

***HFE*-Related Hereditary Hemochromatosis: Patient Information**

HEREDITARY HEMOCHROMATOSIS (HHC) is a condition where adults develop iron overload. It rarely affects children. People with HHC may experience fatigue, joint pain in the hands, bronzing in the skin, heart disease, low sex drive for men, diabetes, and liver disease. Phlebotomy, or blood-letting, can stop the disease from progressing. Most people with this disease who receive treatment can live a relatively normal life.

IF YOUR DOCTOR WANTS TO CHECK YOU FOR HHC, he or she may check your “ferritin,” which represents the iron stored in your heart and liver. Your doctor should also check your “transferrin saturation,” which represents how much iron is being carried in your blood. People with HHC have high levels of *both* ferritin and transferrin saturation. Having a high ferritin *only* is usually not a sign of HHC. Your doctor might look into other causes for high ferritin if that is the case.

IF YOUR DOCTOR THINKS YOU DO HAVE HHC based on your ferritin and transferrin saturation, he or she may order another blood test to look at your genes. The most common form of HHC is caused by changes in the *HFE* gene. Most individuals with HHC have two changes in the *HFE* gene. However, the reverse is not always true. In fact, many people with changes in the *HFE* gene do not actually go on to develop iron overload. The genetic test result might help your doctor confirm whether you do have HHC, or whether your family is at risk to develop HHC. Genetic testing does come with some benefits and risks and you may want to ask your doctor more questions about this test.

YOUR DOCTOR MIGHT ALSO REFER YOU to different specialties for their input. For instance, you may be referred to a liver doctor if he or she is worried about your liver. If you need to have phlebotomies, your doctor might ask you to meet with a hematologist. You may also be referred to a genetics clinic to help explain the chance that other family members might have a similar disease and whether they need to get checked, too.

PEOPLE WHO HAVE HHC should be careful about their lifestyle. They should avoid taking multivitamins that contain iron or vitamin C. They may also eat less red meat. Because they have an increased chance for liver disease, they should avoid drinking excessive alcohol. They should also avoid eating raw shellfish. Finally, some people with HHC will donate blood with Canadian Blood Services even if they no longer need phlebotomies.

This Information is Provided By:
Genetics & Metabolism Program,
Shared Health Phone: (204) 787-2494