Australian Genomics

Research Snapshot
June 2024

Tasmanian Rare Disease Diagnostic Pathways Project

The Tasmanian Clinical Genetics Service within the Tasmanian Health Service was funded by Australian Genomics to undertake research into improving diagnostic pathways for rare diseases in regional Australia.

Aims

The project aimed to:

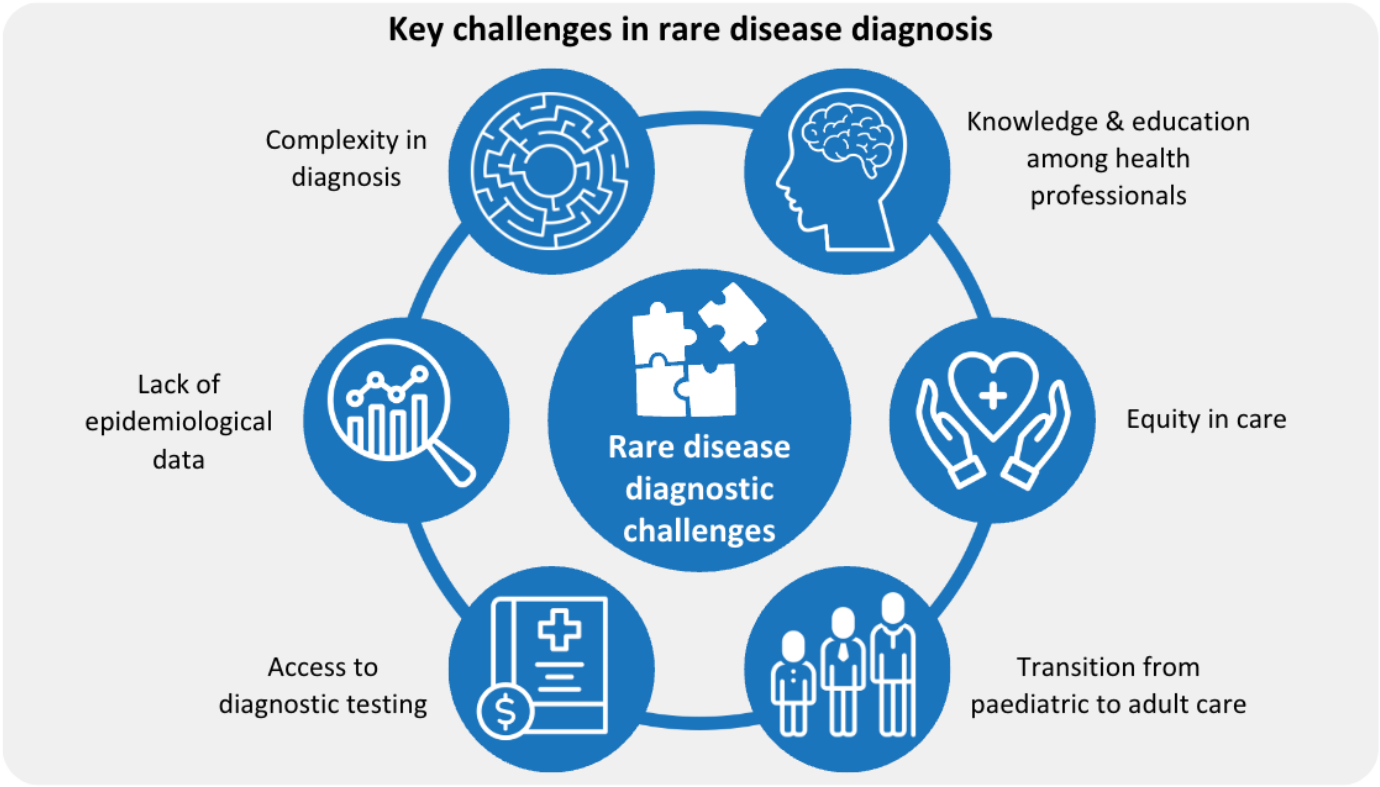
- Map the current pathways to rare disease diagnosis in Tasmania and identify barriers
- Understand community needs with regards to diagnostic pathways for rare disease
- Develop innovative service models and recommendations to address barriers to diagnosis

Background

There are more than 7000 known rare diseases in the world. Individually they are rare, but collectively they are common. They affect between 6 to 8 per cent of Australians and 35,000 to 45,000 Tasmanians. About 80 per cent of rare diseases are genetic. They are often debilitating, life-limiting and affect multiple body systems, but are difficult to diagnose.

About 20 per cent of Tasmania's hospital costs can be attributed to people with a rare disease. The long diagnostic delays often experienced by patients and misdiagnosis, which is common, lead to poor health outcomes and inefficient health care expenditure. These issues are exacerbated in rural and regional populations like Tasmania's.

Rare diseases can share common characteristics at presentation which means earlier recognition is possible. A diagnosis, which is essential to access the most appropriate care, may require genetic testing and the expertise of clinical geneticists and genetic counsellors. Rare disease diagnostic pathways can set out a clear process for clinicians to follow and guidance for post-diagnostic care.



What the community told us

More than 850 patients and carers affected by rare disease in Tasmania participated in the research, along with clinicians, advocates and experts in rare disease. Among the issues identified were:

- A lack of access to GPs and specialists in regional and rural areas
- A lack of empathy, respect and listening from clinicians, which resulted in diagnostic apathy and misdiagnosis
- Poor coordination and communication between health professionals and from health professionals to patients
- A lack of knowledge about rare diseases among primary health clinicians and specialists
- A lack of support for patients to negotiate a complex health system
- Cost and travel barriers for some patients in seeking a diagnosis
- Low awareness among patients, carers and clinicians of public clinical genetic services.

Improving rare disease diagnostic care

The study proposes a range of actions to improve Models of Care for rare disease diagnosis in Tasmania. These actions seek to address the barriers identified by stakeholders and improve access to information; access to the health system; the knowledge and attitudes of clinicians, coordination and integration of care; timely and appropriate investigations; access to research-based treatments; and access to support.

Among the recommendations is a proposal to establish a **Rare Care Centre** that would be a one-stop shop for:

- clinicians and patients seeking information and support
- access to multi-disciplinary clinical teams providing coordinated diagnostic and post-diagnostic care incorporating clinical geneticists and counsellors, specialists, primary care clinicians, nursing and allied health professionals
- the facilitation of access to research-based treatment and
- the facilitation of access to non-clinical support services.

Conclusion

This project implemented mixed methods research to provide a detailed understanding of the barriers to rare disease diagnosis in Tasmania and identified targeted solutions and strategies to overcome these barriers. It provides a comprehensive and up-to-date map of current care pathways and community needs and sets out innovative models of care and recommendations that have been developed in consultation with the community, clinicians, and researchers.

While the research centred on Tasmania, many of the findings and recommendations are relevant to other regional and rural areas of Australia.

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