Blog: Gerri Mortimore

Haemochromatosis: what is it and could you have it?

Genetic haemochromatosis (GH) is the most common inherited genetic disorder among Northern Europeans, thought to affect 1 in 200 people in the UK. But despite this, most people have never heard of the condition. Gerri Mortimore, Lecturer in Post-registration Health Care at the University of Derby, looks at the health implications of this disorder.

Haemochromatosis affects people all over the world but commonly, Northern Europeans, especially those of Celtic descent. According to The Haemochromatosis Society and the British Liver Trust, it is suggested that only 1 in 5,000 people are diagnosed with the condition, making it extremely under diagnosed.

Iron overload and tissue damage

Over time, GH leads to an increased dietary absorption of iron and the excess iron is deposited systemically around the body, leading to iron overload. Multiple organs can be affected, causing inflammation and tissue damage, with the potential to lead to heart disease, diabetes and cirrhosis of the liver.

As the liver is the main storage of iron, liver disease usually occurs before disease in other affected organs. Liver disease can lead to the development of liver cirrhosis – scarring of the liver with associated complications of varices, ascites and primary liver cancer, known as hepatocellular carcinoma.

Why did this genetic disease occur?

Thousands of years ago, it was beneficial to have a higher level of iron to support the body against days or weeks without meat consumption. People survived through the ages due to the fact they had inherited a gene that enabled them to hold onto extra iron.

However, today iron is ubiquitous with iron-rich foods such as meat available from supermarkets and butchers seven days a week. Many foods, including breakfast cereals, also have iron added. If you are unaware that you have this condition, eating iron-rich food could lead to more serious medical problems.

However, not everyone who is diagnosed with GH will go on to develop iron overload and this is thought to be due to genetic factors. GH affects more men than women and this can be explained by the protective mechanism of menstruation in females where there is monthly blood loss.

The symptoms of GH

Unfortunately, the symptoms of GH are non-specific and may include:

- Tiredness or fatigue
- Joint pains, especially in the knuckles of the hand
- Abdominal pain
- Headaches
- Depression
- Skin pigmentation
- Loss of sex drive
- Type 2 diabetes

And more serious clinical signs, indicating advanced liver disease, such as:

- Fibrosis of the liver / cirrhosis
- Enlarged liver
- Cardiomyopathy

Because the symptoms are non-specific, diagnosis of GH can only be made by certain blood tests, in particular transferrin saturation, which indicates how much iron is available to use. If the result is over 50% in men and 45% in women, GH is very likely.

If transferrin saturation is raised, a genetic test called the HFE gene test is offered to determine if you have GH. There are two common mutations of the gene which causes GH, and to develop iron overload, two copies of the mutated gene are required – one from each parent. Having only one copy of the gene, from one parent with GH, makes the person 'a carrier'.

Thousands of people in the UK may be carriers

If it is estimated that one in 200 people have both copies of the gene, then how many thousands of people in the UK are carriers? This may not be of concern, as being a carrier rarely leads to iron overload, but if you have children with a partner who is also a carrier then your children could ~~be homozygous or carriers~~ inherit two copies of the gene and possibly develop iron overload or become a carrier themselves. If a diagnosis of GH is made, a referral to secondary care should be made for further tests and investigations to rule out liver damage.

Find out more

For more information about this condition, contact the Haemochromatosis Society or email Gerri Mortimore at g.mortimore@derby.ac.uk