"You get left behind and lost in a complex world of rare care": equity in access to rare disease care—learnings from the Australian Rare Disease Awareness, Education, Support, and Training (RArEST) project

Elizabeth Emma Palmer,[a,b,*] Krista Recsei,[a] Lauren McKnight,[a] Natalie Roberts,[a] and RArEST Consortium, and Adam Jaffe[a,b]

[a]Discipline of Paediatrics and Child Health, School of Clinical Medicine, Faculty of Medicine and Health, University of New South Wales Sydney, New South Wales, Australia
[b]Sydney Children's Hospitals Network, Randwick, New South Wales, Australia

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Australia is a high-income country, yet the 1 in 12 Australians living with any of the over 8000 rare diseases face significant health inequities.[1,2] These inequities are intrinsically linked to each condition's rarity, which limits recognition, optimal holistic care, and critically needed research and innovation.[2,3] Rare diseases are the biggest killer of children, causing around 60% of childhood deaths.[2,4] People living with rare disease (PLWRD) have more avoidable and prolonged hospitalisations and emergency department attendances compared to people with more common chronic conditions.[5]

Diagnostic delays and misdiagnoses are common. Health professionals have little access to rare disease education and systemic support, so often fail to 'join the dots' to recognise that an individual's myriad of signs and symptoms may represent a rare condition. Over 70% of rare diseases have an underlying genetic basis, but genetic knowledge and access to testing is limited. Consequently, one in two people with a suspected rare genetic condition remains undiagnosed after clinical genomic testing. Even with the promise of advanced therapeutics, rare conditions have few proven treatments (95% lack a targeted therapy[6]) and there is a paucity of natural history data and consensus management guidelines. Thus, PLWRD often experience emerging and unpredictable symptom progression.[2,3] This places a significant burden of hope on clinical trials, yet small individual patient populations, limited knowledge of natural histories, clinical heterogeneity, and inappropriate clinical outcomes are challenges for therapeutic development. PLWRD and their families experience stigma, isolation, uncertainty, and significant educational and financial impacts, leading to a high prevalence of mental health conditions.[7]

These health inequities are greater for certain groups, recognised as priority populations. People living in rural areas generally spend significant time and money travelling for specialist care.[2] Aboriginal and Torres Strait Islander peoples face a legacy of mistreatment, ongoing institutional racism and a lack of culturally safe care.[8] Culturally and linguistically diverse people lack care that reflects their cultural norms and resources in their languages.[2] Further, information on rare diseases is often inaccessible to people with low health literacy and/or disabilities. A paucity of publicly funded rare disease centres of expertise means specialists and therapists may only be accessible privately, with associated out of pocket payments. This compounds financial impacts and can place expert care out of reach.[2,3,8]

The need for equitable rare disease care for *all* was recognised in Australia's National Strategic Action Plan for Rare Diseases (the Action Plan).[2] Its development was led by Rare Voices Australia (RVA), the national peak body for PLWRD. The Action Plan was co-designed with PLWRD and highlights the importance of involving the intended recipients when designing and implementing new models of care. The link between rare disease care and universal health coverage is a key message of the United Nations General Assembly's recently adopted resolution (26/132) "Addressing the Challenges of Persons Living with a Rare Disease and their Families". As we stand on the brink of a World Health Assembly resolution calling for rare diseases to be a global health priority, it is timely to reflect on progress towards that goal in Australia.

How the Rare Disease Awareness, Education, Support, And Training (RArEST) project applied co-design to reduce inequities

The RArEST Project was run by a national consortium and funded by the Australian Government from 2021 to 2024. The project co-designed practical resources to support all PLWRD, health professionals, advocates, and decision makers.[9]

*Corresponding author. Discipline of Paediatrics and Child Health, School of Clinical Medicine, Faculty of Medicine and Health, University of New South Wales Sydney, New South Wales, Australia.
E-mail address: elizabeth.palmer@unsw.edu.au (E.E. Palmer).

RArEST tackled rare disease inequity head on through three innovative approaches. Firstly, traditional power imbalances between academia and those with lived experience were reduced by including RVA as a consortium member, alongside university and health professionals. Secondly, the project prioritised authentic co-design with all intended audiences. Lastly, people facing greater inequities (as outlined above) were involved throughout to improve accessibility, inclusion, cultural safety, and appropriateness.

A stakeholder reference group comprising PLWRD and health professional working groups facilitated co-design and collaboration from start to end. Provision of appropriate remuneration and support demonstrated respect and promoted ongoing engagement with often marginalised communities.

