Hereditary Hemochromatosis (C282Y and H63D mutation) by PCR and RFLP

Clinical Indication and Relevance

Please refer to GeneTests Web "HFE-Associated Hereditary Hemochromatosis".
http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=hemochromatosis

Methodology

DNA is isolated from the patient sample and the *HFE* gene containing C282Y and H63D mutation sites is PCR-amplified, digested with restriction endonucleases, separated by gel electrophoresis, and analyzed by restriction fragment length polymorphisms (RFLP). Results are reported as normal, heterozygous or homozygous for C282Y and/or H63D mutation.

Sensitivity

N/A

Turn-around Time

Five to ten working days

Sample Requirements

Collect

Peripheral blood (PB): 3-5 mL (1mL minimum), in purple top (sodium EDTA) tube; yellow top (ACD) tube acceptable.

Transport

Ambient or 2-8°C (wet ice or cold packs). Do Not Freeze Peripheral Blood.

Stability

Ambient - 8 hours; refrigerated - 48 hours.

Unacceptable Conditions

Serum or plasma; frozen peripheral blood; clotted blood; severely hemolyzed blood samples.

CPT Code(s)

81256: *HFE* (hemochromatosis) gene analysis, common variants (e.g., C282Y, H630)
G0452-26: Molecular pathology procedure; physician interpretation and report