

# Hemochromatosis FACT Sheet

## What is hemochromatosis (HHC)?

- Hemochromatosis is an inherited disorder that causes the body to absorb extra iron. Over time, the extra iron builds to toxic levels in vital organs such as the heart, pancreas, liver, joints, and pituitary. These organs cannot function normally because of iron overload. Without treatment to lower the iron to healthy levels, 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. Excess iron is removed with blood donation and in some rare or difficult cases, excess iron is removed with medications.
- 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 hemochromatosis such as juvenile or neonatal hemochromatosis or African siderosis are not HFE related.

## symptoms & 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"/> Elevated serum iron, TS%* or serum ferritin |

## HEMOCHROMATOSIS

\*TS%=transferrin-iron saturation percentage

## Who is most at risk?

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, osteoarthritis, hormone imbalances, especially hypothyroidism, or infertility.**

**IF** you are homozygous for the C282Y mutation of the HFE gene. Also at risk are H63D homozygotes, compound heterozygote (C282Y/H63D) and some carriers.

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 levels are 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%

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

**Transferrin iron saturation percentage**  
**NORMAL RANGE 25-35%**

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

\*\*In classical hemochromatosis the TS% rises first, then the serum ferritin. Generally both will be elevated if excess levels of iron is present. In some conditions of non-classical iron overload (NASH for example), the serum ferritin might be elevated while the TS% remains normal.

## What is the treatment?

Iron reduction is achieved with therapeutic phlebotomy and diet modification. 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 performed as often 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 of 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 hemochromatosis?

The iron accumulation process in classic HHC is very slow and can take decades before symptoms or organ damage occur. *Very young children have a naturally high iron saturation and serum ferritin. Children also can have an elevated ALP (alkaline phosphatase) a liver enzyme. This can be a normal event for children going through bone growth, but can be misleading and inappropriately attributed to iron loading.*

## Can I drink alcohol if I have hemochromatosis?

Alcohol increases iron absorption. A person with hemochromatosis must be very careful about the use of alcohol because the combination of elevated iron and alcohol are potentially lethal to the liver. For most patients if the liver is healthy and the iron levels normal moderate use of alcohol is permitted. Moderate use is defined as: 8oz wine; 1 oz of spirits; 12oz beer per day.

## 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. When initial 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.

## IMPORTANT POINTS

### #1: Hemochromatosis requires a life-long commitment to iron balance.

Once your iron levels are normal, this does not mean you are "cured". Establish a schedule for blood donation (or therapy recommended by your physician) that suits your unique loading pattern. Get check-ups once a year or at least every two years to assure iron levels continue to be normal.

### #2: Do not get "overbled".

Pretreatment hemoglobin should be 12.5g/dL for most people. Individuals who are overbled are at risk of becoming "iron avid". Read about iron avidity on our website.

Download our newsletter



Education is the cure for Iron-Out-of-Balance™  
**nanograms**  
Fourth Quarter 2009  
www.irondisorders.org

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- Iron in Research: A Tool or Weapon?
- How New Diet Trends May Increase Risk for Iron Deficiency in Kids
- Hypogonadism: The Physical, Mental and Social Impact

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- Serum Ferritin: What Makes it Rise and Fall?
- Hemochromatosis Blood Donor Programs in the USA
- The Other Mutation: H63D
- Iron: Is it a Nutrient, a Medicine or Both?

The Personal Health Profile booklet, perfect for record keeping.



## What can I do to prevent chronic disease caused by hemochromatosis?

Early detection is the best way to reduce your risk of disease from 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.

**Limit supplemental Vitamin C :** Ascorbic acid increases the absorption of iron.

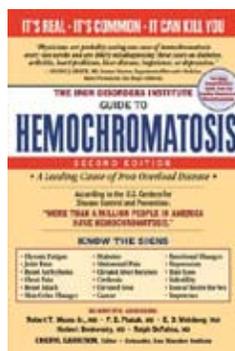
**Consume foods high in calcium:** Calcium inhibits the absorption of both heme and nonheme iron.

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

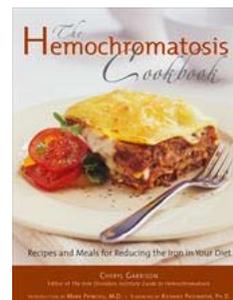
**Do not eat raw shellfish,** which might contain a bacteria that is deadly to people with high iron levels.

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

## Patient support materials are available through Iron Disorders Institute:



Our book about hemochromatosis is written especially for the patient and their family.



And the companion cookbook helps with iron balance.

Learn more about hemochromatosis

Visit our websites  
[www.hemochromatosis.org](http://www.hemochromatosis.org)  
[www.irondisorders.org](http://www.irondisorders.org)