# Genomics of Rare Disease (GRD) Registry

A national, participant-led research platform for Australians with a rare disease of known or suspected genetic cause, led by A/Prof Jodie Ingles and A/Prof Owen Siggs at the Garvan Institute of Medical Research (HREC 95179, Royal Children's Hospital).

- Recently funded by the Clive and Vera Ramaciotti Foundation: 2025 Ramaciotti Biomedical Research Award (https://www.perpetual.com.au/wealth-management/ramaciotti/ramaciotti-awards-and-recipients/)
- A national pathway to enrol monogenic cases for future research and further investigations.
- Increased representation of Australian communities currently missing from international genomic databases
- Returnable, research-grade relevant results, supporting diagnosis, counselling, and management

**Who can be referred (Registry):**

- Any age; alive or deceased (with next-of-kin consent)
- Resident in any Australian state or territory, or an Australian citizen
- Diagnosed with a rare disease (prevalence <1 in 500) of known or suspected genetic cause — or an at-risk family member
- No rare disease populations or conditions are excluded

**Gene Discovery sub-study — for unsolved cases:**

Registry participants with a high likelihood of a monogenic cause but no conclusive genetic diagnosis can be invited to a Gene Discovery Study, where research-grade whole-genome sequencing is applied to identify a cause. Clinically relevant findings are returned to the participant's treating clinician.

**What participation involves:**

- Online surveys at baseline and follow-up questionnaires
- A biospecimen (blood, saliva, or tissue) for the Gene Discovery arm
- A national pathway to enrol unsolved monogenic cases for further investigation
- Increased representation of Australian communities currently missing from international genomic databases