DOUGLASS HANLY MOIR PATHOLOGY
Quality is in our DNA

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Janine WADE
Lab ID : **838756989**
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DOB : 14/01/1982 (42 Yrs)
Sex : Female
Ph : 0404566722

Your Ref : .

Requested : 17/01/2024
Collected : 05/02/2024 15:04
Received : 05/02/2024 15:05
Printed : 15/02/2024 15:59

HFE-related Hereditary Haemochromatosis

Sample Type	**EDTA blood**
Method	**Melt-curve genotyping**
Result:	
HFE: c.845G>A (C282Y)	**Not Detected**
HFE: c.187C>G (H63D)	**Not Detected**
Interpretation	**This result does not support a diagnosis of the most common type of HFE-related HH. In patients with iron overload, investigation for other causes (for example fatty liver disease, metabolic syndrome, and rarer genetic causes of HFE- and non-HFE-related HH) should be considered.**

Comment
Test Information:

The vast majority (>90%) of patients with clinically characterised hereditary haemochromatosis are homozygous for the C282Y HFE variant, referred to as HFE-related HH (PMID 26153218). Genotyping is performed by melt curve analysis on extracted genomic DNA using the TIB MOLBIOL LightMix HFE kit for the detection of the HFE (LRG_749t1) variants c.845G>A (p.(Cys282Tyr), commonly known as "C282Y") and c.187C>G (p.(His63Asp), commonly known as "H63D") that are associated with HFE-related hereditary haemochromatosis (HH). This test will not detect other rare pathogenic HFE variants or genetic causes of non-HFE-related HH. Genetic test results may have significant implications for both the individual and their relatives. Corroboration of this result by reference to other clinical or laboratory information or by repeat testing may be warranted.
As of November 2021, the S65C HFE variant is no longer reported, in accordance with best practice guidelines (PMID:26153218). Please contact the laboratory if this result is required.

Tests Completed: GAH(e). **FINAL REPORT**

Clinical Notes: persistently raised tf saturation previously high iron ?hfe gene heterozygous ho...