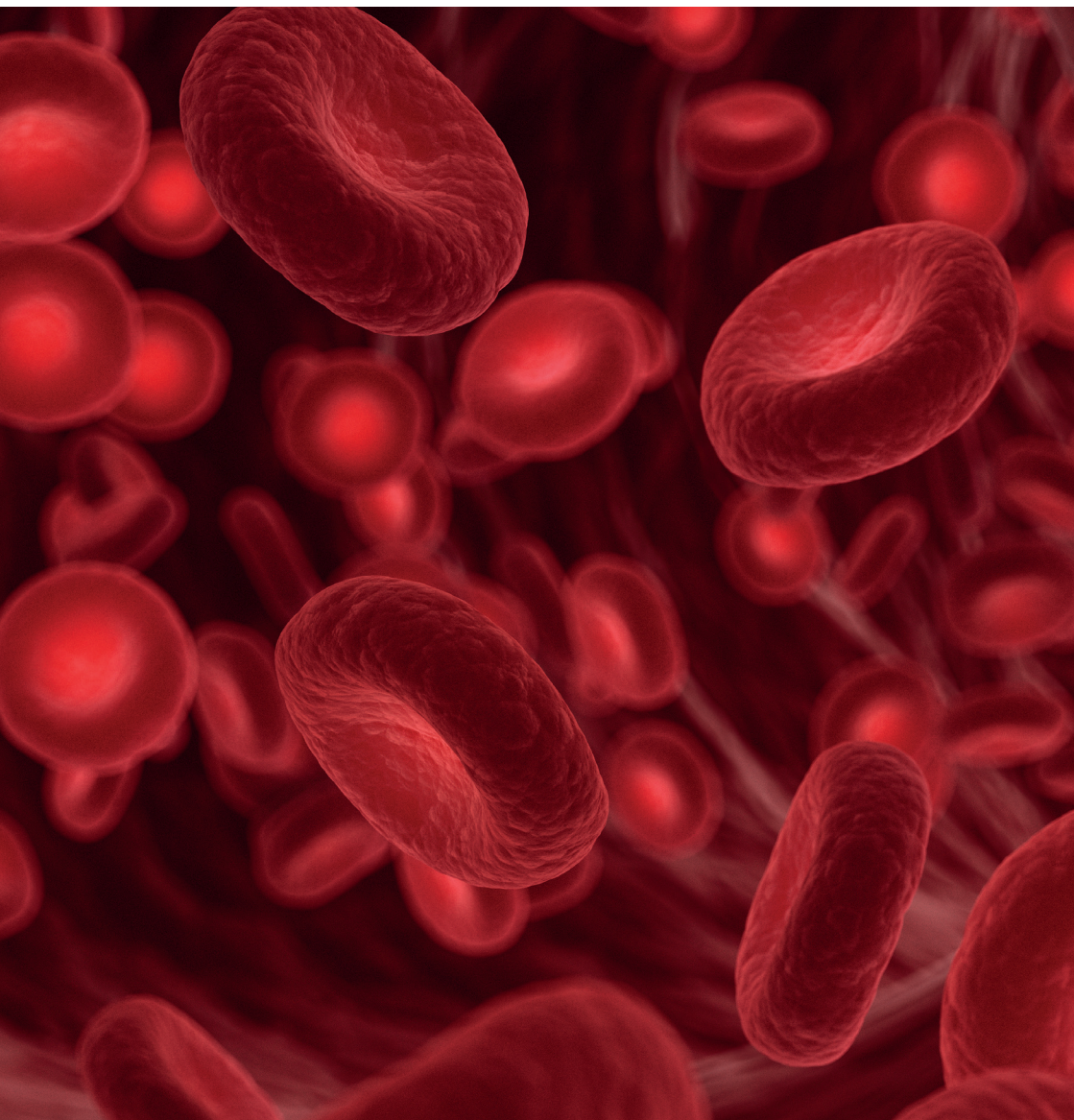


CARRYING AN UNCHARACTERISED HAEMOGLOBIN VARIANT



What does it mean to be a carrier for an uncharacterised haemoglobin variant?

Red blood cells contain haemoglobin.

Haemoglobin is the protein which carries oxygen in your red blood cells. The usual haemoglobin in the red blood cells of an adult person is called haemoglobin A. There are many other types of inherited change in haemoglobin - for example, Haemoglobin S (Sickle cell), haemoglobin C and Beta Thalassaemia. (Haemoglobin is sometimes abbreviated to Hb such as in HbA, HbS, HbD or HbC).

Carrying a variant haemoglobin means that your red blood cells contain both the usual haemoglobin (haemoglobin A) and an unusual type of haemoglobin (variant haemoglobin).

People who carry an uncharacterised haemoglobin variant are as healthy as other people. It is not an illness or infection and cannot be caught like flu.

It is passed on from parents to children like eye colour or the shape of ears. Other people in your family are also likely to carry this uncharacterised haemoglobin variant.



Why is it important to know about carrying a haemoglobin variant?

Haemoglobin is made up of two different parts called alpha and beta.

