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THE CAUSES OF THE OCCURRENCE OF MUCOVISSDOSIS IN CHILDREN, MEASURES FOR THE DETECTION AND TREATMENT OF THE DISEASE

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ANNOTATION: Today, due to various disorders in the genetic material, hereditary diseases are becoming an increasingly important issue. This is mainly due to the accumulation of retsessive mutations in the autosome chromosomes of the population. Mucovissidosis is one of the chronic diseases of a hereditary nature, the main feature of which is the viscosity of the secretion of gland cells and the continued growth of affected structures and organs with secondary muscle degeneration.

Key words:autosom ,mucovissidosis ,enzyme, gene, mutation, microzyme

Today, due to various disorders in the genetic material, hereditary diseases are becoming an increasingly important issue. This is mainly due to the accumulation of retsessive mutations in the autosome (nojunction) chromosomes of the population. One of such diseases is mucovissidosis. It is sometimes also called cystic fibrosis, that is, congenital enterobronchopancreatic dysuria, called Salty child syndrome. Mucovissidosis is the most common pathology among all monogen hereditary diseases of nature. And, basically, europeoids are threeraydi in representatives of the race. The prevalence of the disease is different in different regions, with an average of 2 - 4,5 among newborns, the state of the disease can be encountered. In this regard, there is a need for strategic recommendations based on the latest and most complete evidence for the treatment and care of patients with mucovissidosis. Currently, 176 patients with confirmed diagnosis of mucovissidosis have been registered in Uzbekistan, the most part of which belongs to Tashkent City and Tashkent region. The average age of patients with mucovissidosis is 5.2 years. Their life expectancy in the Republic of Uzbekistan is 21 years (176/2). Mucovissidosis is one of the chronic diseases of a hereditary nature, the main feature of which is the viscosity of the secretion of gland cells and the continued growth of affected structures and organs with

secondary muscle degeneration. Mucovissidosis is one of the proven diseases of hereditary nature. The cause of all occurring disorders is a gene mutation in the middle of the long shoulder of the 7 somatic chromosome. It is called a cystic fibrosis transmembrane regulator, which means that it is considered a transmembrane regulator of the permeability of sodium and chlorine ions. This gene encodes a certain protein structure, which is the basis of chlorine and other ion channels contained in the epithelium. Therefore, the main moment of the disease is a violation of the transport function of the damaged cell membranes with the predominance of the glands of external secretion. But the organs of internal secretion do not participate in the pathological process. The functioning of the cystic fibrosis transmembrane regulator gene does not have a time limit, it works at all stages of life. But the onset of symptoms and their severity can be different. It basically depends on the type of mutation. Currently, its more than 1600 variants are known, and the frequency of occurrence of each of them differs in different regions of the Earth's globe. Cystic fibrosis is a transmembrane regulative mutation retsessiv. This means that in a person with a normal gene, in paired chromosomes, a person with a normal gene has no symptoms of mucovissidosis. There are a lot of such people among the population. They are carriers of the disease and can transfer the pathological gene to their children. Gene transfer does not depend on the sex, so the probability of the disease in boys and girls is the same. The child will be sick with mucovissidosis, having acquired only 2 defective genes. This is most likely, if ikala has at least one chromosome in which both parents have a mutation in the corresponding field. The probability of the birth of a sick child can be different in this case:

The mother is healthy (both chromosomes are mutated) the disease-spreading mother (one mutant gene, another normal) the mother is sick with mucovissidosis (both genes are mutant).

If the father is healthy (both chromosomes are mutated) in 100% of cases, the probability of the birth of a child distributing a healthy disease is 50%, the probability of the birth of the distributor is 50%, the probability of the birth of a sick child is 0%, all children will be the distributor of the disease.

If the probability of the birth of a child is 50%, the probability of the birth of the distributor is 50%, the probability of the birth of a sick child is 25%, the probability of the

birth of a healthy child is 25%, if the probability of the birth of a sick child is 50%, then the probability of the birth of the

The father is sick with mucovissidosis (both genes are mutant) the probability of the birth of a sick child is 0%, all children will be a distributor of the disease. If the probability of the birth of a sick child is 50%, then the probability of the birth of the distributor is 50% that all children will be ill with mucovissidosis.

It should be noted that the probability of the birth of a child with mucovissidosis depends only on the presence of an anomalous cystic fibrosis transmembrane regulator gene in parents. The nature of mutations does not matter. With the participation of two anomalous cystic fibrosis transmembrane regulative genes, a functional incomplete protein of the chlorine-ion transmembrane channel is synthesized in a person. This leads to a cascade that is irreversible and prone to progressive diseases: interruptions with the accumulation of chlorine ions in the cytoplasm of the tube cells. This leads to a change in the membrane potential and worsens the work of other ion channels, mainly the sodium channel. An increase in the concentration of sodium ions in the cells leads to the fact that the cells are more actively taken from nearby water. Since mucovissidosis affects the cells of the gland, the liquid is already absorbed from the secretions released by them. This process is not compensated in any way and leads to irreversible changes in the organs of the second degree. Excessively thick and sticky secretions lead to stagnation, which leads to clogging of the excretory ducts, vessels, bronchi. The inflammation that accompanies it, the adhesive process and aggravates the situation until the development of a rough wound. Channels remain in a state that can not pass gradually. Stagnation of secretion leads to inflammation of the gland organs and surrounding tissues. Therefore, most often in mucovissidosis the following cases occur: pancreatitis (melting of the pancreas with its own enzymes), holangogepatitis (inflammation of the bile ducts and liver), bronchopneumonia (as a result of the closure of the bronchi channels) were detected. Secondly, non-gland organs interfere with the pathological process. For example, the pathology of the bronchi lungs leads to a violation of the cardiovascular system. Deficiency of the enzyme caused by pancreatitis and inflammation of the intestinal walls causes a decrease in the absorption of nutrients, iron and vitamins. In acute disorders of the disease, all organs and, in particular, the brain of the

