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Hemochromatosis mutation

General:

The iron storage disorder, hereditary hemochromatosis, is an autosomal recessive genetic disorder that results from defects in the HFE gene. The HFE gene is located on the short arm of chromosome 6 at location 6p21.3. The protein encoded by this gene is a membrane protein that is similar to MHC class I-type proteins and associates with beta2-microglobulin (beta2M). It is thought that this protein functions in regulating iron absorption by regulating the interaction of the transferrin receptor with transferrin.

The HFE-gene has three mutant alleles at different loci, H63D, C282Y and S65C. The two most common and best characterized mutations result in the substitution of cysteine by tyrosine at codon 282 (C282Y) and histidine by aspartic acid at codon 63 (H63D). A third, rarer mutation, S65C may be associated with a milder disease course when present in conjunction with a H63D or C282Y mutation, although this needs more detailed characterization.

Indication:	Clarification of familial hemochromatosis, hepatic cirrhosis, skin pigmentation (bronzed skin), highly elevated ferritin values
Material:	approx. 5 ml (2 x 2.7 ml EDTA monovette) EDTA blood
TAT:	7 - 10 days*
Method::	real time PCR

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit http://www.fml-dubai.com/parameter-listings/

Updated 03/04/2022