The result was a suite of tailored educational resources, freely available on the RVA website https://rarevoices.org.au/rarest-project/ (Fig. 1).

Highly impactful outputs were co-designed factsheets, e-learning modules, and videos that destigmatise the effect of rare disease on mental health and wellbeing, and signpost credible digital mental health services.[9] These 1) help PLWRD access appropriate supports in a timely manner 2) guide mental health professionals in tailoring their care to PLWRD, and 3) guide other health professionals in considering and asking about the mental health impacts of rare disease. The case study videos have been viewed over 1300 times since their launch in May 2024 and used in undergraduate medical education. The factsheets have been viewed over 2370 times since May 2024.

Australia's first **National Recommendations for Rare Disease Health Care** (The Recommendations)–launched in Australia's Federal Parliament and viewed by over 2000 people globally over the first 7 months–was another key output from the project.[10] They were endorsed by 11 national professional bodies and officially recognised as an Accepted Clinical Resource by the Royal Australian College of General Practitioners (RACGP). The Recommendations are deliberately disease-agnostic and aimed at *all* health professionals to address common challenges, leverage common opportunities, and promote coordinated approaches.

Embedded throughout are links to practical tools that help health professionals apply the recommendations to

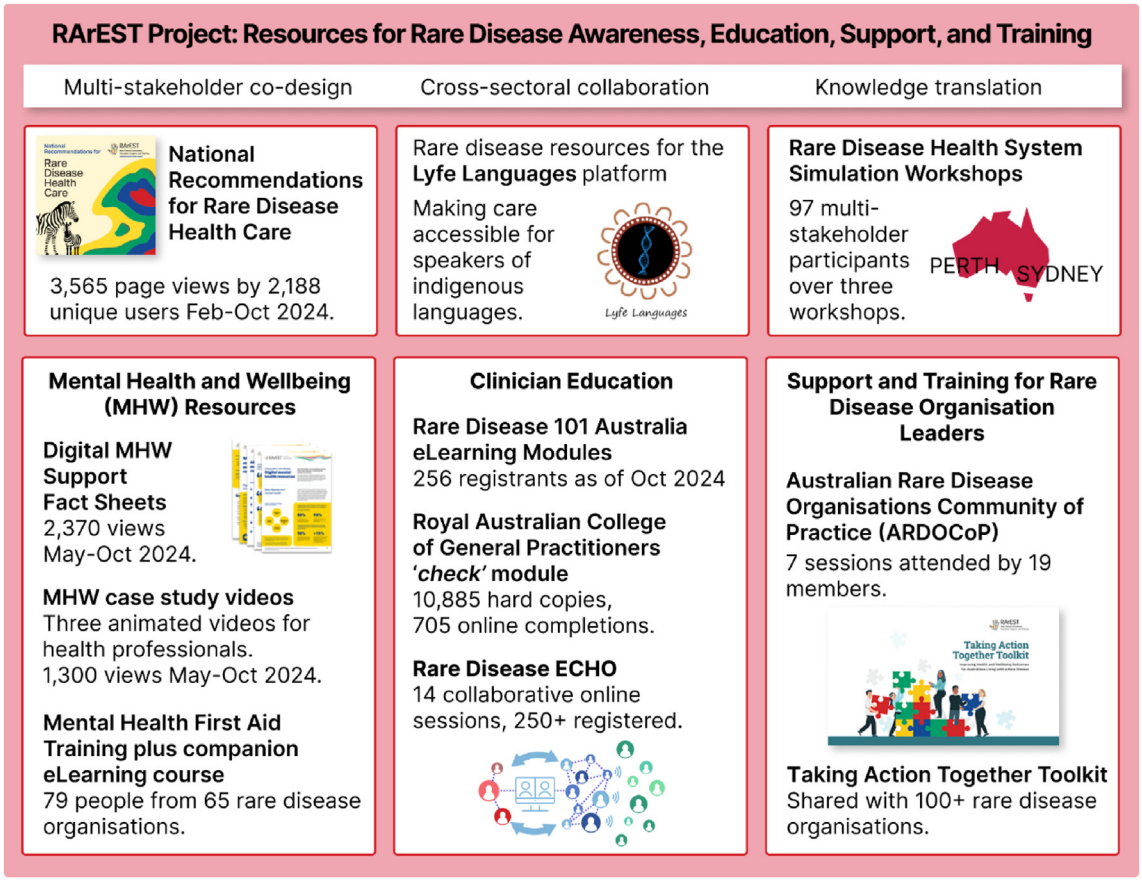

Fig. 1: Co-designed resources from the RArEST Project.

clinical practice, and links to other educational resources co-designed through the project. For example, the Rare Disease Australia 101 e-learning module and a rare disease Project ECHO community of clinical learning practice, both accredited for Continuing Professional Development by the RACGP. This resource also includes suggestions for gathering evidence to monitor the impact of enacting the recommendations.

