NIHR | BioResource

HBP. Haemoglobinopathies

NIHR BioResource – Rare Diseases study project

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Summary

Haemoglobinopathies are inherited disorders of haemoglobin: Sickle cell disease and thalassaemia.

Sickle cell disease (SCD) is due to an abnormality in the B globin chain of haemoglobin leading to the production of abnormal red blood cells. These cells are fragile and sticky so they break down (haemolyse) quickly, causing anaemia and clump together to block small blood vessels (vaso-occlusion). It is associated with acute severe pain episodes (the most common cause of hospital admission), acute stroke and acute respiratory complications. Chronic complications include cardiopulmonary damage, renal dysfunction, splenic damage, retinopathy and leg ulcers. About 15,000 people are affected by SCD in the UK and although survival has improved over the last two decades, it is still associated with early death (median 40-50 years) and significant chronic morbidity. There is marked phenotypic variation in SCD and this is poorly understood.

Dr Jo Howard (top) & Dr Sara Trompeter, HBP project Leads

The BioResource will enable the development of a resource for researchers to investigate this variability. It is often treated with blood transfusion and although this is effective treatment there is a high rate of development of allo-antibodies to blood which can lead to delayed haemolytic transfusion reactions and can render patients untransfusable. The genetics of blood grouping are complex and the BioResource will be a useful tool to understand this better and to ensure we are giving the most appropriate blood to our patients.

Thalassaemia is due to a decreased or absent production of B globin or alpha globin chains leading to the development of anaemia. There are a large number of B thalassaemia genetic variants and the more severe variants cause severe anaemia requiring life long regular transfusion. This in turn leads to iron accumulation, primarily in the heart, liver and endocrine organs, and this iron accumulation leads to early death, most often from cardiac dysfunction. Patients require life long iron chelation therapy. There are around 800 patients with transfusion dependent thalassaemia in the UK. Patients with less severe forms of thalassemia may need intermittent transfusion therapy and iron overload develops later in life. Other complications associated with thalassaemia include an increased risk of pulmonary hypertension, leg ulcers and osteoporosis. The BioResource will help us to understand variation in propensity to iron overload, response to iron chelation and will also help to improve the matching of blood for transfusion.

Recruitment Criteria

Inclusion

Patients with a confirmed clinical diagnosis of sickle cell disease or thalassaemia.

Exclusion

Sickle cell trait.
Thalassaemia trait.