What Is Hereditary Hemochromatosis?

Hereditary hemochromatosis (HH) is a genetic condition that may lead to buildup of iron in tissues throughout the body.

Patients with HH inherit a gene variant from each biological parent that affects the production or function of hepcidin, a protein that regulates use of iron in the body.

Who Is Most Likely to Have HH?
Hereditary hemochromatosis is most common in individuals of northern European ancestry. In this group, approximately 1 in 300 people have HH. Signs and symptoms of iron overload typically develop at a younger age in males than in females, likely because of the release of iron during menstruation.

Which Organs Does HH Affect?
Hereditary hemochromatosis affects the liver in about 25% of patients and may cause elevated liver function values, an enlarged liver, or both. Up to 9% of people with untreated HH develop end-stage liver disease (cirrhosis), and these people are at increased risk of liver cancer (hepatocellular carcinoma). About 1% to 3% of individuals with HH develop a heart condition (cardiomyopathy), which may lead to heart rhythm abnormalities, heart failure, or both. Iron deposits in the pancreas, pituitary gland in the brain, and joints can cause diabetes, decreased production of sex hormones (hypogonadism), and arthritis.

What Are the Signs and Symptoms of HH?
Hereditary hemochromatosis may cause fatigue, joint pain, abdominal pain, loss of sex drive, and bronze- or gray-colored skin in advanced stages of disease. However, approximately 90% of patients with this genetic disorder have no signs or symptoms of HH.

Testing for and Diagnosis of HH
Hereditary hemochromatosis is typically diagnosed if blood testing reveals iron overload, based on an elevated serum transferrin saturation (TSAT) level, increased ferritin level, or both. Genetic testing for variants that cause HH confirms the diagnosis.

People with signs or symptoms suggestive of HH or with certain medical conditions (such as cirrhosis or cardiomyopathy of unclear cause) should undergo testing of their TSAT and ferritin levels. Individuals who have a parent, sibling, or child with HH are also recommended to get these blood tests and to undergo genetic testing for HH, even if they are not diagnosed with iron overload.

Treatment and Lifestyle Modifications for Patients With HH
First-line treatment for patients with HH is regular blood removal (phlebotomy), which decreases blood iron levels, improves liver function, decreases fatigue, and reduces risk of liver and heart problems. Phlebotomy is typically performed weekly for several years until the blood ferritin level is low, at which point it can be decreased to 3 to 4 times per year. Patients who do not improve with phlebotomy or are not able to tolerate phlebotomy (because of severe anemia or heart failure, for example) may receive an oral drug that binds iron, although evidence for this treatment is limited. Patients with HH and end-stage liver disease or hepatocellular carcinoma should be evaluated for liver transplant.

Individuals with HH should avoid taking supplements with iron and vitamin C, but an iron-restricted (vegetarian) diet is not necessary. Patients with HH should maintain a healthy body weight and limit or avoid alcohol intake because being overweight and drinking alcohol increase the risk of cirrhosis and hepatocellular carcinoma.

FOR MORE INFORMATION
Centers for Disease Control and Prevention