To address inequities among Aboriginal and Torres Strait Islanders, RArEST co-designed rare disease information for the Lyfe Languages online platform (https://app.lyfelanguages.com), which translates complex medical terminology into Indigenous languages in text and audiovisual formats. This includes written information and culturally appropriate videos for patients and families on genomics and rare disease.

RArEST worked closely with diverse patient advocacy groups, supporting a community of practice and co-designing the "Taking Action Together Toolkit" which highlights examples of effective systematic advocacy along with useful tools.

Feedback and evaluation show the resources reflect the lived experience of PLWRD and are applicable to clinical practice. Co-design had additional impacts, with participants reporting the experience was empowering. Clinicians valued the practicality of the resources, reporting greater confidence in taking steps to improve rare disease care. Many resources had unanticipated international reach (Fig. 1), suggesting they are filling a global gap.

Next steps needed to bring equity of access to high value rare disease care

The RArEST Project was an important step towards achieving equitable, person-centred, and evidence-based rare disease care in Australia. The project concluded with recommendations to further strengthen the Australian health care system.[9]

Firstly, proactive mental health and wellbeing support should be embedded into rare disease-specific care settings. Complementing this, rare disease awareness should be included in mainstream mental health and wellbeing workforce training.

Rare disease education and training should be provided to all health professionals, not just the motivated 'early adopters'. This requires integration into all health professionals' curricula and mandatory training. Recognising the complexity of both rare conditions and our health systems, education and training must be coupled with clear pathways to rare disease experts and expertise, for patient referral and health professional support and guidance. Equally important is efforts to continually empower and collaborate with PLWRD, so they can make informed decisions including on new and evolving diagnostic and therapeutic technologies.

Clinical Guidelines or Standards for rare disease care should be co-designed, building on the Recommendations, and clinicians should be supported to implement these into routine practice. Rare disease organisations should be given sufficient funding and other supports so they can build their capacity to provide critical support and advocacy for PLWRD.

Investment is needed to co-design and implement the infrastructure to support all PLWRD, including networks of rare disease centres of expertise, registries, and clinical trial networks. Rare disease centres of expertise could harness and foster diverse health professional expertise; support clinicians to deliver evidence-based and coordinated care in the community; connect PLWRD to research pathways to enable timely diagnoses and advanced therapeutics; and coordinate with disability, education, employment, and other services for a cross-sector approach to reduce fragmented care and isolation.

Importantly, these initiatives should respectfully engage with priority populations, so all Australian PLWRD receive the best attainable care.

Authentic co-design and practical implementation can bring health professionals, researchers, and policy makers on the rare journey with PLWRD. Together, we can ensure all PWLRD have access to timely diagnosis and evidence-based care that aligns with their personal and cultural needs, leaving no person living with rare disease behind.

Contributors

EEP, Conceptualisation, data curation, formal analysis, funding acquisition, methodology, project administration, supervision, visualisation, writing–original draft, writing–review and editing.

KR: Literature search, project administration, writing–review and editing.

LM: Conceptualisation, data curation, formal analysis, investigation, methodology, visualisation, writing–review and editing.

NR: Literature search, project administration, writing–review and editing.

AJ: Study design, steering committee membership, data interpretation, review of manuscript.

Declaration of interests

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EEP is a member of the Rare Voices Australia (RVA) Scientific and Medical Advisory Committee and receives flights and accommodation to attend RVA meetings.

AJ has received speaker fees from Vertex Pharmaceuticals in the last 24 months, and in the last 36 months has been the Chair of the RVA Scientific and Medical Advisory Committee.

LM, KR, and NR declare no competing interests.

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Roberts, Discipline of Paediatrics and Child Health, School of Clinical Medicine, Faculty of Medicine and Health, University of New South Wales Sydney, NSW, Australia; Yvonne Zurynski, Australian Institute of Health Innovation, Macquarie University, Sydney.

Appendix A. Supplementary data

Supplementary data related to this article can be found at https://doi.org/10.1016/j.ebiom.2025.105710.

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