There are more than 550 beta chain variant haemoglobins. These have names like haemoglobin G-Saint Jose, haemoglobin J-Baltimore, haemoglobin Andrew-Minneapolis, haemoglobin Kansas, and haemoglobin Richmond. The name is often the place where it was first found or the ancestral home of the first person shown to have the particular variant.

There are over 800 alpha chain variant haemoglobins. These have names like haemoglobin J-Oxford, haemoglobin G-Philadelphia, haemoglobin J-Paris, haemoglobin G-Coushatta.

Your variant haemoglobin has not been characterised (or sub-typed) and does not therefore have an additional name yet.

As far as we can tell your unclassified haemoglobin variant is completely harmless. In which case carrying it should never affect your health or the health of your children.

Why was I tested for haemoglobin variants?

One of the following scenarios probably applied.

a) Your blood was "screened" for unusual haemoglobins because some are more common and important for health. The screening test showed you do not carry any of these important haemoglobin variants. However, the test detects harmless haemoglobin variants as well and occasionally ones that have not previously been reported. This can sometimes be confusing for patients and health workers.

b) The usual way to monitor control of diabetes is to measure a haemoglobin component in the red cells of the blood. The presence of this variant haemoglobin interferes with this measurement. The laboratory staff noticed that something was wrong with the measurement and investigated the cause.

c) On the other hand, a family member may have been found to carry this variant and you wanted to know if you had inherited it as well.

Are there are implications for me or my family?

Now that you know that you carry a variant haemoglobin, it is important for you to know that it should do you no harm.

Not all health workers know this, so someone could tell you that you have a health problem when you do not.

If you have brothers or sisters, or if you already have children, they may also carry this variant haemoglobin. There is no need for them to have a blood test to find out.

However, they could find it useful to know about the variant haemoglobin, in case one of them has a test in the future and finds out that he or she is a carrier. Then they would already know that this particular variant haemoglobin is nothing to worry about.

This haemoglobin variant may interfere with HbA1c measurement by High-performance liquid chromat-ography (HPLC) which is a common test used to test for diabetes. If diabetes monitoring is required, an alternative method may need to be used.

How could the health of my children be affected?

When planning a family, it would be helpful to ask your partner to be tested for haemoglobin variants (also called 'haemoglobinopathy' or 'haemoglobin disorder' testing).

If your partner is also a carrier of a haemoglobin variant, there may be a possibility that your child's health could be affected, if they inherited both haemoglobin variants. You can be referred to a specialist genetic counsellor for a discussion.

If your partner is also a carrier, you could have further testing to classify your haemoglobin variant. This would involve another blood test for genetic testing to read through the sequence of the haemoglobin genes.

Once it is known what type of haemoglobin variant you have, you can be assessed to see whether there are any possible implications for children that you have together.

This is best done before pregnancy if possible.

Remember

- ✓ Most carriers of variant haemoglobins are as healthy as other people. Carrying a haemoglobin variant should not weaken you mentally or physically. You are not likely to need any medical treatment because you carry a haemoglobin variant.
- ✓ Carrying a haemoglobin variant should not affect the food you eat or the work you choose to do.
- ✓ If you are planning to have children, encourage your partner to have a blood test for haemoglobin variants. This is best done before pregnancy.
- ✓ If your partner does not carry a haemoglobin variant, then your children will be as healthy as other children, and will not inherit a haemoglobin disorder, although they may be a carrier like you.
- ✓ If your partner does carry a haemoglobin variant you can ask to be referred to the specialist counsellor or Genetics Department to discuss the possible implications for the health of your children.
- ✓ Only certain haemoglobin variants have any effect on the health of children. Another blood test for genetic testing can tell if you have a significant haemoglobin variant.
- ✓ This haemoglobin variant may interfere with HbA1c measurement by High-performance liquid chromatography (HPLC) which is a common test used to test for diabetes. If diabetes monitoring is required, an alternative method may need to be used.
- ✓ Talk to other people in your family about carrying this variant haemoglobin and give them this leaflet to read. The information may be important for their children's health.



GIG
CYMRU
NHS
WALES

Bwrdd Iechyd Prifysgol
Caerdydd a'r Fro
Cardiff and Vale
University Health Board

**For further information please
contact the Genetic Counsellors at:**

All Wales Medical Genomics Service

Wales Genomic Health Centre
Cardiff Edge Business Park,
Longwood Drive, Whitchurch

Cardiff, Wales, CF14 7YU
Tel: 02921 834000 (option 1)

Acknowledgements

With thanks to the following, whose advice and help were invaluable in the production of this leaflet.

Mrs Ceri Amekuedi
Professor Alan Burnett
Dr Tommy Chung
Mrs Farida Haque
Mrs Patience Karseras
Miss Tessa Liburd
Dr Alison May
Dr Philip Connor

Mrs Naeema Noor
Mrs Sarla Patel
Mr Costa Paul
Mrs Judith Shankleman
Professor Robert Shaw
Dr Charles Wardrop
Mrs Paulette Palmer
Dr Indu Thakur

and others whose names have been accidentally omitted.