Dr Lal PathLabs
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GENEVOLVE
Genomics Division
LPL - PRODUCTION TEST COLLECTION CENTRE
SECTOR - 18, BLOCK-E ROHINIDELHI 110085

Name	**DUMMY**					**Collected**	1/6/2021 5:31:00AM
						Received	1/6/2021 5:34:02PM
Lab No.	DUMMYN235	**Age:**	25 Years	**Gender:**	Male	**Reported**	4/6/2021 4:52:40PM
A/c Status	P	**Ref By:**	DR. DUMMY DUMMY			**Report Status**	Final

TEST CONDUCTED	HEMOCHROMATOSIS DETECTION, HFE GENOTYPING

RESULTS

HFE GENE MUTATION H63D	HFE GENE MUTATION S65C	HFE GENE MUTATION C282Y
MUTATION NOT DETECTED	MUTATION NOT DETECTED	MUTATION NOT DETECTED

Interpretation

RESULT	REMARKS	CONCLUSION
HOMOZYGOUS MUTANT	Two copies of the mutant allele detected	Hemochromatosis could be there
HETEROZYGOUS	One copy of mutant and one copy of wild type allele detected	Hemochromatosis could be there
MUTATION NOT DETECTED	Two copies of the wild type allele detected	Normal individual

Note

- Results must be interpreted in context with clinical findings, family history and other relevant laborator data. Rare polymorphisms exist that could lead to false-negative or false-positive results. If result obtained do not match the clinical findings, additional testing should be considered.
- Genetic Counselling is recommended.
- This is an in-house developed assay. The characteristics of this assay has been validated usin standard guidelines by local and International accreditation agencies.
- Test conducted on EDTA whole blood.
- This test detects only the mutations mentioned above in the HFE gene.
- Bone marrow transplantation from allogenic donor is known to interfere with the test, hence it i recommended to provide complete clinical history of the patient before sending specimen for testing.

Comment

HFE GENE MUTATIONS-HFE-Hereditary hemochromatosis (HFE-HHC), also known as primary hemochromatosis, genetic hemochromatosis or "bronze diabetes", is characterized by inappropriately high absorption of iron by the GI tract, resulting in iron accumulation in the liver, skin, pancreas, heart, joints, and testes. Hepatic cirrhosis may occur in untreated individuals. It is an adult-onset, treatable disease is caused by mutations in the HFE gene. HHC is inherited in an autosomal recessive manner. Two common mutations (C282Y and H63D) account for 87% of cases.

- C282Y - it is the most common mutation in the HFE gene is C282Y; Homozygosity for the C282Y mutation is associated with 60% to 90% of all cases of HH. Additionally, 3% to 8% of individuals affected with HH are heterozygous for this mutation.

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- H63D - this mutation is associated with HH, but the actual clinical effects of this mutation are uncertain. Homozygosity for H63D is insufficient to cause clinically significant iron overload in the absence of additional modifying factors.
- C282Y/H63D - compound heterozygosity for C282Y/H63D has been associated with increased hepatic iron concentrations.
- S65C - . The clinical significance of this mutation appears to be minimal. This rare variant displays a very low penetrance.
- C282Y/S65C - Compound heterozygosity for C282Y and S65C may confer a low risk for mild HH.
- S65C/H63D - Individuals who are heterozygous for S65C and either the wild-type or H63D alleles do not seem to be at an increased risk for HH

DMC-28322

Dr Lopamudra Deka
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Deputy HOD Histopath, Cytopath
& Chief of Academic Program – NRL

Dr Atul Thatai
PhD, Biotechnology
National Head - Molecular Diagnostics
/R & D
NRL - Dr Lal PathLabs Ltd

----------End of report ----------

IMPORTANT INSTRUCTIONS

*Test results released pertain to the specimen submitted.*All test results are dependent on the quality of the sample received by the Laboratory. *Laboratory investigations are only a tool to facilitate in arriving at a diagnosis and should be clinically correlated by the Referring Physician .*Sample repeats are accepted on request of Referring Physician within 7 days post reporting.*Report delivery may be delayed due to unforeseen circumstances. Inconvenience is regretted.*Certain tests may require further testing at additional cost for derivation of exact value. Kindly submit request within 72 hours post reporting.*Test results may show interlaboratory variations.*The Courts/Forum at Delhi shall have exclusive jurisdiction in all disputes/claims concerning the test(s) & or results of test(s).*Test results are not valid for medico legal purposes. *Contact customer care Tel No. +91-11-39885050 for all queries related to test results.
(#) Sample drawn from outside source.