

SUMMARY:

RARE DISORDERS INSIGHTS REPORT: PATHWAYS TOWARDS BETTER HEALTH OUTCOMES

ABOUT RARE DISORDERS NZ

Rare Disorders NZ is the national peak body organisation for rare disorders in New Zealand and provides a strong common voice to advocate for an equitable healthcare system that works for the 300,000 Kiwis with a rare condition.

ABOUT RARE DISORDERS

Rare disorders are often defined as disorders that affect between one in 2,000 people, or one in 3,000 people. While each individual rare disorder is uncommon, in total they are estimated to affect 300 million people worldwide.¹ Rare disorder patients represent a large group in any country's health system, they face more barriers, and suffer significantly more from their conditions than other patients. Therefore, this group needs special attention.

ABOUT THE REPORT

Developed by Business and Economic Research Limited (BERL), Rare Disorders NZ commissioned the report to obtain an insight into the likely prevalence of rare disorders in New Zealand, considering both diagnosed and undiagnosed incidences. The report is based on publicly available data and a high-level literature scan of recent New Zealand and international studies that discuss the prevalence of rare disorders, as well as burden factors that need to be considered in the development of health policies.

KEY FINDINGS

- The lack of data regarding rare disorders in New Zealand is not only a challenge to estimating prevalence, but also for improving diagnosis, treatment, and research.
- The classification system for diseases that New Zealand uses does not include most rare disorders.
- New Zealand may be performing worse than its peers in diagnosis speed and accuracy.
- Pharmac's own definition of rare disorders, at a prevalence of less than one person in 50,000, means that there is a rare disorder medicines funding gap.

RECOMMENDATIONS

Awareness and training

- Advocate for the Government health agencies to fund a study with health and support professionals to gather evidence of their level of awareness and knowledge of rare disorders
- Partner with the University of Otago and the University of Auckland to conduct a similar study with healthcare students
- Advocate for the Government health agencies to fund awareness-raising campaigns and events in New Zealand
- Advocate for the University of Auckland and the University of Otago to further include rare disorders in their curricula
- Advocate with The Royal New Zealand College of General Practitioners and The Royal Australasian College of Physicians, for the Medical Council of New Zealand, and the Australian Medical Council, to further include rare disorders in the Continuing Professional Development (CPD) curricula
- Advocate for the Government health agencies to implement digital resources that support health professionals reach a diagnosis and a treatment plan.

Research and data

- Advocate for the Government health agencies to develop a rare disorder patient registry, which uses the Orphanet classification.

Diagnosis and treatment

- Advocate for the Government health agencies to conduct a comprehensive review of the National Screening Unit.
- Advocate for the Government health agencies to further develop rare disorder guidelines.
- Advocate with the Genetic Health Service NZ for the Government health agencies to allocate additional funding for genetic testing.
- Consider the possibility of further advocacy in regards to genome sequencing in newborns, depending on the results from England in regards to the efficiency of genome sequencing in newborns.
- Advocate for the Government health agencies to adopt an official definition of rare disorders that is in line with international best practice.
- Advocate for Pharmac to expand its rare disorders medicines contestable funding pilot, and to admit that under the current model a significant proportion of people living with a treatable rare disorder are not being covered.
- Advocate for the Government health agencies to take advantage of the health system unification reform and implement a fully integrated country-wide information sharing system.
- Advocate for the Government health agencies to develop an expert centre for rare disorders in the country.