

Hemochromatosis - FAQs

What is hemochromatosis?

Hereditary hemochromatosis (HHC), or **iron overload**, is an inherited disorder that causes the body to absorb two to three times the normal amount of iron. Over the years, the excess iron builds up in the vital organs, joints and tissues where it **can cause a number of debilitating and potentially fatal conditions** such as liver and heart disease, diabetes, impotence and arthritis.

How common is it?

Hereditary hemochromatosis is **the most common genetic disorder affecting Canadians**. One in nine people of mostly Northern European descent are carriers of the most common type of HHC and 1 in 300 are estimated to have two copies of the gene that puts them at risk for iron overload. Up until recently, medical professionals were taught this was an extremely rare disorder, so **your doctor may not be aware of it**.

What are the symptoms?

Because early symptoms can be **non-specific and attributed to many other causes**, hemochromatosis can be **difficult to diagnose** until more extensive damage has occurred. Even then, hemochromatosis is often overlooked as the causative factor in an observable disease.

Some early symptoms include:

- chronic fatigue
- depression
- abdominal pain
- aching joints, especially in the knuckle and first joint of the first and second fingers
- loss of sex drive for both sexes, impotence for men, menstrual irregularities for women or early menopause
- discolouration or bronzing of the skin

Later symptoms may include:

- Type II Diabetes (adult onset)
- hypothyroidism
- disease of the heart muscle and arrhythmia
- liver disorders including cirrhosis and liver cancer

Is there a special test for hemochromatosis?

Yes. A simple blood test can detect iron overload, but it is **NOT part of the standard blood test** ordered in conjunction with an annual check-up. Your doctor must specifically request an **iron series profile** on the lab requisition form. It

measures the amount of iron in the various iron storage and transport proteins in your blood to give a picture of how much iron is stored in your body. This is NOT the same as hemoglobin. If iron levels are elevated, DNA or genetic testing for the most common type of HHC is available and may be used to confirm a diagnosis. If your iron levels are significantly elevated, your physician may also wish to do additional testing to establish the level of damage on the liver.

Is it treatable?

Yes. Currently, the primary treatment consists of the **removal of blood** (like a blood donation, only more frequent) at regular intervals until iron levels return to normal. Once normal iron levels are re-established, they can be maintained by periodic blood removal (2 to 6 times a year depending on the individual). During this maintenance phase, if eligible, blood can be donated through the Canadian Blood Services (CBS). **Treatment will prevent damage that would have been caused by iron overload, and is ongoing for life.**

What happens if it isn't treated?

Not everyone with both copies of the defective gene develops hemochromatosis and, if they do, the degree of iron overload varies. However, once a person exhibits symptoms of iron overload, the disease process can only be arrested by removal of the underlying cause, which is too much iron. Left too long, many symptoms such as cirrhosis of the liver, heart disease, and diabetes will be irreversible and **can lead to early death**. Treated before damage occurs, **disease need never occur**.

Should my entire family be screened?

HHC is a genetic disorder, so all of your **first degree relatives should also be tested** for elevated iron levels. Parents, siblings and adult children may want to confirm their genetic status to evaluate their risk of developing iron overload if their initial blood tests reveal normal iron levels. Spouses should be tested to help evaluate the potential risk to minor children.

How can the CHS help me?

The Canadian Hemochromatosis Society was established to create awareness about this little-known disorder so that early diagnosis would become the rule rather than the exception, and so that needless suffering and premature death from undiagnosed hemochromatosis would become a thing of the past. In the meantime, we are here to provide **information and support** to those adversely affected by iron overload.

How can I help the CHS?

- help us **spread the word** - ask your physician about hemochromatosis
- tell your family and friends
- become a member and / or a volunteer of the Society
- donate to the CHS so that we can continue to help you and others

Unanswered Questions?

If you've consulted our FAQs and our expanded information on our web site at www.toomuchiron.ca and still have questions regarding hemochromatosis, you can ask an expert by e-mailing askanexpert@toomuchiron.ca.

You can also contact our office toll-free in Canada at 1-877-BAD-IRON (1-877-223-4766) or by e-mail at office@toomuchiron.ca.