

Hemochromatosis Iron Overload FACT Sheet

What is hemochromatosis?

- Hemochromatosis, the most common form of iron overload disease, is an inherited disorder that causes the body to absorb and store too much iron. The extra iron builds up in organs such as the heart, pancreas, liver, joints, and thyroid damaging these organs. Without treatment these organs become diseased and eventually fail.
- Healthy people absorb about 10% of iron they ingest. People with hemochromatosis absorb as much as 4 times more iron than normal. Iron cannot be excreted, except through blood loss; therefore, over time iron accumulates in vital organs such as the heart, liver, joints, pituitary and pancreas.
- Hemochromatosis is not a blood disease. The amount of iron in a unit of blood from a person with hemochromatosis is the same as a unit of blood from a person who does NOT have hemochromatosis.
- Classic or Type I hemochromatosis is caused by mutations of the gene *HFE*. The two common mutations are named C282Y and H63D. Other causes of iron overload such as juvenile or neonatal hemochromatosis are not *HFE* related.

What are the signs?

- | | |
|---|---|
| <input type="checkbox"/> Chronic fatigue | <input type="checkbox"/> Weight change |
| <input type="checkbox"/> Joint pain | <input type="checkbox"/> Loss of libido (sex drive) |
| <input type="checkbox"/> Heart arrhythmia, or heart failure | <input type="checkbox"/> Loss of period |
| <input type="checkbox"/> Diabetes | <input type="checkbox"/> Depression |
| <input type="checkbox"/> Abdominal pain | <input type="checkbox"/> Hair loss |
| <input type="checkbox"/> Skin discoloration (bronze, red or ashen-gray/green) | <input type="checkbox"/> Infertility |
| <input type="checkbox"/> Elevated liver enzymes | <input type="checkbox"/> Impotence |
| <input type="checkbox"/> Cirrhosis | <input type="checkbox"/> Stomach problems |
| | <input type="checkbox"/> Headache |

IRON OVERLOAD

Who is most at risk?

Iron overload can occur in anyone, but the type of iron overload caused by classic hemochromatosis (*HFE* related) is more likely:

The gene for hereditary hemochromatosis

IF your ancestors are from Northern Europe

Scotland	Ireland
Germany	Spain
England	France
Italy	Sweden
Scandinavia	
The Netherlands	

IF you are a

Male

or a

Female who is no longer menstruating

IF you have a family history of

heart trouble, especially early death by heart attack* or a history of diabetes, liver disease, arthritis, hormone imbalances, especially hypothyroidism, or infertility.

*casual data only

IF you are homozygous for the C282Y mutation of the *HFE* gene. Or if you are a compound heterozygote for C282Y/H63D mutations of *HFE*.

The two major mutations of *HFE* are C282Y and H63D. Everyone inherits two copies of the *HFE* gene, one from each parent. A person who inherits two mutated copies of *HFE* is called a homozygote. A person who inherits one mutated copy is called a heterozygote (carrier). A person who inherits two different mutations is called a compound heterozygote.

How is HHC detected?

Classical hemochromatosis can be determined with genetic testing. Iron overload however, is determined with blood tests: fasting serum iron, total iron binding capacity and serum ferritin. See charts for normal ranges.

Tests

- transferrin-iron saturation percentage*
- serum ferritin

* Transferrin/iron saturation percentage is serum iron divided by TIBC (total iron-binding capacity) X 100%

Transferrin iron saturation percentage
NORMAL RANGE 25-35%

These tests should be done fasting: Nothing by mouth after midnight except for prescription medications or water.

**In classical hemochromatosis (*HFE* related), both the serum ferritin and the TS% will be elevated if iron overload is present. In some conditions of non-classical iron overload (NASH for example), the serum ferritin might be elevated while the TS% remains normal.

ferritin	Adult Males	Adult Females
Normal Range	up to 300ng/mL	up to 200ng/mL
In treatment*	below 100ng/mL	below 100ng/mL
Ideal	25-75ng/mL	25-75ng/mL
Post Menopausal		25-75ng/mL
Adolescents, Juveniles, Infants & Newborns of normal height and weight for their age and gender		
Male ages 10-19	23-70ng/mL	Infants 7-12 months 60-80ng/mL
Female ages 10-19	6-40ng/mL	Newborn 1-6 months 6-410ng/mL
Children ages 6-9	10-55ng/mL	Newborn 1-30 days 6-400ng/mL
Children ages 1-5	6-24ng/mL	

*phlebotomy treatment

What is the treatment?

De-ironing with therapeutic phlebotomy and diet restrictions is the therapy for iron overload. Therapeutic phlebotomy is the same as routine blood donation except that more frequent blood removal may be needed which requires a physician's prescription. Depending upon the iron levels, phlebotomy may be done as frequently as twice a week so long as the pretreatment hemoglobin remains at 12.5g/dL to avoid overbleeding and unnecessary anemia.

See Common Questions and Answers on the other side.

Commonly Asked Questions and Answers About Hemochromatosis

Do I need a liver biopsy to diagnose hemochromatosis?

Liver biopsy is an important diagnostic procedure; it remains one of the best ways to determine liver damage, such as cirrhosis. This procedure, however, is no longer used to diagnose classic hemochromatosis. Liver biopsy is used to diagnose or document iron levels in non-classical hemochromatosis.

Is the HFE genetic test the best way to diagnose HHC?

