



Hemochromatosis DNA Test

Hereditary hemochromatosis (HH) is the most common genetic disorder known, with a prevalence of 1/200 and a carrier frequency of 1/8 in Caucasians. This disorder involves excess iron absorption and storage in organs and leads to high morbidity and mortality if untreated. Historically, most cases have remained undiagnosed until late in the course of the disease when irreversible damage has already occurred. Early diagnosis allows for prevention of symptoms through treatment by phlebotomy.

Two mutations associated with hemochromatosis have been discovered in the HFE gene. The major mutation, Cys282Tyr (also known as 845A) is present in the homozygous state in 85% of Caucasians with clinically diagnosed hemochromatosis. Most homozygotes (with two copies of the mutation) and some heterozygotes (with one copy of the mutation) develop iron overload and hemochromatosis. A second mutation, His63Asp (also known as 187G), appears to somewhat increase risk of iron overload, although only a small percent of individuals (up to 6%) with this mutation develop hemochromatosis. DNA testing for hemochromatosis provides for effective early diagnosis and prevention.

Indications for Hemochromatosis DNA Testing:

- Elevated transferrin saturation (>50%) or serum ferritin concentration (>400 ng/ml in men and >200 ng/ml in women)
- Clinical diagnosis of hemochromatosis
- Unexplained elevated serum concentrations of liver enzymes
- Cirrhosis, liver failure, or hepatocellular carcinoma
- Chronic unexplained fatigue, abdominal pain, or joint pain
- Hepatosplenomegaly, cardiac arrhythmia, congestive heart failure, hyperpigmentation, hypothyroidism, impotence, hypogonadism, diabetes mellitus
- Relative or spouse with hemochromatosis
- Relative or spouse known to be homozygous or heterozygous for the Cys282Tyr mutation

Our Hemochromatosis DNA Test Service Provides:

- PCR analysis for both the Cys282Tyr and the His63Asp mutations
- Rapid (1 day) turnaround time
- Detailed reports with genetic interpretation, recommendations and education
- Genetic consultation by board-certified genetic counselors and geneticists

Specimen Requirements:

- Check cell sample OR 5 ml blood in an EDTA (lavender top) tube, room temperature

Turnaround Time:

- 1 business day

Please call Kimball Genetics for more information.

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