Iron overload can lead to diabetes, heart failure

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Hemochromatosis is probably the most common disease you've never heard of, as well as the single most common genetic disorder in the American population.

An estimated one in 10 Americans carry the genetic trait for hemochromatosis, and about one in 250 are considered at highest risk because they carry the genetic trait from both parents.

But it is grossly under-recognized and grossly under-treated.

What are the symptoms of Hemochromatosis?

Characterized by iron overload in the blood, hemochromatosis could be the culprit for such otherwise unexplained symptoms as fatigue, joint pain, and infertility issues. Over time, the toxic effects of the excess iron can lead to damaging diseases like diabetes, congestive heart failure, and endocrine system problems.

How is hemochromatosis diagnosed?

A simple blood test to detect iron levels followed by a genetic test can be used to detect the disease. Recognizing the possibility of hemochromatosis at the earliest point possible is important for preventing irreversible complications of the disease.

Hemochromatosis is easy to treat but low on the list of suspected diseases when patients have vague symptoms. Because early signs of the disease mimic numerous other disorders, some physicians are pursuing more frequent screenings for hemochromatosis, especially among family members of patients diagnosed with the disease.

If the disease can be caught before people manifest complications from hemochromatosis, they can be treated with very low-tech, cheap and safe methods, preventing complications and allowing them to lead a perfectly normal life.

How is hemochromatosis treated?

Newly diagnosed patients receive regularly-scheduled phlebotomy (blood-letting) to eliminate excess iron in the body. Once their iron levels reach a normal level, patients are put on a maintenance schedule, receiving phlebotomies every two to three months.

If treatment is started before organ damage occurs, people with hemochromatosis can be symptom-free and essentially "cured" as long as their iron levels remain within a normal range.
What specialist treats hemochromatosis?

Hemochromatosis is usually treated by a patient’s primary physician or a hematologist. A primary physician usually makes the initial diagnosis based on iron-panel testing.

Also, confirmatory genetic testing can be ordered. It is helpful for a genetic counselor or geneticist to assist in interpreting the genetic testing results and providing genetic counseling and risk assessment for the patient and other family members.

What does hemochromatosis genetic testing involve?

Typically, genetic testing for hereditary hemochromatosis includes a simple blood test for the two most common mutations in the hemochromatosis (HFE) gene. These are named C282Y and H63D.

Most laboratories that offer this testing charge a couple hundred dollars, and it may not be covered by your insurance company.

If you have two mutations, then you are predisposed to the development of “iron overload” or hemochromatosis. If one mutation is identified, there are two possibilities:

• You are only a carrier of hemochromatosis.

• You have a second, rare mutation that predisposes to hemochromatosis that was not identified on the genetic test. In either scenario, your doctor should use your iron levels to determine whether you should be treated.

Why are men more likely to be diagnosed with hemochromatosis than women?

Although men and women can inherit the HFE gene, men tend to develop the problems caused by iron overload at an earlier age than women. Most women with hemochromatosis don’t display symptoms until after menopause, since they are releasing blood through their normal monthly menstrual cycle.