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ACADEMIC MEDICAL CENTRE

Genomic
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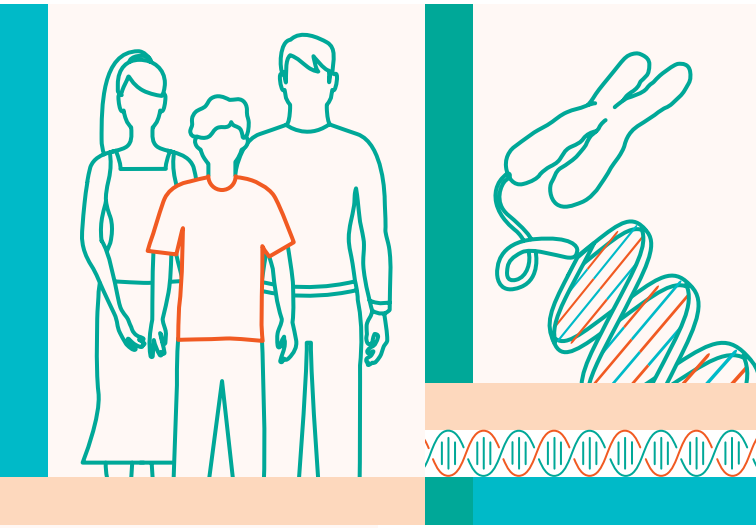
Inherited Bone Marrow Failure Syndromes



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What are inherited bone marrow failure syndromes?

Inherited bone marrow failure syndromes (IBMFS) are a group of genetic conditions characterised by the inability of the bone marrow to produce sufficient blood cells.

These syndromes are usually caused by a fault (i.e., mutation) in a gene that is meant to repair DNA (the instructions our body needs to function). This may lead to bone marrow failure, and in some cases, a higher risk of developing solid tumour(s) and/or blood cancer such as acute myeloid leukaemia (AML).

Not all cases of bone marrow failure are inherited and passed down within families. Some cases may be acquired – caused by environmental factors, chance events and age.

Genetic testing can help identify if a person's medical history is caused by a faulty gene that may be inherited.

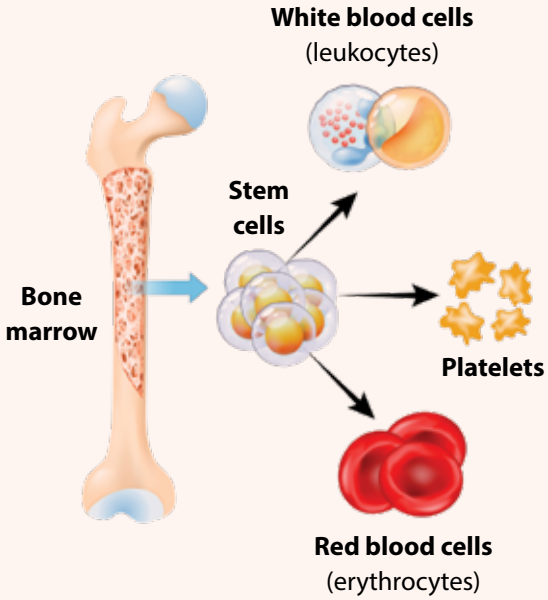
What is bone marrow failure?

The bone marrow contains blood stem cells, which function to produce the different types of blood cells.

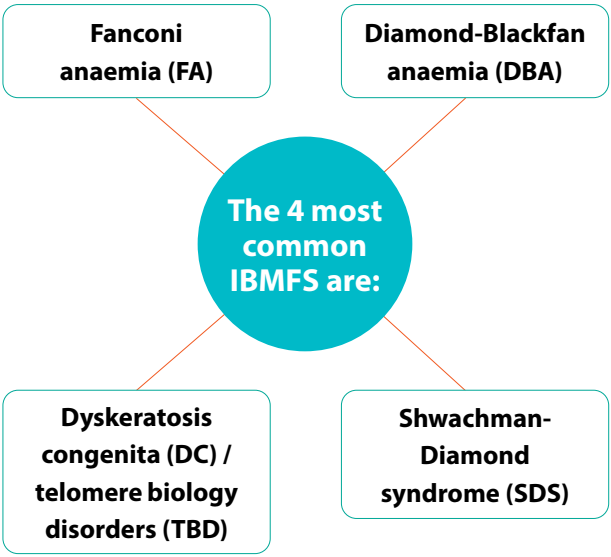
Individuals with bone marrow failure typically start by exhibiting low blood count(s) in the following cell types:

- Low red blood cell (RBC) count *(anaemia)*
- Low platelet count *(thrombocytopaenia)*
- Low white blood cell (WBC) count *(leukopaenia)*

The different cells made from blood stem cells in the bone marrow



IBMFS may present at birth, infancy or childhood, and sometimes in adults. Both males and females can be affected.



These conditions are associated with different faulty genes.

