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This website is dedicated to the millions of thyroid patients who are being ignored and left to suffer unnecessarily, and to healthcare practitioners, who want to better serve those patients.

MENU

Haemachromatosis/iron overload FAQ's

Please note: This information has been compiled with the advice of leading doctors/researchers as well as HH patients themselves. This information is based on the following premise that this is the information we would give to a family member, where "money was no object", and the latest information on health options was wished. Through this premise, the most thorough and aggressive health care can be suggested. Each patient should confer with his/her physician about their own health care. If a physician does not regularly treat HH patients, he/she should consult with a medical expert. AHS can provide such experts.
Sandra Thomas, President, American Hemochromatosis Society

Q: What is iron overload, hemochromatosis?

A: Hemochromatosis (pronounced: He-mo-chro-ma-toe-sis) is a genetic condition of abnormal iron metabolism that permits absorption of too much iron from an ordinary diet. Hereditary hemochromatosis is an autosomal recessive disorder. It is NOT a blood disease. It is also known

as iron overload or iron storage disease. It is possible for someone who has never had an iron pill in his/her life to have iron overload.

Q: Can iron overload be acquired?

A: Yes, iron overload can be acquired. The genetic form is known as primary hemochromatosis, hereditary hemochromatosis (HH) or (HHC), or genetic hemochromatosis (GH) and idiopathic hemochromatosis (from an unknown origin), a term which is rarely used anymore. The acquired form (through massive doses of iron pills or blood transfusions) is known as secondary hemochromatosis, acquired hemochromatosis, or transfusional iron overload.

Q: How common is iron overload/hemochromatosis?

A: Frequency (incidence in the general population) of the abnormal gene is: 1 in 100-200 people has hemochromatosis (double gene mutation known as a homozygote) and 1 in 8-10 people is a carrier of hemochromatosis (single gene mutation known as a heterozygote or “het” for short). That’s approximately 32 million Americans who are carriers and 1.5 million Americans have the double gene which can lead to full blown hemochromatosis. Recent studies in Ireland, show a frequency of 1 in 4 as carriers of the single mutation and 1 in 64 as double gene mutation. Because of this high frequency, routine screening for hereditary hemochromatosis is medically indicated.

Q: Who is affected by iron overload/hemochromatosis?

A: Most affected people DO NOT KNOW they are accumulating dangerous stores of iron. Tragically underdiagnosed, no race, age, or gender is immune. (Premenopausal women do have iron overload as well as young children) The American Hemochromatosis Society (AHS) has made an official position statement and issued guidelines for diagnosis, treatment, and management of iron overload/hereditary hemochromatosis, including recommendations that all Americans age 2 years and older be routinely and universally screened for iron overload as well as genetic screening. All ethnic groups can be affected, but those with an Irish/Scottish/Celtic/British heritage have an even higher prevalence of the HH mutation. Hispanics and Afro Americans also have iron overload.

Q: How serious is iron overload, hemochromatosis?

A: The excess iron injures body organs and KILLS unless detected in time for adequate iron

storage removal. It is a very serious disease, but quite benign if detected early before organ damage has occurred. That is why routine screening is so important. HH is a lethal but treatable disease. Don't let anyone tell you that iron overload/HH is "nothing to worry about". The higher the ferritin level, the potential for serious organ damage is increased. An early diagnosis offers the patient a normal life span.

Q: Is there anything that can be done to treat or prevent iron overload?

A: Yes. Hereditary hemochromatosis is one of the few genetic diseases which has a prevention plan so that all organ damage and premature death can be completely prevented. When the excess iron IS detected EARLY and is ADEQUATELY removed, the individual can enjoy a normal life span in normal health. The motto of the American Hemochromatosis Society is: **"Prevention through Genetic Testing".**

Q: What are the symptoms of iron overload, hemochromatosis?

A: Patients can have iron overload and NOT have symptoms (asymptomatic) and that is the best time to diagnose the patient. Many doctors have been taught to look for "signs and symptoms" of HH but by the time symptoms appear, it is often too late to save the patient's life. Iron overload and storage in vital body organs can damage and may cause:

- chronic fatigue (the most common complaint by patients);
- cirrhosis/cancer of the liver (with or without a history of alcohol use);
- arthritis/joint pain;
- impotence/sterility/infertility; early menopause/irregular menses;
- hair loss; hair thinning
- diabetes (bronze diabetes, a darkening, graying of the skin not caused by sun exposure);
- cancer (cancer thrives on iron); (especially primary liver cancer)
- abdominal pain/swelling;
- weight loss;
- frequent colds/flu/infections, compromised immune system;
- headaches;
- hypothyroidism; (low thyroid)
- heart irregularities/heart failure/heart attack (especially in younger men);
- cirrhosis of the liver (with or without a history of alcohol use);
- hepatoma/liver cancer (the leading cause of death in HH);

- premature death.

