Understanding Hemochromatosis

Hemochromatosis is a disorder in which the body absorbs too much iron. Usually inherited, it can also be caused by repeated blood transfusions, sickle cell anemia, liver disease or alcoholism.

Healthy people absorb about 8 to 10 percent of the iron they consume through diet. If you have hemochromatosis, you can absorb four times more iron than normal; over time, you may retain 5 to 20 times the amount of iron the body needs. Untreated, excess iron can accumulate in the liver, heart, pancreas, thyroid and other organs, leading to liver disease, diabetes, heart attack, hypothyroidism and other serious or life-threatening medical conditions.

Symptoms
The most common complaint is joint pain. Other symptoms include:
- fatigue/lack of energy
- abdominal pain
- unexplained weight loss or weight gain
- loss of sex drive
- irritability
- bronze or yellowish skin color

Sometimes there are no symptoms; a routine physical exam or blood test reveals the condition. Because many medical conditions can cause similar symptoms, it is important to see your doctor for further evaluation.

Genetics
Hereditary hemochromatosis is most often caused by abnormalities in the gene HFE, which helps control the amount of iron absorbed from food. Everyone inherits two copies of HFE, one from each parent. Two specific mutations of this gene account for the vast majority of iron overload: C282Y and H63D. (There are also other, less common genetic causes.)

If you inherit one mutated (altered) copy, you are a carrier, or heterozygote. You will not have hemochromatosis, but you could pass it on to your children.

If you inherit two copies of the same mutations, you are a homozygote. If you inherit two different mutations, you are a compound heterozygote. The exact mutations determine your likelihood of risk for iron overload. Only 1 in 400 people is likely to have genetic mutations in both copies of the HFE gene.

Diagnosis
Hemochromatosis is usually discovered through a routine blood test. Specific tests may cover these areas:
- Transferrin saturation (TS): The best measure of excess iron storage, this blood test measures how much iron is bound to the protein that carries iron in the blood.
- Ferritin: Levels of this protein in the blood indicate how much iron is stored in the liver and the chance of liver damage.
- Liver biopsy: This allows a physician to see how much iron is accumulating and whether there is inflammation or cirrhosis (scarring).
- Genetic testing: This is best used after diagnosis to understand why someone has hemochromatosis and to determine which, if any, genetic mutations caused it. It is good to remember that not all genetic mutations can be tested for, nor does their presence mean that you will experience hemochromatosis.

Treatment
Treatment aims to remove excess iron from the blood and treat organ damage.

Periodic phlebotomy (therapeutic blood removal) is similar to regular blood donation but requires a doctor’s order. You may initially need to donate as often as once or twice a week, then less often (but still regularly) as iron levels drop to normal. It is safe to donate to blood banks and hospitals.

If you have hereditary hemochromatosis, but have not experienced organ damage, phlebotomy can improve issues, like fatigue and abdominal pain—but other issues (joint pain and diabetes) will likely be unaffected by treatment.

Recommended dietary changes include consuming less iron and avoiding iron and vitamin C supplements (vitamin C enhances iron absorption). If you have liver damage, your doctor may advise you to avoid alcohol and raw seafood.

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