Diagnosis, discovery and novel phenotype characterisation using multimodal genomics in patients with inherited bone marrow failure and related disorders (IBMDx study)

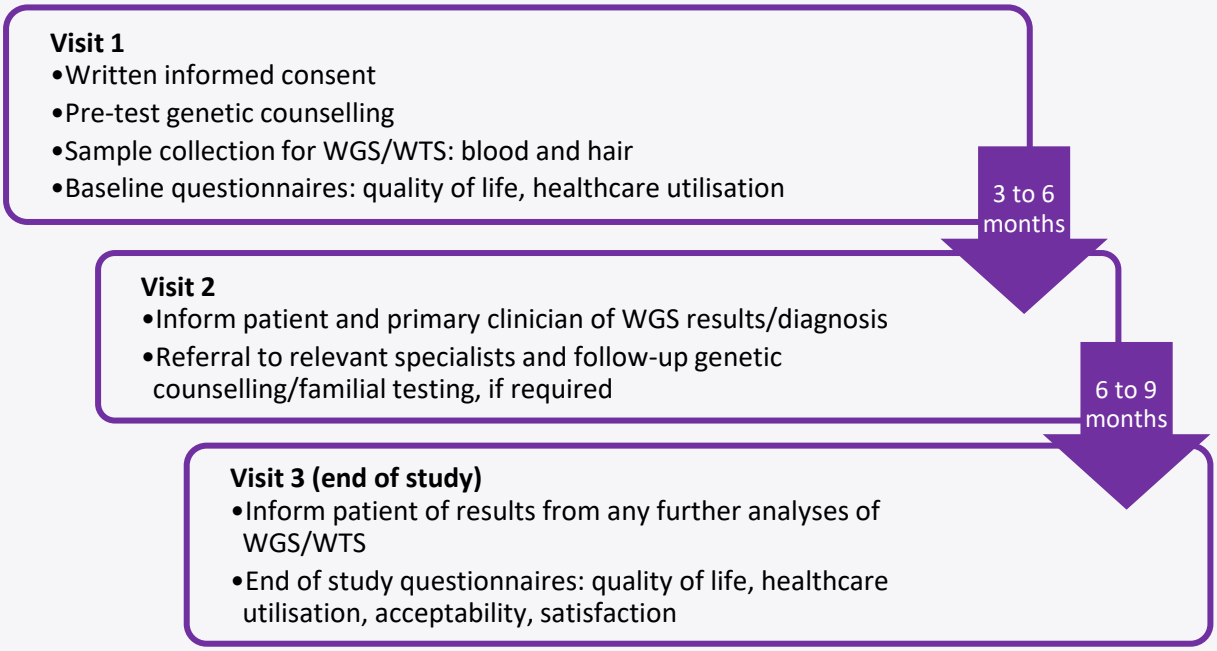
Inherited bone marrow failure syndromes and related disorders (IBMFS-RD) are a group of genetic diseases that are phenotypically heterogeneous and can present throughout lifetime. These syndromes are individually and collectively rare, and may be difficult to diagnose based on clinical presentation and examination. The Melbourne Genomics Health Alliance (MGHA) BMF Flagship, which provided whole exome sequencing for patients with bone marrow failure between 2017-18, demonstrated the impact of comprehensive genomic testing on achieving an accurate diagnosis (doi:10.3324/haematol.2019.237693).

The IBMDx study aims to provide whole genome and transcriptome sequencing (WGS/WTS) to 350 patients with IBMFS-RD. The efficacy of WGS/WTS in establishing an IBMFS-RD diagnosis will be examined as well as exploration of health economic impacts and health implementation challenges.

Study population

Inclusion criteria	Exclusion criteria
• age ≥ 3 months • able to give informed consent (or parent/guardian able to give informed consent) • a clinicopathological diagnosis (or differential diagnosis) of IBMFS-RD	• A clinicopathological diagnosis of an acquired BMFS-RD (including acquired aplastic anaemia and hypoplastic myelodysplastic syndrome) • Existing definitive genomic diagnosis for patient's haematological phenotype

Study visits



Please email IBMDxstudy@petermac.org if you have any potential patients to discuss, or if you have any questions for the study team.