What is Hereditary Persistence of Fetal Haemoglobin (HPFH)?

Haemoglobin (Hb) is a protein in red blood cells that carries oxygen around our bodies. It is composed of iron and globin chains called alpha (α), beta (β), and gamma (γ). Mutations in the globin chains leads to a different from normal (variant) haemoglobin being produced (resulting in a condition called a haemoglobinopathy). At birth, fetal haemoglobin (HbF) levels are high but several months post birth falls to <1% of the total detectable haemoglobin.

HbF can be increased in one of two ways. The most common is via *acquired* causes (e.g. pregnancy, bone marrow failure syndrome(s), some drug therapies such as hydroxyurea) or *congenital* (ie a genetic cause inherited from parents) which is called ***Hereditary Persistence of Fetal Haemoglobin (HPFH).*** Acquired causes are transient unlike HPFH which is present for life.

Inheritance of HPFH

- A person normally inherits two alpha and two gamma genes (one from each parent), to produce the alpha & gamma globin proteins in haemoglobin F.
- A person may have a deletion or mutation in one of their two gamma globin or promotor genes.
- HPFH can be inherited with other haemoglobinopathies and this co-inheritance can potentially improve the clinical presentation of the disorder.
- Just like the genes that determine hair or eye colour, the gene for HPFH is inherited from one or both parents.
- Importantly HPFH is NOT contagious and NOT transmitted by germs and is of no major clinical concern.

Diagnosis

- Can be identified via a haemoglobinopathy/thalassaemia screening blood test
- Newborn infants normally have large amounts of HbF. Testing is therefore recommended (if needed) after 12 months of age.

Treatment

HPFH is a *benign* clinical condition, and so treatment for this condition is not required.

HPFH and family planning

HPFH tends to be found most frequently in populations in which haemoglobinopathy disorders are common, such as those of African and Greek descent. This is because increased amounts of HbF can be beneficial (as it can lead to a milder disease).

However, rarely when co-inherited with HbS (sickle cell) then this can look very similar to sickle cell anaemia (HbSS) and may require ongoing follow up. *In those couples of whom one or both partners are sickle cell carriers(trait) or have sickle cell disease, screening for HPFH is recommended.*

Testing can be arranged by your local doctor. By testing we can determine whether there is a risk of having a child with a haemoglobinopathy/thalassaemia.

References:

Is elevated fetal haemoglobin a cause for concern in antenatal screening? - PHE Screening (blog.gov.uk)

For Medical Professionals seeking further information or advice, please contact Pathlab to discuss with one of our Haematologists: **07 858 0795**