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Dr.SAMPLE REPORT
TEST HEALTH CENTRE
123 TEST STREET
BURWOOD VIC 3125

SAMPLE REPORT

09-May-1990 Female

16 HARKER STREET
BURWOOD VIC 3125

LAB ID : 3814220
UR NO. :
Collection Date : 09-May-2022
Received Date:09-May-2022



3814220

Genomic Assessments

BLOOD - EDTA Result Range Units

HAEMOCHROMATOSIS

C282Y mutation: C282Y mutation not detected: Result: Normal

H63D mutation: H63D mutation DETECTED on one allele. Result: **Heterozygote**

Test performed by accredited laboratory NATA: 2133

COMMENT: H63D Heterozygote.

H63D heterozygosity or homozygosity are not associated with hereditary haemochromatosis when C282Y alleles are normal. This genotype carries only a very slight increased relative risk of hereditary haemochromatosis. Patients with hereditary haemochromatosis usually have a serum transferrin saturation greater than 50% and an elevated ferritin.

Approximately 90% of people with symptoms of hereditary haemochromatosis are homozygous for the C282Y mutation, while approximately 2% of affected individuals are compound heterozygotes for both the C282Y and H63D mutations.

Those individuals who are heterozygous for only the C282Y mutation account for approximately 1% of people with symptoms of hereditary haemochromatosis, while those who are homozygous for the H63D mutation only rarely develop the condition.

Approximately 8-10% of people with the symptoms of haemochromatosis do not exhibit either of these genetic abnormalities.

Tests ordered: HAEMO,CFee