

Haemochromatosis StripA^{ssay} B

INTENDED USE

The *ViennaLab* Haemochromatosis StripA^{ssay} B provides materials for the isolation of DNA from human whole blood, the *in vitro* amplification of HFE exon 2 and 4 gene sequences, and the subsequent detection of three mutations by reverse-hybridization.

INTRODUCTION

Hereditary haemochromatosis (HH) is a very common autosomal recessive disorder of iron metabolism. Among individuals of Northern European descent the carrier frequency is estimated 1 in 10, resulting in up to 1 in 200 homozygous subjects being predisposed to develop the disease. HH is characterized by progressive accumulation of iron in various organs (liver, heart, pancreas), ultimately leading to liver cirrhosis, diabetes, arthritis, cardiomyopathies and premature death.

A number of mutations within a novel MHC class I-like gene (HFE) have been identified and related to HH. In the majority of Caucasian HH patients homozygosity for a single point mutation (C282Y) in exon 4 is observed. In addition, compound heterozygotes for C282Y and one of two other common mutations (H63D, S65C) within exon 2 of the HFE gene are at increased risk of developing haemochromatosis.

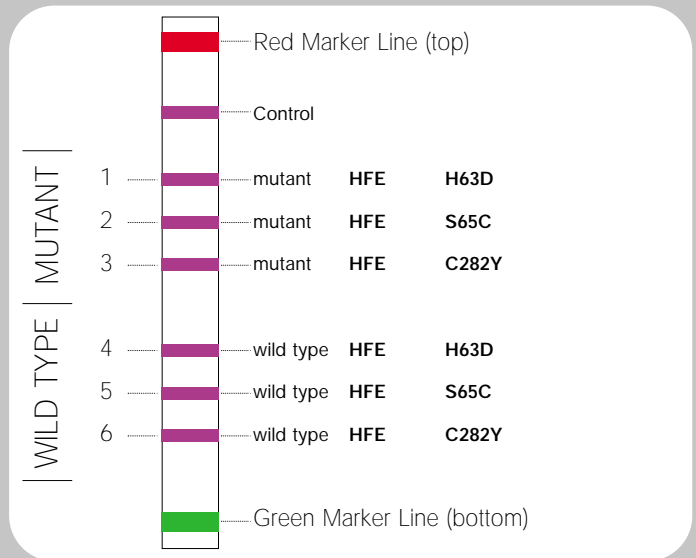
Molecular genetic testing for HH-associated mutations is considered valuable for carrier identification, as well as for pre-symptomatic diagnosis of the disease. With early detection and simple and very effective treatment by therapeutic bleeding (phlebotomy) in order to remove the iron overload, irreversible organ damage can be completely prevented and survival of patients is virtually normal.

PRINCIPLES OF THE ASSAY

The Haemochromatosis StripA^{ssay} B is based on the reverse-hybridization principle, and includes three successive steps: DNA is isolated from anticoagulated blood by a rapid and convenient procedure. Then, HFE exon 2 and 4 gene sequences are simultaneously *in vitro* amplified and biotin-labelled in a single («multiplex») amplification reaction. Finally, the amplification products are selectively hybridized to a test strip, which contains oligonucleotide probes (wild type- and mutant-specific) immobilized as parallel lines. Bound biotinylated sequences are detected using streptavidin-alkaline phosphatase and color substrates.

The assay covers the 3 most common mutations in the HFE gene (H63D, S65C, C282Y).

References: Feder, J.N., Gnirke, A., Thomas, W., et al. (1996), *Nature Genetics* 13, 399-408. Mura, C., Ragueneas, O., Ferec, C. (1999), *Blood* 93, 2502-2505.



Haemochromatosis^{StripAssay} B

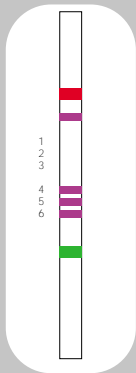
TEST RESULTS:

For each polymorphic position, one of three possible staining patterns may be obtained:

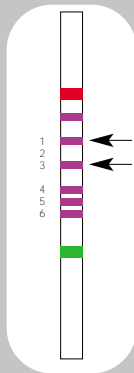
1. wild type probe only: *normal genotype*
2. wild type and mutant probe: *heterozygous genotype*
(«carrier» individual)
3. mutant probe only: *homozygous mutant genotype*
(«affected» individual)

EXAMPLES:

(A.) normal



(B.) H63D - C282Y compound heterozygous



(C.) C282Y homozygous



**VIENNA
LAB**

Labordiagnostika GmbH

Manufactured by:
ViennaLab
Vienna, Austria

www.viennalab.com

CAT.NO.:
4-210 / 4-211