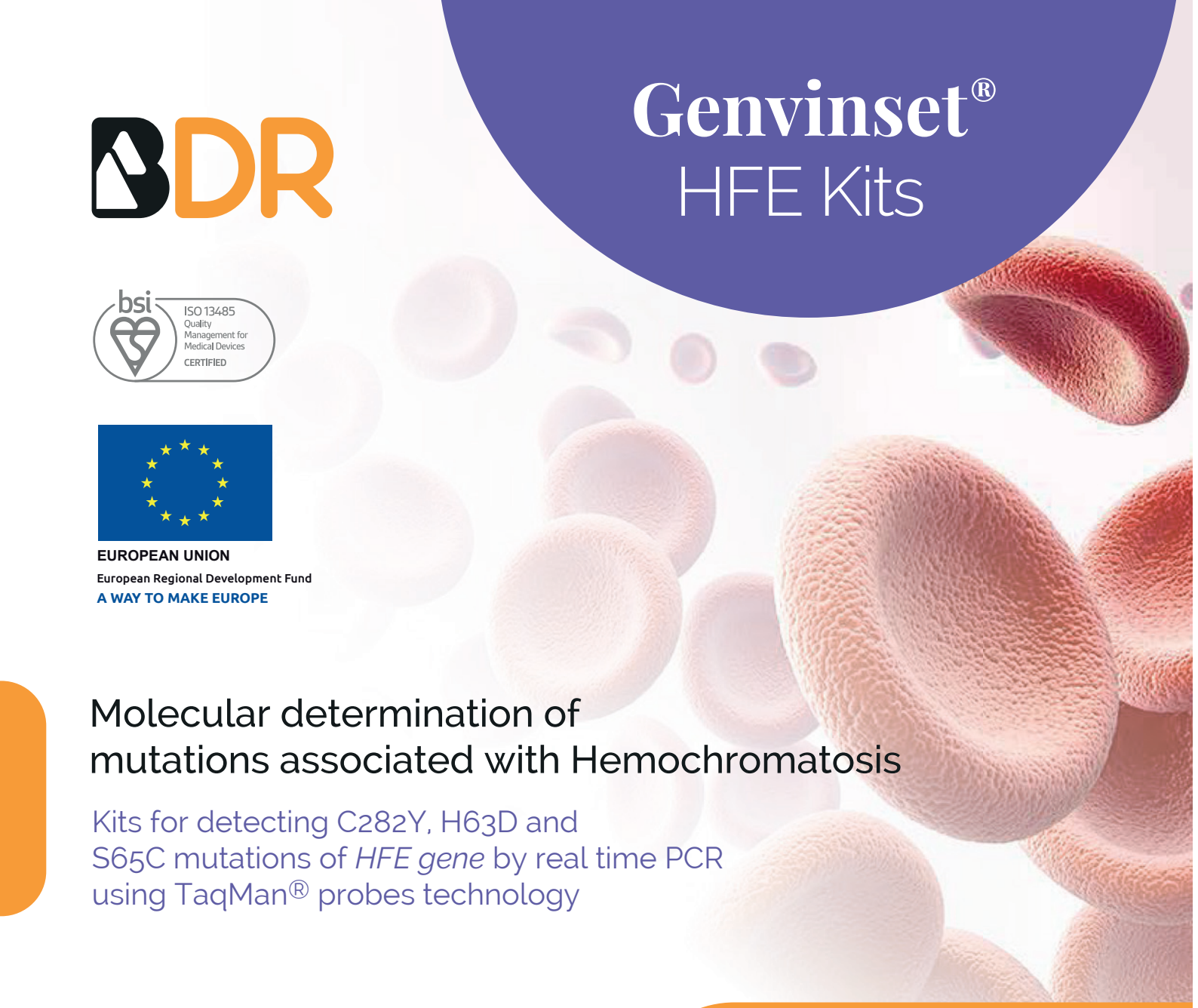
Genvinset®
HFE Kits



ISO 13485 Quality Management for Medical Devices CERTIFIED

EUROPEAN UNION
European Regional Development Fund
A WAY TO MAKE EUROPE

Molecular determination of mutations associated with Hemochromatosis

Kits for detecting C282Y, H63D and S65C mutations of *HFE gene* by real time PCR using TaqMan® probes technology

About Genvinset® HFE Kits

Hereditary hemochromatosis (HH) is an inherited autosomal recessive disorder of iron metabolism. Due to excessive intestinal absorption, iron accumulates in the parenchymal cells of the liver, pancreas, heart, and other organs resulting in structural injury and impaired function. The symptoms are often non-specific, and the organ damage is often irreversible once it has occurred. Therefore, early detection and treatment are critical as part of preventive medicine.