How are IBMFS diagnosed?

Different tests are used to diagnose and differentiate IBMFS.

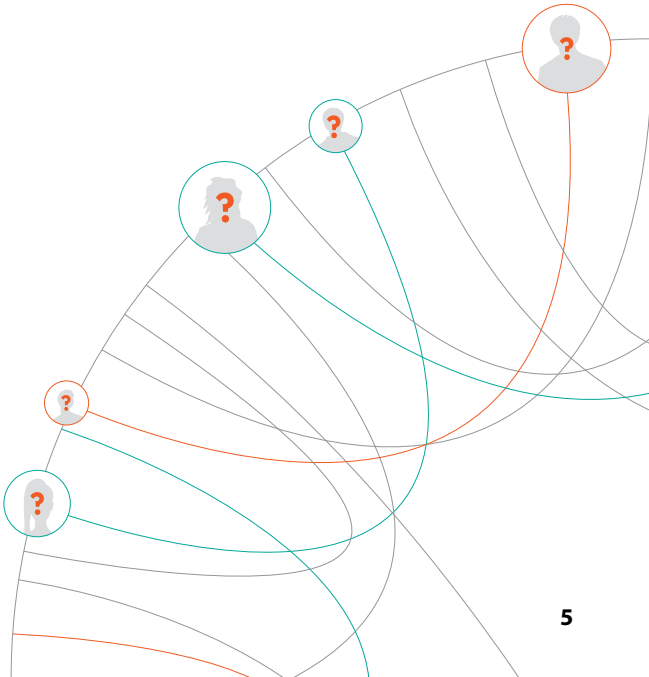
These include:

1. Complete blood count (CBC) test to check for low RBC, WBC and platelet counts.
2. Bone marrow aspiration and biopsy. The bone marrow sample is then viewed under a microscope to determine if the bone marrow is working.
3. Genetic testing to identify a causative faulty gene (if any), using a blood, saliva or skin sample.
4. Chromosome breakage test to rule out FA.
5. Telomere length test to exclude other genetic conditions that affect telomeres like DC or TBD.

Who should undergo genetic testing for IBMFS?

You should consider genetic testing if you or your family members meet one or more of the following criteria:

- Personal history of a bone marrow disorder suggestive of IBMFS
- Diagnosis of aplastic anaemia (AA), myelodysplastic syndrome (MDS), acute myeloid leukaemia, primary immunodeficiency and other bone marrow disorders below age 40
- Excessive bleeding or toxicities while on treatment for a solid tumour
- Family history of bone marrow disorders, blood problems, cancer or immune issues
- A previously identified faulty gene causing IBMFS in the family



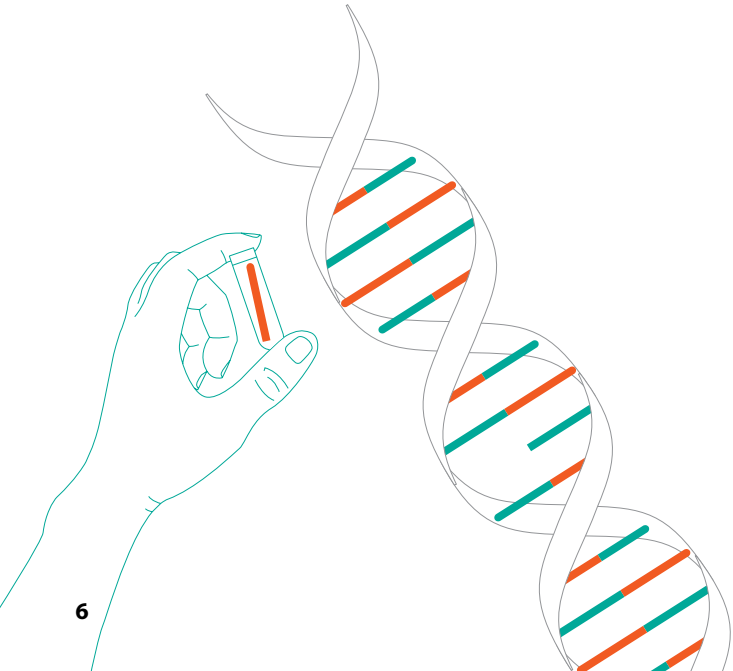
How can your genetic test result help you?

If a faulty gene is identified, it can help guide your options for treatment and management.

- By identifying which treatments to use/avoid
- Individuals with Fanconi anaemia (FA) are sensitive to DNA-damaging agents and would require modified treatment regimens
- Guide the selection of potential donors for bone marrow transplant

If you are currently asymptomatic, it can help guide relevant screening and blood tests to detect bone marrow failure or cancer at its earliest, most treatable stage.

Genetic testing to identify these faulty genes will allow for personalised treatment and management options.



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If you wish to support the Cancer Genetics Service's education, research and patient support efforts, please scan the QR code to make a donation.

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