He[illegible]emochromatosis

Reviewed by: Christopher N. Frantz, MD

Listen Listen | AAA |

Hereditary hemochromatosis (hee-muh-kro-muh-TOE-sus) is a genetic disease that causes the body to absorb and store too much iron.

Iron is an important nutrient that our bodies get from food. It's in every red blood cell and is the main component of hemoglobin, the substance in red blood cells that carries oxygen away from the lungs to the rest of the body.

About Hemochromatosis

In hemochromatosis, the body absorbs about twice as much iron as it should. This excess iron can't leave the body. Instead, it's stored in the joints and major organs such as the liver, heart, brain, pancreas, and lungs. Over time, this iron can build to toxic levels that can cause diabetes and damage or even destroy an organ.

Even though kids are born with hemochromatosis, the very slow buildup of iron means that it might not be diagnosed until adulthood, when symptoms begin.

Therapies and diet changes can help slow the progression of the disease. That's why it's especially important for kids with a family history of hemochromatosis to be tested early on.

Causes

Hereditary hemochromatosis is caused by a mutation in a gene that controls how much iron the body absorbs from food. It's actually fairly common, affecting about 1 in 200 people. The disease is considered "autosomal recessive," which means that someone must have two copies of the mutated gene — one from each parent — to get it.

In the United States, 1 out of every 8 to 10 people has one copy of the mutated gene. These people are **carriers** — they can pass the gene on to their children but won't get the disease. Many people who inherit two mutated genes will absorb extra iron, but only some will absorb enough to cause health problems.

Carriers and other people who don't have hereditary hemochromatosis can still have iron build up if they have another genetic defect or a health problem that affects iron absorption, such as alcohol abuse or hepatitis, an inflammation of the liver.

Signs and Symptoms

Some people with hemochromatosis never develop symptoms. Kids who are diagnosed with it rarely have symptoms because iron takes years to build up.

An adult with the disease eventually may have:

- muscle aches and joint pain, mostly in the fingers, knees, hips, and ankles
- impotence and hypogonadism (low production of sex hormones by the testicles or ovaries)
- gallbladder disease
- cirrhosis (disease and scarring of the liver)
- ongoing fatigue

MORE ON THIS TOPIC

Parents | Kids

Hepatitis

Blood Test: Ferritin (Iron)

Your Child's Immunizations: Hepatitis A Vaccine (HepA)

Your Child's Immunizations: Hepatitis B Vaccine (HepB)

Iron

View more

- depression, disorientation, or memory problems
- stomach swelling, abdominal pain, diarrhea, or nausea
- loss of body hair other than that on the scalp
- early menopause
- gray or bronze skin similar to a suntan
- heart problems
- diabetes
- an enlarged liver (called hepatomegaly)
- a higher likelihood of bacterial infections
- organ failure

Fortunately, not all adults with too much iron in the system develop these conditions.

Sometimes, people with hemochromatosis are diagnosed with conditions that can have similar symptoms, like chronic hepatitis, some forms of diabetes, Alzheimer's disease, iron deficiency, or menstrual problems.

Diagnosis

If hemochromatosis is diagnosed and treated early enough, damage from iron buildup can be prevented. Doctors usually diagnose iron overload with these blood tests:

- serum ferritin: measures the amount of ferritin, a protein that contains iron
- serum iron: measures iron concentrations in the blood
- total iron-binding capacity (TIBC): measures the amount of iron that the blood can carry

Genetic testing for the gene mutation that causes the disease will find most, but not all, cases of hemochromatosis. Sometimes doctors need to do a biopsy, taking a small sample of liver tissue to look for iron deposits or changes in the cells.

If you have a family history of hereditary hemochromatosis, tell your child's doctor. The doctor may recommend checking iron levels or doing genetic testing, especially if a close relative (like a spouse, parent, child, or sibling) has the disease.

Treatment

Doctors treat the iron overload from hereditary hemochromatosis by regularly drawing blood to lower the level of iron. This process, called phlebotomy (flih-BOT-uh-mee), is similar to making blood donations.

During the initial "de-ironing" phase, blood is removed once or twice a week until a person's iron levels drop to normal. This phase may take 2 to 3 years. After that, a person may need phlebotomy done three or four times a year to make sure iron levels stay down. Iron levels then have to be checked about once a year, and treatment continues for life. The goal is to prevent any organ damage from iron overload.

Caring for Your Child

Treatment for kids usually isn't as aggressive as for adults. Minor changes in diet often can help slow iron buildup.

Your child's doctor will recommend ways to delay or lower iron overload. These might include:

- **Drinking tea and milk products.** Black, green, and oolong teas have tannins, which help reduce iron absorption. (Herbal teas won't help because they don't contain tannins.) Milk also reduces iron absorption.

- **Getting vaccinated.** Your child should be immunized against hepatitis A and B.

- **Limiting red meat.** Iron-rich vegetables are fine because the body doesn't absorb iron from plant sources very well.

- **Limiting iron-enriched foods.** Avoid breakfast cereals, breads, and snacks that

are enriched with iron.

- **Limiting vitamin C.** Try to keep any vitamin C supplements under 100 milligrams a day because vitamin C makes the body absorb more iron.

- **Using iron-free vitamins.** Check the label to find a children's multivitamin that doesn't have iron.

- **Not cooking with uncoated cast iron cookware.** It may be possible for iron from the pan to get into the food you cook, although doctor's aren't entirely sure about this.

- **Skipping raw seafood.** Sometimes raw shellfish can carry bacteria that might harm someone who has an iron overload.

- **Not drinking alcohol.** Alcohol is bad for hemochromatosis because it harms the liver — which is already damaged — and also makes the body absorb more iron.

Reviewed by: Christopher N. Frantz, MD
Date reviewed: September 2014