The discovery of the *HFE* gene in 1996 resulted in the inclusion of its molecular analysis in the diagnostic strategy for HH. Three mutations in the *HFE* gene (C282Y, H63D, and S65C) have been strongly associated with the development of iron overload, leading to a clinical diagnosis of HH.

Genvinset®
HFE C282Y

Intended use

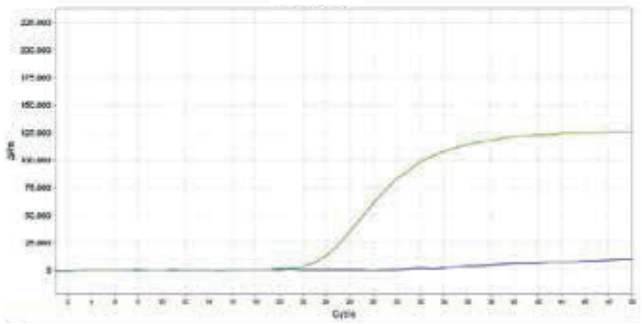
Genvinset® HFE C282Y is a semi-automated *in vitro* diagnostic kit for the qualitative detection of the C282Y mutation (NCBI dbSNP rs1800562; NM_000410.4:c.845G>A), in the *HFE* gene (OMIM: 613609) associated with primary hemochromatosis, in genomic DNA extracted from whole blood using Real-Time PCR technology with specific TaqMan® probes.

The patient referred by the corresponding health specialist (haematologist), and taking into account the compatibility of the symptoms presented; joint pain, abdominal pain, fatigue, weakness, heart failure, liver failure, bronze-coloured skin, and/or his family history (for example, a direct ascendant diagnosed with hereditary hemochromatosis) may be subject to the determination of the mutation in the *HFE* gene. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

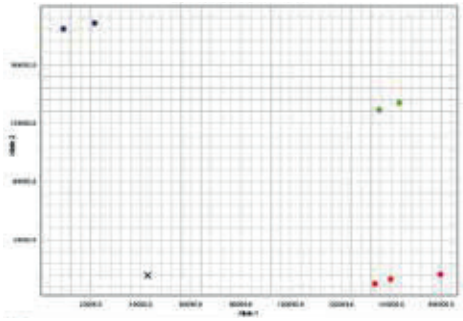
Results

Homozygous wild-type sample



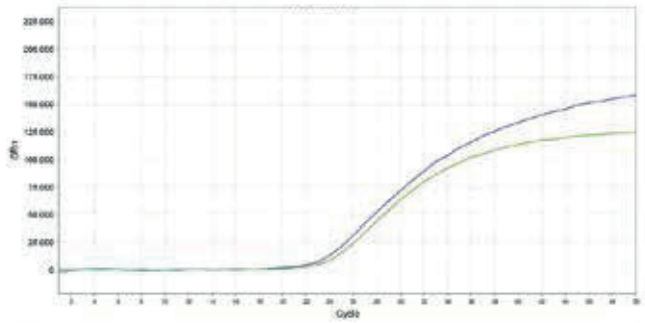
Wild-type allele Mutant allele

Allelic Discrimination Plot



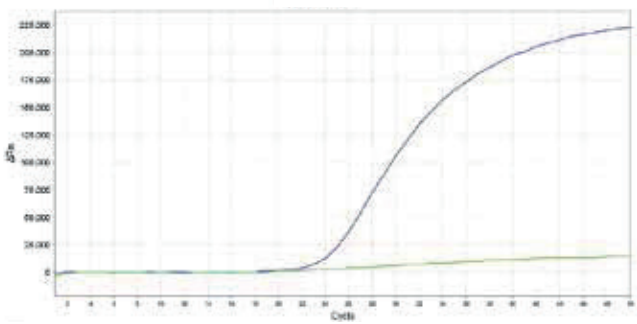
wt / wt mut / mut
wt / mut Reaction Blank

