CRM ID	: 0000000	Sample Type	: NA
Name	: DUMMY	Date & Time Collected	: DD-MM-YYYY
Sex/Age	: NA	Date & Time Received	: DD-MM-YYYY
Bill. Loc.	: NA	Date & Time Reported	: DD-MM-YYYY
Ref. By	: NA		

Hereditary Hemochromatosis:
HFE Gene three common mutation screening report

Specimen Description: Sample quality is optimum for the test. DNA conc.:52.5 ng/µl

Methodology

The test is performed by end point PCR using gene specific primers followed by automated DNA sequencing of the amplicon using BigDye™ Terminator Chemistry on an ABI Genetic Analyzer 3500DX platform. This assay only screens H63D, S65C and C282Y mutation in HFE gene.

Result

Serial Number	Mutation	Remarks
1.	c.845G>A; p.C282Y	No Variant Identified
2.	c.193A>T; p.S65C	No Variant Identified
3.	c.187C>G; p.H63D	No Variant Identified

Clinical Background

Hereditary hemochromatosis (HH)is an autosomal recessive disorder of iron metabolism. Accumulation of excess iron results in damage to multiple organs including the liver, pancreas, heart, joints, and the brain. Individuals may develop cirrhosis of theliver,liver tumors, diabetes, arthritis, and/or heart disease. For individuals with clinical symptoms consistent with HH orbiochemical evidence of iron overload, an HH diagnosis is typically based on the results of transferrin-iron saturation and serum ferritin concentration The majority of HH patients have three common mutations in hereditary hemochromatosis (HFE) gene: H63D, S65C and C282Y. Clinically significant iron overload also can occur in the

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absence of known HFE gene mutations, so a negative HFE test does not exclude a diagnosis of iron overload or hemochromatosis.

Limitation of the test

1. This assay will only screen the H63D, S65C and C282Y mutation in HFE gene.
2. This assay will not detect all of the mutations that cause hereditary hemochromatosis. Therefore, the absence of a detectable mutation does not rule out the possibility that an individual is a carrier of or affected with this disease.
3. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data.
4. Diagnostic errors can occur due to rare sequence variations.Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

References

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2. Walsh A, Dixon JL, Ramm GA, et al: The clinical relevance of compound heterozygosity for the C282Y and H63D substitutions in hemochromatosis. Clin Gastroenterol Hepatol 2006;4(11):1403-1410.
3. Whitlock EP, Garlitz BA, Harris EL, et al: Screening for hereditary hemochromatosis: a systematic review for the U.S. Preventive Services Task Force. Ann Intern Med 2006;145(3):209-223.

---------- End Of Report ----------

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2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

3. It is to be noted that variations in results may occur between different laboratories and over time, even for the same parameter for the same Customer. The assays are performed and conducted in accordance with standard procedures, and the reported outcomes are contingent on the specific individual assay methods and equipment(s) used, as well as the quality of the received specimen.

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