Anyone with any combination of these symptoms, or a family history of these symptoms, should be tested for HH immediately. But remember, two important facts: 1.) There can be numerous generations of “silent carriers” of the mutation who never become ill and live to old age thereby giving a “false security” that HH doesn’t “run in the family” 2.) Some patients do not have symptoms until they are end stage and their lives cannot be saved. Early detection should be achieved through: 1.) Knowledge of genetic risk through DNA Testing 2.) Annual screening with serum iron, TIBC, and serum ferritin to assure that iron storage is not taking place.

Q: I went to the blood bank and they told me I was anemic; how could I have iron overload at the same time?

A: Blood banks do NOT screen for iron overload/hemochromatosis. They are basing their comments on the hematocrit or hemoglobin readings that they take prior to a blood donation (the finger prick test) and these are not the correct tests for iron overload storage! Yet blood banks continue to give out false information to their clients, telling them that they have low “iron” or even in some cases that their iron is high! The iron-overloaded person may be anemic at the same time. There are several types of anemia that are iron-loading! Hematocrit and hemoglobin are NOT tests for iron overload/hemochromatosis; ask your physician to test you with transferrin saturation (TS) which is calculated by dividing the serum iron by the TIBC (total iron binding capacity) and serum ferritin to confirm or rule out iron overload.

Q: How can I know if I have iron overload/hemochromatosis? What tests should be performed? I hear that there is a DNA genetic test kit for hemochromatosis, is that true?

A: A simple series of blood tests which can be performed by any doctor or lab can indicate iron levels. They must be proper iron measures: Total Iron Binding Capacity (TIBC) together with Serum Iron. Divide TIBC into Serum Iron to get the percentage of transferrin saturation. It is important that the serum ferritin is also performed at the same time and it should be done, if possible, while fasting. Refrain from iron pills for a week prior to the tests. A new test, serum ferritin-iron assay may also be available in the near future. The discovery of the hemochromatosis gene was announced in August 1996 by Mercator Genetics Inc. of Menlo Park, California (which was purchased by Progenitor in 1997. Bio-Rad Laboratories of Hercules, CA bought the patent from Progenitor. Bio-Rad currently holds the patent to the HFE mutation). The new genetic DNA test (HLA-H now known as HFE or HFe) has been commercially commercially available from many labs around the nation since 2/1997, including SmithKline

Beecham Clinical Laboratories currently known as Quest Diagnostics on a nationwide basis. Many university labs and other smaller independent genetics labs across the nation now offer DNA testing for HH. In the early years, many of them only tested for the one mutation (845A also known as Cys282tyr), but today most labs test for BOTH HH mutations (845A and 187G also known as cys282 and his63). There is a 3rd gene mutation, 65S, but it is not included in most labs' testing protocols. Several labs also test for both gene mutations and offer a handy "cheek brush" tissue collection kit or saliva test kit which collects saliva in a tube, which you can get through the mail and perform in the privacy of your own home. AHS president, Sandra Thomas, and Annette Taylor, PhD, president of Kimball Genetics, Denver, Colorado, partnered to offer the public a home test kit which patients could order themselves, without a doctor's order.

This method of genetic testing revolutionized the DNA testing format and helped many patients to get the life saving information that they needed. The "cheek brush" method (no needles/blood/pain) is great for kids and adults. More info on how to order these tests is available from the AHS office at 407-829-4488. If you wish to get genetic DNA testing without using a doctor or having the results on your medical records, you may contact HealthCheckUSA (www.healthcheckusa.com (http://www.healthcheckusa.com)) or call on their toll free number: 1-800-929-2044, to order the HFE test. The results are private and confidential and are mailed directly to the patient, thusly protecting the patient's medical information. You may contact the lab directly for current prices. Sometimes genetic counselors are available to patients as part of the cost of the testing. Ask the lab you select about this service. Two other labs which offer genetic testing are: 23and me (www.23andme.com (http://www.23andme.com)) which tests for three gene mutations for HH, and which gives the patient raw data but not an interpretation for $99.00 and GeneTrack (www.hemochromatosisdna.com (http://www.hemochromatosisdna.com)) which offers a discounted price of $156.00. The American Hemochromatosis Society strongly urges patients and their family members (male and female) to be genetically tested, including children, teens, young adults and seniors. If you have a question about your genetic testing, feel free to contact the AHS office directly for more information.

Q: I had the blood tests for iron overload and my doctor says I am "fine"; do I need to worry about it now?

A: First of all, always get copies of your medical lab reports for your home medical file and review them yourself. Make sure that the serum iron, TIBC, and serum ferritin tests are on the report and double check to make sure that you fall into the "safe zone" set by AHS–a ferritin under 150 and a saturation percentage of under 40%. Some labs have very "high" normal levels and you

might not really be in a safe zone. Many patients have contacted us who have iron studies in the "danger zone" but their doctors have told them that they are fine. It is prudent to find out for yourself. The same philosophy applies to the DNA test–make sure you get copies of the report for your own files and know if you have the single or double mutation and which of the two mutations you carry (you can even carry one of each mutation which would make you known as a compound heterozygote).

CONTINUED/.... (http://www.americanhs.org/faq.htm)

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