TEST SYSTEMS FOR REVEALING Cys282Tyr, His63Asp, AND Ser65Cys ALLELES OF THE HFE GENE ASSOCIATED WITH INHERITED HUMAN HEMOCHROMATOSIS

Description
Inherited hemochromatosis (IH) is a human in-born aberration of ferrum metabolism that leads to unregulated accumulation of ferrous pigments in liver, endocrine glands, joints and muscles. In the case of untimely diagnostics the pathology leads to arthritis, myocardiac defects, diabetes, hypogonadism, aberrations in skin pigmentation and, finally, to cerosis and carcinoma of the liver. The disease can be easily treated by bloodletting at early stages preceding irreversible involvement of viscera. Early diagnostics of hemochromatosis or prediction of predisposition are very important. Common clinical methods fail to do that: biochemical blood analyses for ferrum content are not precise; the results may considerably deviate in patients depending on the consumption of alcohol, presence of infections, etc. Liver biopsy reveals the disease at a relatively late stage. Moreover, this method involves a certain risk for a patient. It was established that worldwide from 50 to 100% of people suffering from IH are carriers of the Cys282Tyr allele of the HFE homozygous gene and mixed heterozygotes on the Cys282Tyr and His63Asp alleles, as well as Cys282Tyr and Ser65Cys. Besides, a higher frequency of the His63Asp allele was noted in the samples of patients with ferrum overload syndrome, sporadic skin porphyria, and some other diseases. Test systems for revealing these alleles in human genome based on the method for polymorphism of the length of restriction fragments have been worked out at the Institute of Cytology and Genetics.

Technical appraisal and economic benefits
The suggested test systems provide a unique opportunity for detecting the disease at early stages and predicting the predisposition to it.

Application areas
Medicine.

Development stage
The test systems are being successfully used at the hospital of the Scientific-Research Institute of Therapy, at the Novosibirsk Regional Diagnostic Centre (Novosibirsk), at the Hematological Research Center (Moscow), and at the Central Clinical Hospital of the Ministry of Communications (Tambov).

Patent situation
RF patent No. 2144566.

Commercial offers
License agreement, test system sales, performance of tests.

Estimated cost
To be negotiated.

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