head are affected, and this is especially dangerous in early childhood. In the case of mucovissidosis, all exocrine gland tumors, regardless of their size, are affected. But the appearance of pathological changes in different organs is usually different. In this regard, several clinical forms of the disease are distinguished: the closure of the Meeconial intestinal tract. It now develops in the first days in babies born, and in them it is expressed mainly by thickening the primary feces (mekonia). Bronchial lung form associated with obstruction of parts of the respiratory system (obstruction with mucus) and violation of bronchial epithelial activity of the glands. Manifested through bronchopneumonia davriy with chronic obstructive disease of the lungs and the formation of bronchoectasis . On the development of intestinal form and enzyme deficiency include lesions of the pancreas. The connection of the colon to the opening of the gastrointestinal tract is billiards liver cirrhosis. Also, very "soft" forms of the disease are threeraydi, that is, the existing violations do not roughly disrupt the quality of life and do not create life-threatening conditions. For example, in men, mucovissidosis may appear in the form of an isolated obstructive azospermia infertility due to the obstruction of the seed pathways. In addition, there are abortive forms, including diseases of the sweat glands, sinusitis, chronic pancreatitis, which do not form cysts, etc. In clinical practice, the terminology of the International Classification of 10 diseases is used. Mucovissidosis Ye has 84 cipher, the cipher is classified with diseases of the lungs, intestines and other organs. There is also a concept of "unspecified mucovissidosis". In this case, the doctor will have to encrypt only the most severe cases, since in 70% of cases a mixed (pulmonary) form of the disease is manifested. At the time of diagnosis, they are described as already developing complications, indicating the degree of rejection of the existing disease. In children, mucovissidosis begins in the first years of a child's life, in cases from 90 to 94% of cases. Sometimes its symptoms are found in newborns and grow for several days. The main symptoms of mucovissidosis in childhood are: intestinal disorders due to enzyme deficiency. In most cases, they appear when introducing complementary foods or complementary foods. In the vast majority of cases, with a large amount of fat in the composition is often observed malignant feces and abdominal swelling. Soon anemia associated with hypopolivitaminosis and iron deficiency develops. There is a high probability of the appearance of diabetes mellitus. Predisposition to recurrent long-term complex

pneumonia. Difficult to swallow food, it depends on the extreme stickiness of the saliva and the dryness of the digestive tract and esophagus. The child is inclined to drink food. Violation of thermoregulation in a warm room in the warm season. This is due to a violation of sweating. Intestinal and bronchopulmonary forms of the disease in childhood lead to a delay in the physical development of the child, the formation of chronic polyorgenic insufficiency. Ualal depression is not uncommon, but certain metabolic disorders can lead to poor functioning of the brain. It should be noted that the therapy prescribed for mucovissidosis does not allow to cure the disease. It only helps to eliminate diseases, improves the quality of life of the patient, facilitates symptomatology, helps to eliminate serious complications and reduces the risk of their development. Gene therapy, which affects the defective gene, is still in development and is undergoing clinical trials. Enzyme preparations are very important, especially when the intestine is sick. Multivitamin complecs and especially vitamins are important to compensate for the lack of absorption in the intestine. The use of enzyme preparations in mucovissidosis can partially eliminate the deficiency of digestive enzymes. Such digestive disorders are noted in people with almost all this disease, since the work of the pancreas is its usual manifestation. Special attention here requires correction of the level of lipase - that is, an enzyme that allows sufficient digestion of fats in the small intestine. For this purpose, in the disease of mucovissidosis, a microsyma is appointed. Previously, with the manifestation of mucovissidosis, mainly pediatricians collided, and this was mainly considered threeraydi in children. Currently, this pathology is often encountered in adults, which is explained by the possibilities of modern advanced pharmacology. The drugs used can partially compensate for the impaired secretory functions of the mucous membrane of the bronchial tree and pancreas. With the chosen therapy desired, the child with mucovissidosis is able to grow healthy.

Used literature:

1.Maksudov.Y. Z. "General genetics". T. Teacher, 1980.

2.B. Zire " Lost In Test Match T. Selomov, M. N. Abdullayeva " Children's diseases" T.:Medical publishing house named after Abu Ali ibn Sina, 1997 .

3. Hadsan M., Geddes D., Bush A. cystic fibrosis. London: Huddi Arnold. 2007.

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4.Tatochenko V. K., Rachinsky S. V., Volkov I. K., Fedorov A. M.Childhood practical pulmonology: a guide. Moskow.2007

5. Kapranov N. I., Kashirskaya N. Y. cystic fibrosis. Modern achievements and existing problems. The method. recommendations. Hilalazmi.ad the 4. Moskow

6. Simonova O. I. Complex treatment of children with cystic fibrosis: recommendations for a pediatrician. Children's pharmacology. 2006.

7. Baranov A. A., Valadin N. N., Samsygina G. A. National pharmacotherapy of children's diseases. Volume. 1. M.: Littera. 2007.