Heterozygous sample



Wild-type allele Mutant allele

Homozygous mutant sample



Wild-type allele Mutant allele

Genvinset®
HFE H63D

Intended use

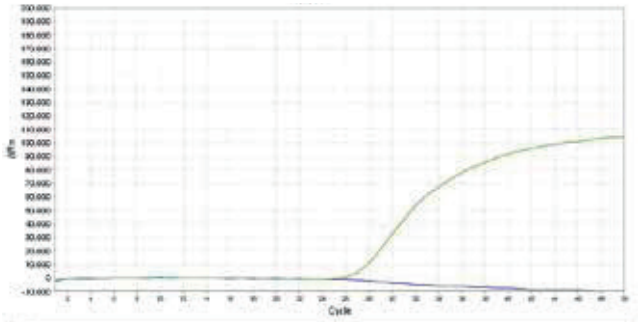
Genvinset® HFE H63D is a semi-automated *in vitro* diagnostic kit for the qualitative detection of the H63D mutation (NCBI dbSNP rs1799945; NM_000410.4:c.187C>G), in the *HFE* gene (OMIM: 613609) associated with hereditary hemochromatosis, in genomic DNA extracted from whole blood using Real-Time PCR technology with specific TaqMan® probes.

The patient referred by the corresponding health specialist (haematologist), and taking into account the compatibility of the symptoms presented; joint pain, abdominal pain, fatigue, weakness, heart failure, liver failure, bronze-coloured skin, and/or his family history (for example, a direct ascendant diagnosed with hereditary hemochromatosis) may be subject to the determination of the mutation in the *HFE* gene. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

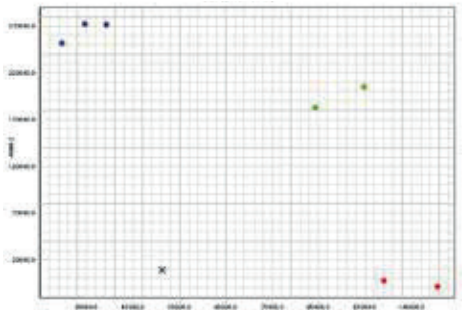
Results

Homozygous wild-type sample



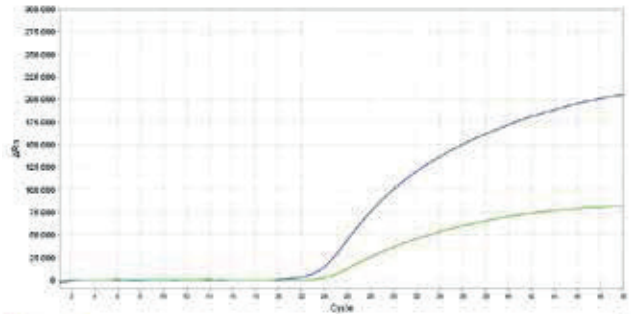
Wild-type allele Mutant allele

Allelic Discrimination Plot



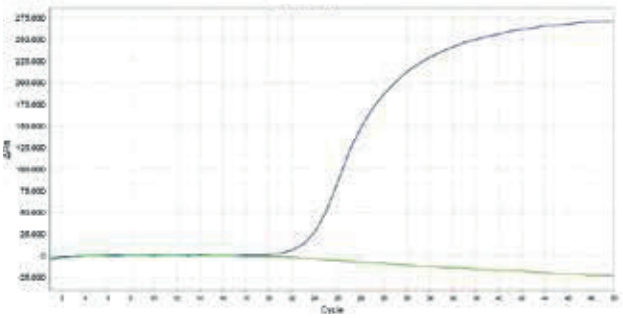
wt / wt mut / mut
wt / mut Reaction Blank

Heterozygous sample



Wild-type allele Mutant allele

Homozygous mutant sample



Wild-type allele Mutant allele

Genvinset®
HFE S65C

Intended use

Genvinset® HFE S65C is a semi-automated *in vitro* diagnostic kit for the qualitative detection of the S65C mutation (NCBI dbSNP rs1800730; NM_000410.4:c.193A>T), in the *HFE* gene (OMIM: 613609) associated with primary hemochromatosis, in genomic DNA extracted from whole blood using Real-Time PCR technology with specific TaqMan® probes.

