Diagnosis and Phenotype Characterisation Using Genomics in Patients With Inherited Bone Marrow Failure (IBMDx Study)

NCT05196789

Status	RECRUITING
Sponsor	Peter MacCallum Cancer Centre, Australia
Enrollment	350 participants

Key Eligibility Criteria

Inclusion (3)

- age e 3 months
- able to give informed consent (or parent/guardian able to give informed consent)
- a clinicopathological diagnosis (or differential diagnosis) of inherited bone marrow failure syndrome or related disorder (IBMFS-RD) as per the study team

Exclusion (2)

- A clinicopathological diagnosis of an acquired bone marrow failure syndrome (including acquired aplastic anaemia and hypoplastic myelodysplastic syndrome) as per the study team
- Existing definitive genomic diagnosis for patient's haematological phenotype

Locations (1 total)

Peter MacCallum Cancer Centre, Melbourne, Victoria, Australia