Hemochromatosis (HFE) 3 Mutations
ARUP test code 0055656

HFE PCR Specimen | Whole Blood
---|---

C282Y Hemochromatosis Mutation | Homozygous

H63D Hemochromatosis Mutation | Negative

S65C Hemochromatosis Mutation | Negative

Hemochromatosis Mutation Interpretation
See Note

Indication for testing: Carrier screening or diagnostic testing for hereditary hemochromatosis.

Hemochromatosis Interpretive Results:
Homozygous C282Y:
C282Y Homozygous - The patient is homozygous for the HFE C282Y mutation and is at high risk for hereditary hemochromatosis. Homozygosity for this mutation accounts for 80-90 percent of the hemochromatosis patients of Northern European descent. The frequency of this mutation in other populations is lower.

H63D: Negative - This patient is negative for the HFE H63D mutation.

S65C: Negative - This patient is negative for the HFE S65C mutation.

This result has been reviewed and approved by Pinar Bayrak-Toydemir, M.D., Ph.D.
BACKGROUND INFORMATION: Hemochromatosis (HFE) 3 Mutations

CHARACTERISTICS: Disorder of iron metabolism resulting in excessive iron storage leading to increased skin pigmentation, arthritis, hypogonadism, diabetes mellitus, heart arrhythmias/failure, cirrhosis and liver carcinoma.

INCIDENCE: One in 300 individuals of Northern European descent; unknown in other ethnicities.

INHERITANCE: Autosomal recessive.

PENETRANCE: 5 percent of C282Y homozygotes, 1 percent of C282Y/H63D compound heterozygotes and rare H63D homozygotes develop clinical symptoms.

CAUSE: Two pathogenic HFE gene mutations on opposite chromosomes.


CLINICAL SENSITIVITY: 85 percent of hereditary hemochromatosis in Northern Europeans is caused by C282Y homozygosity and 5 percent by C282Y/H63D compound heterozygosity.

METHODOLOGY: PCR and fluorescence monitoring.

ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.

LIMITATIONS: HFE mutations, other than those targeted, will not be detected. Diagnostic errors can occur due to rare sequence variations.

This test is performed pursuant to an agreement with BioRad Laboratories, Inc.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

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H=High, L=Low, *=Abnormal, C=Critical

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