DNK s.r.o.
diagnostické centrum

Laboratory diagnosis of hereditary hemochromatosis

Diagnostic kits for detection of SNP polymorphisms associated with the development of hereditary hemochromatosis

Hereditary hemochromatosis

Iron overload (hemochromatosis) is a disease characterized by excessive accumulation of iron in tissues with subsequent damage and functional failure of organs. Hemochromatosis may be acquired or hereditary.

HFE – hereditary hemochromatosis gene

Hereditary hemochromatosis – autosomal recessive disease common in people of European descent. The disease is caused by mutations in the genes responsible for stimulating the expression of the protein hepcidin, the main regulator of iron metabolism.

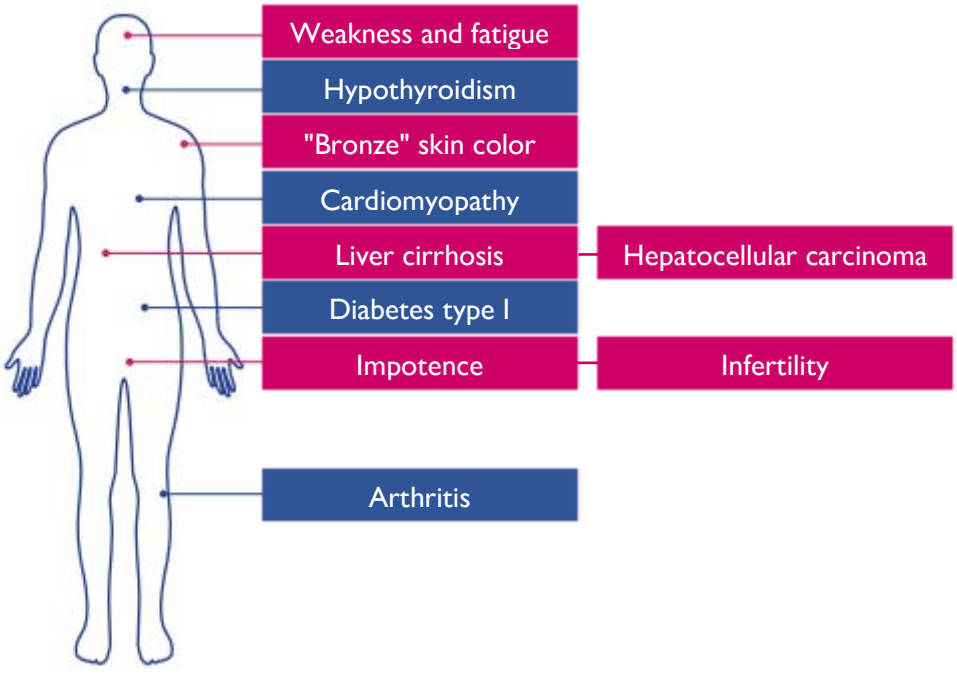
The cause of 85-90% of cases of the disease is a homozygous mutation of **Cys282Tyr** (HFE: 845 G/A) in the hereditary hemochromatosis gene (*HFE*). Less common (10-15%) are the heterozygous mutations **His63Asp** (HFE: 187 C/G) and **Ser65Cys** (HFE: 193 A/T).

The product of the HFE gene controls the expression of hepcidin in response to transferrin saturation with iron.
A mutation in the HFE gene leads to a lack of hepcidin with consequent excessive accumulation of iron in target organs.

Excessive accumulation of iron leads to the development of various pathologies

Frequency of occurrence among the Eurasian race

The ratio of men and women with hereditary hemochromatosis



Indications for molecular-genetic testing of mutations in the HFE gene

- deviation of laboratory tests: increase in ferritin and decrease in serum transferrin
- hereditary predisposition to hemochromatosis
- presence of clinical signs, e.g. hyperpigmentation of the skin (bronze skin colour), liver cirrhosis, diabetes mellitus

As a result of the gradual accumulation of iron, obvious clinical signs of the disease usually develop at the age of 40-60 years, when the iron level reaches critical values and leads to serious organ damage. Early diagnosis helps prevent the development of serious pathologies by timely monitoring and reducing iron levels in the body (through diet or appropriate therapy).

Diagnostic kits for detection of gene mutations in the HFE gene by real-time PCR method with melting curve analysis

Diagnostic kits can be used

- to confirm the hereditary nature of hemochromatosis and thereby exclude the secondary nature of iron overload
- for early diagnosis of hereditary hemochromatosis in case of family history

Features and advantages of the diagnostic kit

- Ready Master Mix for PCR: simplification of analysis procedures and high stability of test quality
- Specimens: whole blood or buccal epithelium
- Number of tests: the kits are intended for the analysis of 48 samples, including control samples
- Compatible devices: CFX96 (Bio-Rad, USA), DT-96 a DTprime (DNA-Technology, Russia), Gentier 96E/R (Xi'an TianLong, Science and Technology Co., Ltd., China)
- High stability of the kit: storage at a temperature of 2–8 ° C; transport up to 26 °C for not more than 10 days

Cat. №	Kit name	Number of tests
3822 CE	**RealBest-Genetics Hemochromatosis (HFE 187 C/G)**	48
3823 CE	**RealBest-Genetics Hemochromatosis (HFE 193 A/T)**	48
3824 CE	**RealBest-Genetics Hemochromatosis (HFE 845 G/A)**	48

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