testing lipoarabinomannan in the urine as a prognostic marker in screening TB / HIV-infected individuals, persons with latent form of TB and progressing in the active form of TB with initiation of therapy as soon as possible with medicine against TB including cost effectiveness for patients’ clinical LAM investigation.

References.


3. Lawn SD, Gupta-Wright A. Detection of lipoarabinomannan (LAM) in urine


Key words: Lipoarabinomannan (LAM), TBC, M. tuberculosis, TB LAM.

DOI: 10.29256/v.03.01.2019.escbm21

**CLINICAL CASE OF COMBINED GENETIC PATHOLOGY IN A PATIENT**


I.M. Sechenov First Moscow State Medical University, Russia

Familial hypercholesterolemia (FH) is a form of genetically determined increase of lipid levels in blood associated with a high risk of cardiovascular disease in young age. Disease is usually diagnosed only when the first symptoms of atherosclerosis appear, including acute cardiovascular disorders; that particularly complicates diagnosis and prevention in patients without clinical signs [1, 2]. Gilbert’s syndrome is a hereditary benign hyperbilirubinemia associated with a mutation in the UGT1A1 gene and a decrease in the functional activity of the enzyme uridine diphosphate glucuronidase (UGT) 1A1 in hepatocytes. The UGT gene is located in the long arm of chromosome 2 (q 37.2). More often there is an increase in the number of TA-repeats, which reduces the activity of the enzyme. The incidence of this syndrome in different regions of the world ranges from 3 to 10 % [3, 4]. Genetic testing of patients with familial hypercholesterolemia is not included in the routine examination, but it can be recommended to first-line relatives for timely diagnosis and assessment of the risk of cardiovascular diseases [5]. Demonstration of clinical observation of genetic combination of familial hypercholesterolemia and Gilbert’s syndrome in a young patient and analysis of possible protective role of increased bilirubin level in atherosclerosis progression in Gilbert’s syndrome.

**Materials and Methods.** This paper describes a clinical case of a combination of two genetic diseases in a 24-year-old male patient: familial hypercholesterolemia and Gilbert’s syndrome using analytical, logical, monographic methods, the results of anamnesis and objective examination of the patient, methods of laboratory, functional, ultrasound diagnosis and genetic testing.

**Results.** During genetic testing, a mutation R3500Q of apoB 100 gene was detected, which leads to a violation of low-density lipoprotein (LDL) binding, their accumulation...
in the bloodstream, and an increase in the number of TA-repeats in the homozygous state (7 repeats) of the uridine diphosphate-glucuronyltransferase gene, which violate the functional activity of the enzyme involved in bilirubin conjugation. Mutation of apoB 100 gene R3500Q along with LDL receptor mutations is one of the causes of high cholesterol in Russian patients. A search for a causal mutation in the apoB gene in the region encoding the receptor-binding domain of the protein revealed a single nucleotide substitution in codon 3500, resulting in the amino acid arginine being replaced by the amino acid glutamine. The R3500Q mutation is the most common familial defect. Another missense mutations causing a family hypercholesterolemia, such as R3480W, R3500W and R3531C are much rarer [6, 7]. On the basis of the research the patient’s diagnosis was formulated: Dyslipidemia Fredrickson Type IIa (mutation R3500Q) and benign hyperbilirubinemia of the Gilbert’s syndrome type. The patient is recommended to follow the principles of healthy nutrition, graduated exercise, the exclusion of alcohol, reducing insolation. In order to reduce the risk of cardiovascular disease the patient was prescribed therapy with lipid-lowering drugs (Statins), which showed a positive effect (reduction of total cholesterol and LDL by 32% and 38%) according to the results of blood tests in the dynamics.

Prospects for further research. This case is significant because the combination of familial hypercholesterolemia and Gilbert’s syndrome in clinical practice is rare. The possible protective properties of increased bilirubin levels in the development of cardiovascular diseases are interesting and not fully elucidated. Early detection, treatment and availability of genetic testing of first-line relatives of patients with familial hypercholesterolemia play a key role. Long-term therapy of patients with familial hypercholesterolemia significantly reduces or eliminates the excess risk of coronary heart disease, persisting throughout the life of the patient, thereby reducing the risk in the general population.

References.


Key words: gene mutation, genetic testing, familial hypercholesterolemia, Gilbert’s syndrome.