

"Genetic variation is a key aspect of biological systems, and this understanding is not restricted to modern science – indigenous peoples have known this too, for millennia." says Phillip

Professor Louise Bicknell

Congratulations to Professor Louise Bicknell who has recently been appointed to the Human Genetics Society of Australasia (HGSA) Board of Directors.

Louise is a Professor of Human Genetics at the University of Otago in Dunedin, New Zealand. Her lab are gene hunters – they are passionate about using their science expertise to help NZ families get answers and they love discovering new variants and especially new candidate disease-associated genes.

They tend to focus on neurodevelopmental and growth disorders and undertake both functional genomics and zebrafish-based studies to generate the needed evidence to support pathogenicity. Their bigger picture goal is trying to help improve diagnostics in New Zealand.

Aside from her research, Louise is involved in the Rare Disorders Research Network, and led New Zealand's involvement in ERDERA (European Rare Disease Research Alliance). She is now Co-Deputy Chair for the ERDERA-aligned National Mirror Group. Louise also keeps busy teaching lots of genetics and molecular biology papers, and supervising students.



Rare Disorders Month, March 2027

In March Rare Disorders NZ will once again be marking Rare Disorders Month in Aotearoa New Zealand to raise awareness about rare disorders and raise vital funds to support our work.

Rare Disorders NZ is passionate about being the collective voice and ‘connector hub’ for rare. We are the first point of contact for many people when diagnosed with a rare disorder. We offer advice and guidance, help navigate the health and support systems and connect people with others with the same condition. We fight for systemic changes to improve the services and support for people with rare disorders. We are also really excited about the potential of the rare disorders research network. Financially however, things are tough and the future less certain.

We are reaching out to you, our friends in the research network, to get behind Rare Disorders Month in 2027 by hosting a morning tea at your workplace just one day during March. Invite your team members along - or even your whole department - to share a plate and contribute a small donation to Rare Disorders NZ.

It is a wonderful chance for your team to come together and enjoy some kai for a cause close to your hearts.

We are grateful for your support so we can deliver more for the rare disorder community.

For more information please contact RDNZ’s Communications Manager Angela Nielsen Angela@raredisorders.org.nz

Rare Disorders Research Network membership

Membership of RDNZ’s Rare Disorders Research Network (RDRN) is open to any recognised New Zealand based researcher who is, has been, or expects to be actively engaged in any aspect of rare disorders research.

If you’d like to join please click here where you’ll be asked to provide details of your credentials including rare disorders research areas of interest, work in New Zealand, institutional affiliations, levels and sources of funding etc.

Benefits of membership include:

- Invitations to member only rare disorders research networking events and forums;
- Opportunities for your research to be promoted to the rare disorders community through RDNZ communications channels;
- Enhanced prospects for the results of your research to be embedded into practice in New Zealand as a result of RDNZ and Support Group advocacy;
- Sharing of knowledge and ideas among colleagues in the rare disorders research community;
- Being kept up to date about international and local rare disorders sector collaborations and developments;
- Opportunities to contribute to the growth of New Zealand's rare disorders research capacity, including the development of a rare disorders research strategy;
- Receipt of and an open invitation to contribute to the Rare Disorders Research Network newsletter.

If you don't meet the criteria to be a network member you're warmly encouraged to sign up to receive our newsletter.

If you don't want to receive this newsletter please contact us and we'll remove you from the distribution list. In either case please email RDNZadmin@raredisorders.org.nz

Ngā mihi nui,
Chris Higgins

RDRN Chair
Chief Executive
Rare Disorders NZ