ClinLab
Clinical Testing Laboratory, Inc.

Clinical Testing Lab of Washington
2150 Pennsylvania Avenue NW
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Phone: 202-555-1212

Specimen Number	Specimen Type	Control Number	Account Number	Account Phone Number	Route
	Peripheral Blood				

Jeff (Patient Last Name)			Patient Barcode
Patient First Name	Patient Middle Name		
Patient SS#	Patient Phone	Total Volume	
46 y.o.	Male		
Age (Y/M/D)	Date of Birth	Sex	Fasting
Patient Address		Additional Information: **Indication: Suspected Hemochromatosis** **Family History: No known family history** **Ethnicity: Western European Caucasian**	

Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID
			Jane Ferreiro, MD		

Tests Ordered: **Hereditary Hemochromatosis Panel**

General Comments: **Please send a copy of the final report to the Molecular Science/M1 Training office via Fax at (202) 555-1212**

Clinical test results for Hereditary hemochromatosis

6 conditions tested:

- Hereditary hemochromatosis (type 1)
- Hemochromatosis type 2A
- Hemochromatosis type 2B
- Hemochromatosis type 3
- Hemochromatosis type 4
- Juvenile hemochromatosis

GENE	TEST RESULTS	EXPLANATION
HAMP	Negative	No known pathogenic variant detected
HFE	p.Cys282Tyr p.Cys282Tyr	**This result confirms the diagnosis of or predisposition for Hereditary hemochromatosis** (type 1). This result should be interpreted in the context of clinical presentation and results of other laboratory tests (e.g., serum transferrin-iron saturation and serum ferritin). A PCR/sequencing study has detected two copies of the Cys282Tyr (HFE g.10633G>A, c.845G>A, p.Cys282Tyr) variation. The Cys282Tyr mutation is a G to A change at nucleotide position 10633 in the HFE gene, 845 in the primary HFE transcript and results in a change from cysteine to tyrosine at amino acid position 282. In addition, this individual's result has important implications for other family members. Clinical and laboratory evaluations should be considered for at risk individuals. Genetic counseling is recommended for at risk individuals.
HFE2	Negative	No known pathogenic variant detected
SLC40A1	Negative	No known pathogenic variant detected
TFR2	Negative	No known pathogenic variant detected

DISCLAIMER:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.

ASSAY METHODS

Full-Gene Sequencing covers the full gene coding sequence, +/- 10 base pairs of adjacent intronic sequence, and other non-coding sequence positions containing select known pathogenic variants. Deletion/Duplication Analysis detects most intragenic deletions and duplications at single exon resolution. Rarely however, single-exon duplication events may be missed due to inherent sequence properties or isolated reduction in data quality.

CLINICAL DESCRIPTION

HFE-associated hereditary hemochromatosis (HFE-HH) is characterized by inappropriately high absorption of iron by the gastrointestinal mucosa. The phenotypic spectrum of HFE-HH is now recognized to include Those with clinical HFE-HH, in which manifestations of end-organ damage secondary to iron storage are present; Those with biochemical HFE-HH, in which the only evidence of iron overload is increased transferrin-iron saturation and increased serum ferritin concentration; and Non-expressing p.Cys282Tyr homozygotes in whom neither clinical manifestations of HFE-HH nor iron overload are present. Clinical HFE-HH is characterized by excessive storage of iron in the liver, skin, pancreas, heart, joints, and testes. In untreated individuals: early symptoms may include abdominal pain, weakness, lethargy, and weight loss; the risk of cirrhosis is significantly increased when the serum ferritin is higher than 1,000 ng/mL; other findings may include progressive increase in skin pigmentation, diabetes mellitus, congestive heart failure and/or arrhythmias, arthritis, and hypogonadism. Clinical HFE-HH is more common in men than women.

REFERENCES

- Rochette J, et al. Factors influencing disease phenotype and penetrance in HFE haemochromatosis. Human Genetics.2010;128(3):233–248.
- Whitlock EP, Garlitz BA, Harris EL, et al. Screening for hereditary hemochromatosis: a systematic review for the U.S. preventive services task force. Annals of Internal Medicine. 2006;145(3):209–223.
- Bacon BR, Adams PC, Kowdley KV, Powell LW, Tavill AS. Diagnosis and management of hemochromatosis: 2011 practice guideline by the American Association for the study of liver diseases. Hepatology. 2011;54(1):328–343.