Genetic testing is one way to diagnose classic hemochromatosis. Classic HHC is HFE related. Iron Disorders Institute does not recommend using the genetic test for screening purposes or on persons younger than age 19. Appropriate use of genetic testing for Classic HHC is to confirm diagnosis in adults or for couples who are planning a family to determine carrier status.

How is the genetic test done?

A tissue sample can be obtained by taking blood or by doing a cheek swab. Both ways are reliable. About 15% of those with iron overload do not have mutations of HFE. Genetic testing does not provide information about tissue iron levels.

Are my children at risk for iron overload?

HFE related iron overload is not known to occur in youths. The iron accumulation process in classic HHC is very slow and takes 20-30 years to manifest in symptoms or organ damage. Very young children have a naturally high iron saturation, which can be misleading and inappropriately attributed to iron loading.

How do I know if I have iron overload?

Iron levels can be determined by measuring serum ferritin and transferrin-iron saturation percentage (TS%). Both levels will be elevated in adults with classic hemochromatosis. Note that these levels are naturally elevated in newborns, infants and possibly in adolescents.

What if only one level is elevated?

If only serum ferritin is elevated, you may have some other condition present such as hyperferritinemia-cataract syndrome or inflammation due to chronic disease. Phlebotomy, which is the treatment for iron overload may not be appropriate and could even be harmful. If only TS% is elevated, you could be in the first stages of loading iron, dehydrated or possibly have another condition such as abnormal blood glucose. Your physician can determine if any of these conditions are present and whether phlebotomy is warranted.

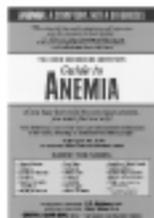
If I have iron overload, how often should I have a phlebotomy?

Patients with serum ferritin over 1,000ng/mL need aggressive iron removal and may need a phlebotomy twice a week until ferritin is lowered to a normal range. Once ferritin is below 1,000ng/mL, the risk of cirrhosis is less than 1%. Iron loading patterns vary with the individual depending upon a person's age, compliance with therapy, general health, tissue iron levels, modifying genes not yet discovered, tolerance to blood extractions, and diet, especially the amount of red meat consumed. Ferritin levels need to be checked periodically to assure iron levels are dropping. Serum ferritin drops at an estimated 30 ng/mL for each full unit of blood removed. Once de-ironed phlebotomies might be as infrequent as 3 or 4 times a year depending upon the patient.

Why is my blood thrown away?

At centers that do not have an FDA variance to use HHC blood for transfusion the blood must be discarded. However, centers that have the variance can take HHC blood every two weeks with a doctor's order and they can use the blood for transfusional purposes. There are many centers with variances in the US; contact Iron Disorders Institute to see if there is one in your area. One of the most prestigious in the US is the Hemochromatosis Protocol at the Warren G. Magnuson Clinical Center in Bethesda, MD. If you live within driving distance of Bethesda, and you have HHC, you might be eligible to participate in this unique program. Contact us for details.

Important! Do not get overbled. Pretreatment hemoglobin should be 12.5g/dL. For patients who have been overbled and who are having difficulty with anemia, you might like to read our book: *Iron Disorders Institute Guide to Anemia*.



Read more about hemochromatosis in Iron Disorders Institute *Guide to Hemochromatosis* Available through major bookstores or online at www.IronDisorders.org

Is there anything I can do to help prevent chronic disease caused by iron overload?

Early detection is the best way to reduce your risk of disease from iron overload/hemochromatosis. However, there are preventive measures you can take to help lower your risk of disease; here are some important ones:



Donate blood: One blood donation a year can lower a male's risk of heart attack by 50%

Don't smoke: Tobacco is loaded with iron and when inhaled, this iron bypasses the defense system the body uses to control the amount of iron you absorb.

Cut back on alcohol or stop altogether: Alcohol increases the absorption of iron and damages the liver.

Cook in glass or ceramic cookware: Iron filings can get into food from cast iron skillets and some grills.

Cut back on red meat: Red meat such as beef, lamb and venison contain high amounts of heme iron, which is the type of iron most easily absorbed by the body. Chicken, fish and pork have less heme iron.

Eat more fruits, vegetables, nuts and grains: These foods contain nonheme iron, which is the type of iron that is not so easily absorbed by the body.

Avoid Vitamin C at mealtime: Ascorbic acid increases the absorption of iron.

Consume foods high in calcium: Calcium inhibits the absorption of both heme and nonheme iron.

Drink tea, coffee, or dairy with the main meal: These inhibit the absorption of iron.

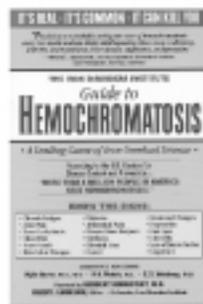
Do not eat raw shellfish, which might contain a bacteria that is deadly to people with high iron levels.

Keep good records with IDI's Personal Health Profile and Read up on diet and other important iron matters in our books and our magazine *idInsight*.

Patient support materials are available through Iron Disorders Institute:

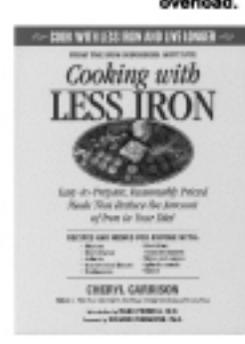


The Personal Health Profile booklet, perfect for record keeping.



Our book about HHC, which is written especially for the patient and their family.

Our unique magazine devoted to iron related health issues.



And the companion cookbook for people with iron overload.