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PATIENT REPORT

Patient Age/Sex: 31 years Female

Specimen Collected: 5/6/2025 08:03 MDT

Hemochromatosis (HFE) 3 Variants |Received: 5/6/2025 08:04 MDT Report/Verified: 5/7/2025 15:47 MDT

Procedure	Result	Units	Reference Interval
HFEPCR Specimen	Whole Blood		
C282Y Hemochromatosis Variant	Homozygous		
H63D Hemochromatosis Variant	Negative		
S65C Hemochromatosis Variant	Negative		
Hemochromatosis Interpretation	See Note f1 i1		

Result Footnote

f1: Hemochromatosis Interpretation

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 [redacted]

Test Information

i1: Hemochromatosis Interpretation

BACKGROUND INFORMATION: Hemochromatosis (HFE) 3 Variants

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.

Test Information

i1: Hemochromatosis Interpretation

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.