# The EMBRACE Trial Evaluating MultidisciplinaRy Bone maRrow fAilure CarE trial

Principal investigators: Dr Lucy Fox and Dr Piers Blombery

2020 ongoing

The Evaluating MultidisciplinaRy Bone maRrow fAilure CarE (EMBRACE) trial is a prospective observational clinical trial being undertaken by the combined Clinical Haematology Department at the Peter MacCallum Cancer Centre and The Royal Melbourne Hospital. The trail is open to patients from one month old, and offers molecularly-guided individualised care to Australian patients with a either a suspected Bone Marrow Failure Syndrome (BMFS) or an inherited predisposition to blood cancer, and expert management advice to clinicians referring these patents.

Patients enrolled on the trail will undergo molecular genetic testing for approximately 90 known current genes responsible for inherited BMFS and predisposition to haematological malignancy. Prior to the availability of genomic testing, establishing an accurate diagnosis was extraordinarily complicated, and only achieved in a proportion of cases with traditional testing. Patient stories of the 'diagnostic odyssey' (the time taken from the onset of symptoms to the receiving a definitive diagnosis) are absolutely heartbreaking – families who had no idea what was so desperately wrong with their child for years, enduring multiple hospital admissions, failure to thrive, and only a generic diagnosis of 'ugly bone marrow'.

Access to genomic testing has revolutionised the ability to provide an accurate clinicogenomic diagnosis in a matter of weeks. The EMBRACE trail will ensure positive and lasting impact to Australian patients by providing a co-ordinated and efficient approach to case identification, diagnosis, multidisciplinary management, and genetic counselling and management to affected family members