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Client:

Physician:

Patient:
DOB:
Gender:
Patient Identifiers:
Visit Number (FIN):
Collection Date:

Hemochromatosis (HFE) 3 Mutations

ARUP test code 0055656

HFE PCR Specimen	Whole Blood
C282Y Hemochromatosis Mutation	Heterozygous
H63D Hemochromatosis Mutation	Heterozygous
S65C Hemochromatosis Mutation	Negative
Hemochromatosis Mutation Interpretation	See Note

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

Hemochromatosis Interpretive Results:
Compound Heterozygous C282Y/ H63D:
C282Y Heterozygous - The patient is heterozygous for the HFE C282Y mutation.
H63D Heterozygous - The patient is heterozygous for the HFE H63D mutation.
S65C: Negative - The patient is negative for the HFE S65C mutation.
The patient may be at an increased risk for hereditary hemochromatosis. Approximately 5 percent of hemochromatosis patients have this genotype. The penetrance of this genotype is 0.5-1.5 percent. Due to this reduced penetrance, compound heterozygotes may have a milder phenotype than C282Y homozygotes. Less than 2 percent of individuals with the compound heterozygote genotype develop hereditary hemochromatosis.

This result has been reviewed and approved by Yuan Ji, 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.
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.

H – high L – low * – abnormal C – critical

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 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

Order Comments

Hemochromatosis (HFE) 3 Mutations

Client Accession number:

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
HFE PCR Specimen	17-021-104444	1/21/2017 08:49	1/23/2017 13:38	2/3/2017 14:02
C282Y Hemochromatosis Mutation	17-021-104444	1/21/2017 08:49	1/23/2017 13:38	2/3/2017 14:02
H63D Hemochromatosis Mutation	17-021-104444	1/21/2017 08:49	1/23/2017 13:38	2/3/2017 14:02
S65C Hemochromatosis Mutation	17-021-104444	1/21/2017 08:49	1/23/2017 13:38	2/3/2017 14:02
Hemochromatosis Mutation Interpretation	17-021-104444	1/21/2017 08:49	1/23/2017 13:38	2/3/2017 14:02

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H – high L – low * – abnormal C – critical