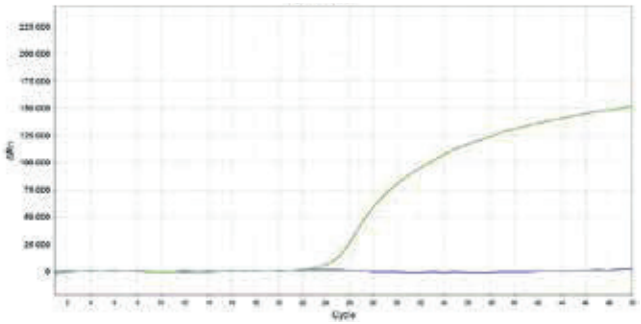
The patient referred by the corresponding health specialist (haematologist), and taking into account the compatibility of the symptoms presented; joint pain, abdominal pain, fatigue, weakness, heart failure, liver failure, bronze-coloured skin, and/or his family history (for example, a direct ascendant diagnosed with hereditary hemochromatosis) may be subject to the determination of the mutation in the *HFE* gene.

Patients who can benefit from this determination are those referred by a specialist. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

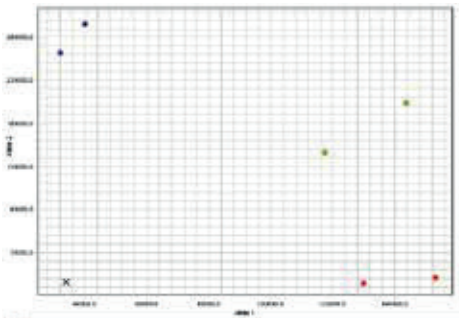
Results

Homozygous wild-type sample



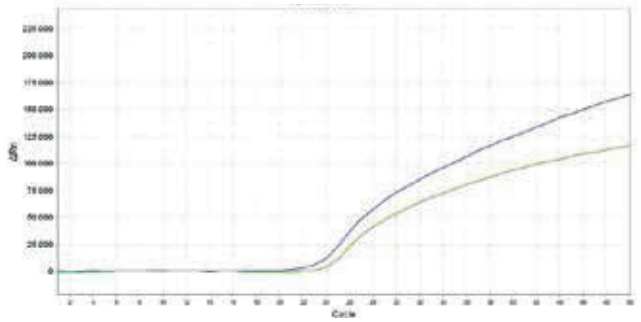
Wild-type allele Mutant allele

Allelic Discrimination Plot



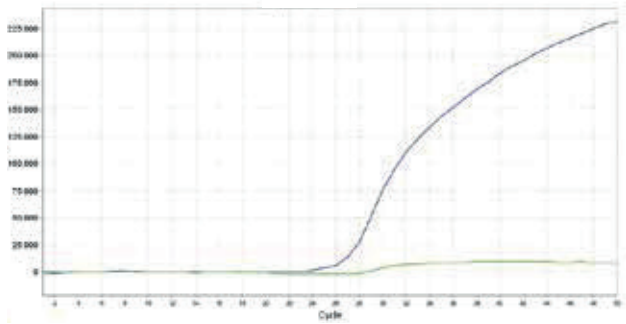
wt / wt mut / mut
wt / mut Reaction Blank

Heterozygous sample



Wild-type allele Mutant allele

Homozygous mutant sample



Wild-type allele Mutant allele

Genvinset®
H63D-C282Y multiplex

Intended use

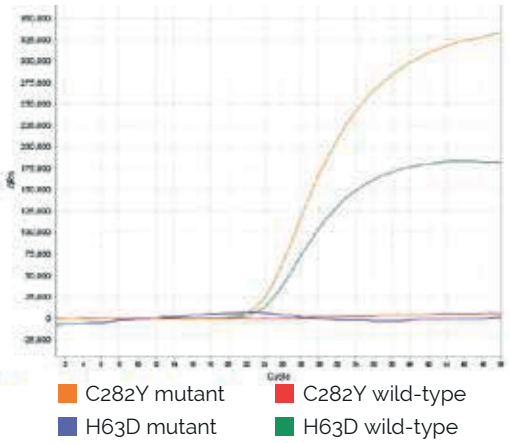
Genvinset® HFE multiplex is a semi-automated *in vitro* diagnostic kit qualitative detection of the C282Y mutation (NCBI dbSNP rs1800562; NM_000410.4:c.845G>A) and H63D mutation (NCBI dbSNP rs1799945; NM_000410.4:c.187C>G) associated with hereditary hemochromatosis, in the *HFE* gene (OMIM: 613609) in genomic DNA extracted from whole blood using Real-Time PCR technology with specific TaqMan® probes.

