Hereditary Persistence of Fetal Haemoglobin (HPFH): Information Sheet

Produced by Specialist Haematology, Canterbury Health Laboratories

🩸 What is Haemoglobin?

Haemoglobin (Hb) is a protein found in red blood cells; it carries oxygen from the lungs to all parts of the body and gives blood its red colour. Hb is made up of haem groups, which contain iron and globin chains which are made of protein.

The combination of different globin chains with the haem groups produces the different types of haemoglobin present in people. The normal adult haemoglobin is Hb A.

Where a person has inherited Hb A from both parents, they will have the usual Hb combination, Hb AA.

Foetal Hb (Hb F) is the main Hb produced during the foetal period, but from birth is gradually replaced by the production of adult Hb (Hb A).

🧬 What is Hereditary Persistence of Fetal Haemoglobin (HPFH)?

It is a name used to describe a group of disorders where the production of unusually large amounts of fetal haemoglobin continues into adult life.

Heterozygous hereditary persistence of fetal haemoglobin (HPFH) occurs when a person inherits the normal haemoglobin from one parent and the HPFH from the other parent.

When a person inherits HPFH from both parents, they are said to have homozygous HPFH.

🌎 Who does it affect?

HPFH occurs most frequently among people of southern European, Mediterranean or African origin, but can affect all races.

💊 What is the Treatment?

*HPFH is **not** an illness and requires no regular medical follow-up or treatment.*

HPFH may be mistaken for iron deficiency anaemia and iron tablets may be given unnecessarily.

Iron tablets should only be taken if your doctor has done a special blood test to check that you lack iron.

👨‍👩‍👧 How is HPFH inherited?

HPFH is inherited from either one or both parents who carry the HPFH gene on the DNA in their cells.

If a person is born with HPFH, they will have it all their life. It is **not** contagious or infectious.

How Will This Affect My Children?

Every child inherits haemoglobin genes from each parent. The type of haemoglobin gene inherited from the parent determines the type of haemoglobin the child will have.

If only one parent is a carrier for HPFH there is a 50% chance that any child will also be a carrier for HPFH.

When **two** people with heterozygous HPFH become parents, there is a 25% chance that the child will inherit a HPFH gene from each parent and be homozygous for HPFH.

Why is Knowing About HPFH so Important?

The inheritance of HPFH produces a mild disorder with no requirement for medical follow-up or treatment although sometimes it is difficult to make a definitive diagnosis.

You need to know about it for several reasons:

- Carriers of HPFH can be mistaken for carriers of another haemoglobin disorder called delta-beta thalassaemia
- During pregnancy high levels of haemoglobin F could be mistaken as an indication of a foetal-maternal bleed

Is There a Test for HPFH?

YES ! Blood tests and genetic studies can show whether an individual has HPFH.

Carrying HPFH is **not an illness** and will never cause you health problems. However, documenting your status is highly valuable for the following reasons:

1. **Prevents Unnecessary Testing:** It provides a clear answer for why your fetal hemoglobin levels are high, preventing unnecessary future medical diagnostic tracks.
2. **Clarifies Future Family Planning:** HPFH is passed down genetically. If you are planning a family, it is important to know your status so you can discuss inheritance probabilities with a healthcare professional.

Early diagnosis of a possible HPFH with combination of other haemoglobin variant may ensure and help with family planning.

Can HPFH Be Prevented?

HPFH cannot be prevented at this time, but a programme of health education, testing for the trait, genetic counselling and prenatal diagnosis can provide families with full medical information to help them have healthy children.

People who think they may have HPFH should go to their family doctor for advice and testing.

Your family doctor can contact a Haematologist at the Community Laboratory or the Haematology Department at Canterbury Health Laboratories on (03) 364 0300 if further clinical input is required.

The Specialist Haematology Laboratory at Canterbury Health Laboratories provides a comprehensive diagnostic service in conjunction with the family doctor.

Contact for Canterbury Health Laboratories

Laboratory Services: Specialist Haematology Laboratory, Canterbury Health Laboratories.
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On behalf of the Clinical Haematology Team at Christchurch Hospital