

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB [REDACTED]
Gender: Male
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Hemochromatosis (HFE) 3 Mutations

ARUP test code 0055656

HFE PCR Specimen whole Blood

C282Y Hemochromatosis Mutation Negative

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:
Negative WT:
C282Y: Negative - The patient is negative for the HFE C282Y mutation.
H63D: Negative - The patient is negative for the HFE H63D mutation.
S65C: Negative - The patient is negative for the HFE S65C mutation.
Mutations in unidentified genes or other mutations in the HFE gene are not ruled out.
This result has been reviewed and approved by Yuan Ji, Ph.D.

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

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.
MUTATIONS TESTED: p.C282Y (c.845G>A), p.H63D (c.187C>G), and p.S65C (c.193A>T).
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

Procedure	Accession	Collected	Received	Verified/Reported
HFE PCR Specimen	18-216-103751	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C282Y Hemochromatosis Mutation	18-216-103751	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
H63D Hemochromatosis Mutation	18-216-103751	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
S65C Hemochromatosis Mutation	18-216-103751	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemochromatosis Mutation Interpretation	18-216-103751	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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