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Effective Date: 08/11/17 Supersedes: 2.01

File Type: Master Assigned to: Quality Department

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Issue No.	Revision Details	Effective Date
1.01	Original	20/09/12
1.02	Storage temp for primary sample tube = -80. TAT for haemochromatosis, factor II and V updated to 10 days.	16/10/12
1.03	Updated TAT for factor II, V and haemochromatosis.	25/10/12
1.04	Updated accreditation status. Addition of revision table.	25/02/13
1.05	Updated TATs.	27/05/13
1.06	Updated samples requirements and storage post analysis for CT/NG testing.	01/08/13
1.07	Updated TATs	27/03/14
1.08	Removed Factor II, Factor V and CT/NG from PSM as no longer performed in house.	22/09/15
2.01	Eurofins Biomnis rebranding	24/05/17
2.02	Updated HFE units and reference ranges.	08/11/17

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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	Yes
Method	Molecular Biology - Real time PCR SOP: MB01
Sample Requirements	Tube Type: Whole blood EDTA Temperature: + 4°C Miscellaneous: Non fasting
Turnaround Time	10 days
Stability	DNA extracts: 3 Months at -20°C Primary EDTA tube: 3 Months at -80°C
Units - Reference Ranges	C282Y and H63D mutations not detected.

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