ARUP LABORATORIES
800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	10/29/1991
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	Homozygous
S65C Hemochromatosis Mutation	Negative
Hemochromatosis Mutation Interpretation	See Note

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

Hemochromatosis Interpretive Results:
Homozygous H63D:
C282Y: Negative - The patient is negative for the HFE C282Y mutation.
H63D: Homozygous - The patient is homozygous for the HFE H63D mutation.
S65C: Negative - The patient is negative for the HFE S65C mutation.
This genotype has not been significantly correlated with hereditary hemochromatosis. If clinical symptoms consistent with hemochromatosis persist, please follow the patient accordingly and /or refer to a hematologist.

This result has been reviewed and approved by Pinar Bayrak-Toydemir, M.D., 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 SENSITIVTY 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 was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
HFE PCR Specimen	22-088-140044	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
C282Y Hemochromatosis Mutation	22-088-140044	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
H63D Hemochromatosis Mutation	22-088-140044	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
S65C Hemochromatosis Mutation	22-088-140044	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemochromatosis Mutation Interpretation	22-088-140044	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