PreGen Inclusion Criteria

General Inclusion Criteria

- PreGen is limited to those families who will undergo genomic diagnostic testing as a family trio or where both the mother/egg donor and father/sperm donor are available for testing.

Single parent families or those requiring a gene panel or singleton test may access prenatal genomic testing but cannot be included in the translational PreGen project.

Eligible participants must also be able to:

- Provide informed consent
- Speak/read conversational English
- Be eligible for Medicare

Testing in PreGen is limited to:

- Families where the fetus is believed to be alive at the time of enrolment (preterminal imaging findings are exclusion criteria)

Clinical Inclusion Criteria

A fetus with a structural anomaly likely to have a single gene germline aetiology including:

- A significant/ severe brain abnormality
- A significant cardiac abnormality
- Renal anomalies with a likely Mendelian basis
- A phenotype consistent with skeletal dysplasia
- Evidence of multi-joint arthrogryposis
- Non-immune fetal hydrops

Significantly abnormal biometry:

- Growth restriction (< 1st centile) without placental insufficiency

Clinical Exclusion Criteria

- The family do not wish to take part in PreGen
- A definite decision already made for pregnancy termination
- Likely non-genetic or undiscoverable aetiologies including teratogenesis, viral infections, and poorly controlled maternal diabetes