

Hereditary Hemochromatosis

Individuals who are concerned about the risk of carrying the HH gene are encouraged to discuss genetic testing with their physicians or healthcare providers and to consider having genetic counseling at GeneCare. During counseling, a detailed family history will be reviewed and appropriate testing determined for each individual situation.

Specimen Collection:

- Call GeneCare at (800) 277-4363 to discuss clinical indications, current testing, informed consent, fees and payment method.
- Complete Consent Forms and Laboratory Request. Enclose family history and/or pedigree.
- Label each specimen tube with patient name, birth date, and collection date.

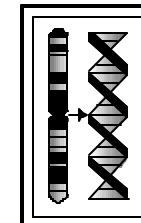
Specimen Transport:

- Enclose Laboratory Request / Consent Forms
- **SHIP AT ROOM TEMPERATURE** in our kit to:

GeneCare Medical Genetics Center
201 Sage Road, Suite 300
Chapel Hill, NC 27514
(800) 277-4363

- **DHL/Airborne** (800) 247-2676 **priority overnight** (or by courier) to reach GeneCare Monday - Thursday. Delivery required within 24 hours.
- Notify GeneCare of shipment date and DHL/Airport airbill /tracking number.

GeneCare Medical Genetics Center
201 Sage Road, Suite 300
Chapel Hill, NC 27514
(800) 277-4363 ? (919) 942-0021
Fax (919) 967-9519



GeneCare[®]
Medical Genetics Center

www.genecare.com

WHAT IS HEREDITARY HEMOCHROMATOSIS?

Hereditary Hemochromatosis (HH) is the most common genetic disorder known. It occurs in about 1 in 200 to 1 in 400 Caucasians, and approximately 12% (1 in 8) of Caucasians carry the gene for HH. HH occurs less frequently in other ethnic groups.

WHAT ARE THE MOST COMMON FEATURES OF HH?

HH is characterized by excess iron absorption from the gastrointestinal tract leading to storage of iron in the organs and eventually multiple organ damage, if left untreated. Symptoms of HH usually appear in adulthood (40-60 years), but there are documented cases of HH in children and young adults. Early symptoms of HH are often non-specific and include malaise, fatigue, impotence, abdominal pain, and joint pain. In the absence of treatment, serious complications include chronic liver disease and cirrhosis, hepatic carcinoma (liver cancer), diabetes (also referred to as bronze diabetes) and heart disease. Arthritis may develop and there may be a gradual darkening of the skin from a grayish hue to the characteristic bronze color known to be associated with HH. All of the above complications of HH can be prevented by early detection and treatment with phlebotomy (routine removal of blood via venipuncture). Because women lose blood during menstruation and pregnancy, symptoms occur less often or at later ages in women than in men.

HOW IS HH INHERITED?

HH is an autosomal recessive disorder. Autosomal means the abnormal gene is not on a sex chromosome. Recessive means the effect of the gene would only be seen when a person inherits an abnormal recessive gene from both parents. People who are carriers of HH have one abnormal (mutated) copy of the gene and one normal copy of the gene. When both parents are carriers of HH, they have a 25% risk (1 in 4 chances) of having a child who may develop HH, a 50% risk (1 in 2 chances) of having an unaffected carrier child, and a 25% chance of having an unaffected, non-carrier child with each pregnancy.

HOW IS THE HH TEST PERFORMED?

A DNA blood test can identify changes (mutations) within the HH gene. DNA studies detect individuals with the most common HH mutation, Cys282Tyr (C282Y), and a second mutation implicated in HH, known as His63Asp (H63D). Approximately 85-93% of people with HH have two copies of the Cys282Tyr mutation (homozygous for Cys282Tyr). Most homozygotes of the Cys282Tyr mutation develop hemochromatosis, and some heterozygotes (carrying one copy of the Cys282Tyr mutation) may have mild symptoms of HH such as lethargy, joint pain, and weakness. Another 3-5% of affected individuals are considered compound heterozygotes and have one Cys282Tyr mutation and one His63Asp mutation. Of people identified to be compound heterozygotes, only 1.5% in the general population develop clinical hemochromatosis.

WHO SHOULD HAVE HH TESTING?

- Individuals with elevated transferrin saturation (>50%) or serum ferritin concentration (>400 ng/ml in men and >200 ng/ml in women)
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Individuals with a clinical diagnosis of hemochromatosis

- Individuals with unexplained elevated serum concentrations of liver enzymes
- Individuals with cirrhosis, liver failure, or

hepatocellular carcinoma (liver cancer)

- Individuals with chronic unexplained fatigue, abdominal pain, or joint pain
- Individuals with hepatosplenomegaly (enlarged liver and spleen), cardiac arrhythmia (irregular heart beat), congestive heart failure, hyperpigmentation (bronze color), hypothyroidism, impotence, hypogonadism (small testes), diabetes mellitus
- Individuals with a relative or spouse with hemochromatosis
- Individuals with a relative or spouse known to be homozygous or heterozygous for the Cys282Tyr mutation
- Couples with a family history of hemochromatosis.

HOW LONG DOES IT TAKE TO COMPLETE THE TEST?

The DNA test for Hemochromatosis is completed within 2-4 days.

HOW CAN HH TESTING BE ARRANGED?

Physicians or healthcare providers may contact one of our Centers for consultations. Blood specimens may be sent by a physician or drawn at GeneCare if testing is recommended.