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ETIOPATHOGENETIC AND DIAGNOSTIC CRITERIA FOR GILBERT'S SYNDROME

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Abstract
Gilbert's syndrome is characterized by episodes of jaundice and increased unconjugated bilirubin in the blood serum. This syndrome is usually a relatively harmless condition and does not pose a serious health threat. However, sometimes, especially with other existing diseases or an incorrect attitude towards one's health, complications may arise in the form of stone formation in the gallbladder. In addition, there is an increased risk of complications after some surgical procedures, especially those involving the biliary tract, and there are problems with the absorption of certain medications, which can complicate the treatment of concomitant diseases. Frequent exacerbations significantly reduce the patient's quality of life. The article is devoted to consideration of etiopathogenetic factors, clinical symptoms, diagnosis and treatment of Gilbert's syndrome.

Keywords: Gilbert's syndrome, etiopathogenetic factors, clinical symptoms, dysfunctional disorders of the biliary system, diagnostic criteria, therapeutic approach.

Introduction

Gilbert's syndrome is a hereditary disease associated with a predominant violation of the uptake and conjugation of bilirubin, which is based on a genetically determined persistent increase in the content of indirect bilirubin in the blood serum.

The direct cause of the development of Gilbert's syndrome is a disorder of bilirubin metabolism caused by a mutation in the UGT1A1 gene, which encodes the enzyme uridine diphosphate - glucuronyl transferase. This protein works in hepatocytes, and its main function is the conversion of bilirubin.

The first to draw attention to the unusual condition in some young patients was the French doctor Augustin Gilbert. Such patients developed jaundice, which then went away on its own. And in 1901, Gilbert described this disease, from which the syndrome got its name.

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From 7% to 10% of the globe suffer from this pathology. Most often, the disease is diagnosed in adolescents, which is associated with the effect of male sex hormones on the metabolism of bilirubin. In men - more often than in women. The disease is detected in Africans more often than in Europeans [1, 6, 11].

The disease is transmitted in an autosomal recessive manner, i.e. it develops in people who inherited a certain defect of the second chromosome from both parents: in the place that is responsible for the formation of one of the liver enzymes, glucuronyl transferase. As a result, the enzyme content is reduced to 80% [1, 14]. Glucuronyl transferase is involved in the metabolism of bilirubin. In conditions of deficiency of this enzyme, indirect bilirubin cannot be associated with the glucuronic acid molecule in the liver, which leads to its increase in the blood. Indirect bilirubin is a toxic substance for the body and its neutralization is possible only by converting it into direct bilirubin in the liver. The latter is excreted from the body along with bile. This chromosomal defect often makes itself felt only starting from adolescence, when the metabolism of bilirubin changes under the influence of sex hormones. Due to the active influence of androgens on this process, Gilbert's syndrome is recorded more often in the male population [2, 8, 17].

Typically, the syndrome does not develop out of nowhere, since 20–30% of uridine phosphate - glucuronyl transferase meets the body's needs under normal conditions. The first symptoms of Gilbert's disease appear after: fasting or, conversely, overeating, eating fatty foods; taking certain medications; drinking alcohol; excessive physical activity; stress; operations, injuries; colds and viral diseases. These same factors aggravate the course of the disease and provoke its relapses [1, 5, 10].

As a rule, Gilbert's syndrome is asymptomatic or with minimal clinical manifestations. Therefore, many experts consider it not as a disease, but as a physiological feature of the body.

In most cases, the only manifestation of the syndrome is moderate jaundice (staining of the skin, mucous membranes, and whites of the eyes yellow. Other symptoms are extremely rare and mild [12, 15].

Neurological symptoms are minimal, but may include: increased fatigue, weakness, dizziness, insomnia, and sleep disturbances. Even more rare symptoms include symptoms of dyspepsia: decreased or lack of appetite, bitter taste in the mouth, bitter belching after eating, heartburn, nausea, rarely vomiting, constipation or diarrhea, bloating, feeling of fullness in the stomach, discomfort and pain in the right hypochondrium. Sometimes there is an increase in the size of the liver [3, 4, 9].

With additional examination methods, a clinical blood test reveals high hemoglobin, an increase in red blood cells, and possible reticulocytosis.

In a biochemical blood test, changes in ALT, AST, thymol test, and bilirubin can be observed.

The diagnosis is confirmed by a genetic analysis for Gilbert's syndrome (a study of human DNA obtained from venous blood).

Thus, the diagnosis is made on the basis of the following data: onset of the disease in prepubertal and pubertal age, hereditary predisposition, low intensity of jaundice, chronic (wavy) course, appearance or intensification of jaundice due to intercurrent diseases, fasting, insolation, physical or psycho-emotional stress, taking alcohol and a number of medications. Hepatomegaly is absent or insignificant, bilirubin increases 2-5 times mainly due to the free fraction, normal ALT/AST activity, markers of viral hepatitis are negative, there is no evidence of hemolytic anemia, the result of genetic analysis is positive.

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In general, the disease proceeds favorably, without causing unnecessary inconvenience and anxiety, but with frequent exacerbations of the disease, the development of some complications is possible, such as: dysfunctional disorders of the biliary tract, cholelithiasis, sludge syndrome. Also, indirect bilirubin plays a key role in the formation of gallstones, being the nucleus for the formation of pigmented and mixed stones.

The main goals of therapeutic intervention for Gilbert's syndrome are: reducing the level of bilirubin to reduce intoxication and the effect on the central nervous system, as well as to prevent the development of complications.

There is no etiotropic or pathogenetic treatment. The condition can only be alleviated or the symptoms removed. To do this, restrictive treatment is used: adherence to the regime, exclusion of provoking factors, adherence to a diet, table No. 5. Also strictly prohibited: fresh baked goods, lard, sorrel, spinach, fatty meat, fatty fish, mustard, pepper, ice cream, black coffee, alcohol, etc.

To relieve the symptoms of jaundice, inducers of enzymes of the monooxidase system of liver cells are taken: phenobarbital and zixorine for 2-4 weeks. They allow you to reduce the level of bilirubin in the blood and get rid of dyspeptic symptoms. To enhance the excretion of bilirubin from the intestine, enterosorbents are used. Phototherapy is the destruction of bilirubin fixed in tissues by exposure to light, usually blue lamps. Eye protection is required to prevent burns. Hepatoprotectors are agents that protect liver cells from damaging influences. Drugs that normalize the function of the gallbladder and its ducts: ursodeoxycholic acid preparations. For dyspeptic disorders, antiemetics and digestive enzymes are used. Vitamin therapy [2, 7, 13, 16].

Thus, the following conclusions can be drawn:

1. Gilbert's syndrome is a hereditary disorder of bilirubin metabolism, the timely recognition and correction of which is essential.
2. The modern stage of development of medicine has made it possible to objectively confirm the diagnosis of Gilbert's syndrome using genetic methods.
3. The benign nature of the syndrome does not exclude such adverse consequences as diseases of the biliary tract, including cholelithiasis.
4. To prevent and treat these adverse effects, it is advisable to use ursodeoxycholic acid.
5. Since the disease is hereditary, married couples where at least one of the spouses suffers from this disease should consult a geneticist before planning a pregnancy.

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