Hemochromatosis (HFE) Mutation Testing

Shodair CHILDREN'S HOSPITAL
www.shodair.org

DESCRIPTION:

- Hereditary Hemochromatosis (HH) is a recessive disorder associated with iron overload. Iron overload produces a myriad of symptoms, including joint and abdominal pain, malaise, fatigue, and impotence. Complications include hyper-pigmentation, diabetes, heart failure, cirrhosis, liver cancer, and hypogonadism.
- Early symptoms and signs are minimal. However, early detection is critical to long term prognosis, given the effective treatment (phlebotomy) for this disorder.
- Carrier frequency is as high as 1 in 8 in populations of northern European origin.

REASONS FOR REFERRAL:

- Confirm or rule out the diagnosis of HH in an individual with symptoms and/or signs, positive family history or suggestive laboratory findings.
- This test detects two mutations in the HFE gene known to be associated with clinical hemochromatosis (Feder et al., *Nature Genet,* 1996, 13:399-408). Detection of hemochromatosis is over 85% in Caucasians.

METHOD OF ANALYSIS:

- DNA is amplified by PCR using pairs of primers flanking regions coding for amino acids 63 and 282, and sizes of the resulting fragments are determined by high resolution gel electrophoresis after digestion with MboI and RsaI, respectively.
- Results are reported within 2 weeks or less of receipt of sample and can be expedited upon request.

REFERENCE RANGES:

- Homozygous for C282Y (likely to develop hemochromatosis).
- Heterozygous for C282Y (carrier, probability of hemochromatosis about 0.5% due to undetected mutation in the other allele).
- Compound heterozygous for C282Y/H63D (risk for hemochromatosis about 1.5%)
- Homozygous for H63D (risk of clinical disease less than 0.4%).
- Heterozygous for H63D (very low risk, carrier of a low-penetrance HH mutation).
- Normal homozygote for C282 and H63 (very low risk for disease).

SAMPLE REQUIREMENTS:

- For DNA testing, 5 to 10 milliliters of blood (minimum 1 ml) in EDTA (purple top) tubes should be sent by overnight carrier at room temperature.
- Prenatal testing: Not available

TEST CPT CODES:

CPT 83890 DNA extraction
CPT 83894 DNA separation
CPT 83898 DNA amplification
CPT 83912 DNA interpretation and report

Discounts from list price are available for institutional billing under contractual arrangement with the laboratory. Contact Ellen Livers at 800-447-6614 ext 7523.

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