The patient referred by the corresponding health specialist (haematologist), and taking into account the compatibility of the symptoms presented; joint pain, abdominal pain, fatigue, weakness, heart failure, liver failure, bronze-coloured skin, and/or his family history (for example, a direct ascendant diagnosed with hereditary hemochromatosis) may be subject to the determination of mutations in the *HFE* gene. The results of this test should not be the only ones on which the therapeutic decision is based and should be used as an aid in the diagnosis together with results of other markers of the disease.

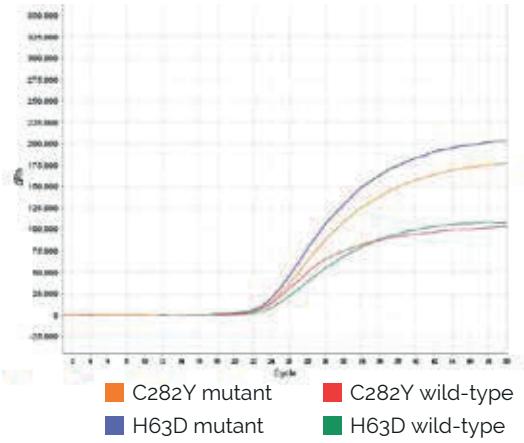
The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

Results

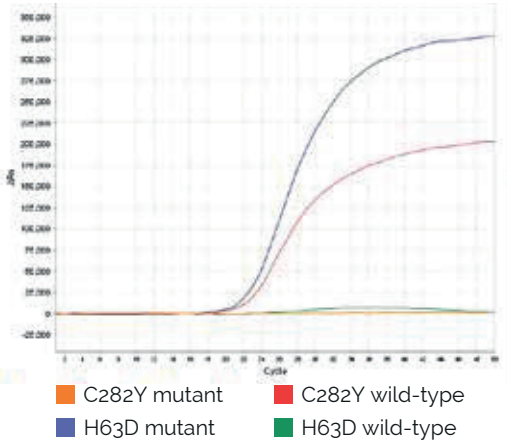
Homozygous wild-type H63D and homozygous mutant C282Y sample



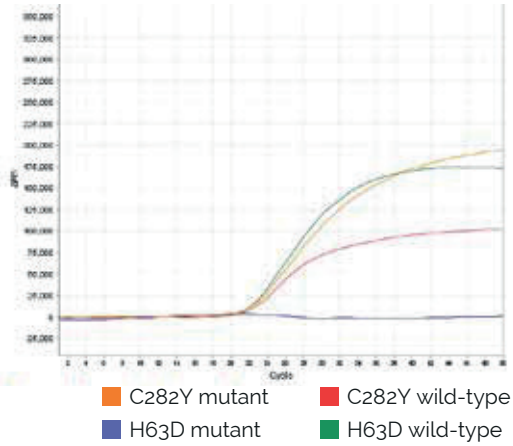
Heterozygous H63D and C282Y sample



Homozygous mutant H63D and homozygous wild-type C282Y sample



Homozygous wild-type H63D and heterozygous C282Y sample



Product Information

HFE H63D

48 tests - Code NLM AA1605/48
CE-IVD certified

DESCRIPTION: Genvinset® HFE H63D Real Time

HFE C282Y

48 tests - Code NLM AA1607/48
CE-IVD certified

DESCRIPTION: Genvinset® HFE C282Y Real Time

HFE S65C

48 tests - Code NLM AA1606/48
CE-IVD certified

DESCRIPTION: Genvinset® HFE S65C Real Time

HFE multiplex

24 tests - Code NLM AA1741/24
48 tests - Code NLM AA1741/48
CE-IVD certified

DESCRIPTION: Genvinset® HFE multiplex, 24T
Genvinset® HFE multiplex, 48T

Limitations

- Mutations or polymorphisms at annealing primer/probe sites are possible and may result in the lack of allele definition. Other technologies could be necessary to resolve the genotyping.
- Data and result interpretation should be revised by qualified personnel.
- This product is an auxiliary tool for the diagnosis of patients with suspected hereditary hemochromatosis. Use these results in conjunction with clinical data and results of other tests performed on the patient.

EXCLUSIVE DISTRIBUTOR
Nuclear Laser Medicine srl
Viale delle Industrie, 3 - 20049 Settala (Milan), ITALY
Phone +39 02 952451 - Fax +39 02 95245237-8
VAT 08763060152
segreteria@nlm.it - **www.nlm.it**