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Title: Primary Sample Manual: Molecular Biology – Haematology

Changes made since previous version: Original. Replacement for PSM MOLECULAR BIOLOGY

Note: *Please refer to the document record on QPulse for the revision history of this document*

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1. HAEMOCHROMATOSIS

Haemochromatosis (HH) is an autosomal recessive disorder of iron metabolism that affects approximately 0.2-0.5% of the Caucasian population, with a higher than average incidence in the Irish population. The disease is characterized by the excessive accumulation of iron in the body and caused by an increased absorption of dietary iron at the intestinal mucosa level. The HFE gene is responsible for the disease and was identified in 1996 (Feder JN et al., 1996); it is localized on the short arm of chromosome 6, near the locus of the HLA-A gene. A mutation of this gene causes the synthesis of an abnormal protein unable to interact with the transferrin receptors, favoring the transport of iron through the intestinal mucosa. However, the exact mechanism with which the mutated HFE protein contributes to the increased intestinal absorption is not completely clear. The two most frequent mutations found in the HFE gene correspond to the C282Y mutation (substitution of a cysteine with a tyrosine in position 282 of the protein) and the H63D mutation (substitution of the histidine with an aspartic acid in position 63).

Preparation of patient: There is no physical preparation for the haemochromatosis test.

Precautions: None.

Accredited	No
Method	Molecular Biology - Real time PCR SOP: MB55
Sample Requirements	Tube Type: Whole blood EDTA Temperature: + 4ºC Miscellanous: Non fasting
Turnaround Time	7 working days from sample receipt.
Stability	Whole blood at room temperature: 7 days. Long term storage (>10 days) of blood samples intended for nucleic acid isolation should be in EDTA tubes at 4 °C. DNA extracts: 3 Months at -20˚C. Primary EDTA tube: 3 Months at 4˚C.
Units - Reference Ranges	Homozygous for C282Y and H63D mutations. Heterozygous for C282Y and H63D mutations. C282Y and H63D mutations not present.

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