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SMD. Stem Cell and Myeloid Disorders

NIHR BioResource – Rare Diseases study project

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Summary

All blood cells are made in the bone marrow. They all start off as stem cells and then become more and more specialised into red blood cells (carry oxygen around the body), different types of white blood cells (fight off infections), and platelets (stop bleeding).

Genetic abnormalities in blood cell production can result from defects all along this maturation pathway and patients can present with anaemia, infections or bleeding. A single type of blood cell can be affected (cytopenia), or several types can be affected together. Sometimes all three types of blood cells are reduced (pancytopenia).

It is important for patients to know their genetic diagnosis for several reasons. In some cases, there is overlap between different types of inherited cytopenias, and a definitive diagnosis will allow selection of the appropriate treatment. This can result in a dramatic change in management and a better prognosis. In other cases, a genetic diagnosis gives important information for genetic screening of family members- this can be critical when certain types of treatments are considered, such as bone marrow transplantation, or for family planning. Finally, sometimes knowing the exact genetic cause for the condition can inform the family about potential responses to treatment or long term prognosis.

Recruitment Criteria

Inclusion

A suspected or likely diagnosis of a rare genetic disorder causing persistent or recurrent cytopenia or cell function disorder or pancytopenia of known or unknown cause following review by a haematologist. (*Please turn over.*)

For patients with isolated thrombocytopenias, ***see: BPD project*** *('Bleeding thrombotic and Platelet Disorders') recruitment criteria*.

Exclusion

Acquired causes of cytopenia or pancytopenia including drugs, viral infections and clear-cut autoimmune cytopenias responsive to first-line treatment.