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

Clinical Indication and Relevance

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