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ALBINISM DISEASE - CLASSIFICATION, DIAGNOSIS, SYMPTOMS AND TREATMENT MEASURES

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Abstract: Albinism is a group of hereditary pathologies characterized by a violation or complete absence of skin, hair, eye pigmentation. The main symptoms of the disease are very pale skin and hair, blue or red eyes, and in some cases, vision impairment. The diagnosis of albinism is made based on the current condition of the patient, as well as genetic tests. To date, there is no specific treatment for albinism, palliative therapy (correction of vision) is used, and there are a number of recommendations for patients to behave in the sun, protect the skin and reduce the likelihood of complications.

Key words: albinism, melanin pigment, Chediak-Higashi syndrome, Germansky-Pudlak disease, tyrosinase, mutation,

General information

Albinism is a set of genetic pathologies in which the processes of formation or accumulation of melanin pigment in skin cells, its appendages, iris and retina are disturbed. This condition has been known since ancient times and affects people of any nationality or race. However, the frequency of occurrence of albinism varies in different nations - it ranges from 1:10,000 to 1:2,000,000. Also, in recent years, this indicator is not the same for various forms of the disease classified according to genetic characteristics. Previously, only two forms of albinism (ocular and skin-ocular) were distinguished, although modern genetics distinguishes at least seven different types of

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pathology. In addition, there are a number of hereditary diseases, the complex of symptoms of which includes, among other things, albinism - for example, Chediak-Higashi syndrome, Germansky-Pudlak disease.

Causes of albinism

The main reason for the development of albinism is a violation of the metabolism of tyrosine amino acids and, as a result, a complete blocking or weakening of the synthesis and deposition of melanin pigment. This condition can lead to various mutations in genes directly or indirectly involved in the process of melanin formation. For example, the most severe form of albinism, eye-skin 1A, is caused by a complex mutation in the alb-OCA1 gene, located on chromosome 11. It encodes the sequence of the enzyme tyrosinase, and with nonsense mutations, its production in the body stops completely. As a result, the production of melanin also stops completely, which is the cause of severe skin-eye albinism. This condition is inherited by an autosomal recessive mechanism.

Another type of this disease, oculocutaneous albinism 1B, is caused by a disruption of the same alb-OCA1 gene, but its activity continues. More than 50 mutations of the above gene are associated with this pathology, and each of them affects the activity of tyrosinase to a different degree. Therefore, the severity of symptoms of cutaneous-ocular albinism 1B is also very variable - from the almost complete absence of melanin in the tissues to a slightly lighter color of the skin and hair. In some cases, patients with this form of pathology can look at the sunset, their hair darkens with age, and pigment spots can appear. An interesting subtype of this disease is temperature-sensitive albinism, in which tyrosinase activity decreases sharply at temperatures above 37 degrees. This causes pigmentation to appear more on colder parts of the body - hands, feet. Head, eyes, armpits often remain without pigment.

Type 2 cutaneous-ocular albinism is the most common type of this pathology. However, in this case, genetic diseases do not affect the synthesis of tyrosinase, which remains at a sufficient level, and the activity and structure of the enzyme are not affected. This type of albinism is caused by a mutation of a gene located on chromosome 15. It is thought to encode a melanosomal membrane protein (P-protein) responsible for tyrosine transport. Melanin deficiency in this form of albinism is also highly variable, and pigmentation may increase over time. The reasons for this incident

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have not yet been determined. Type 2 cutaneous-ocular albinism is inherited in an autosomal recessive manner.

Another form of the disease, type 3 cutaneous-ocular albinism, occurs almost exclusively in the Negroid race. Under it, genetic studies revealed mutations in the TRP-1 gene located on chromosome 9. A similar gene in mice is responsible for the brown color of the coat, but its function in humans is not clearly known. It controls the formation of the black fraction of melanin (eumelanin), and it is assumed that the disruption of its structure leads to the superior synthesis of the brown pigment variety. Type 3 albinism, like other skin-eye forms, is transmitted through an autosomal recessive mechanism.

Each type of albinism is characterized not only by the loss of melanin from the skin and its appendages, but also by the visual apparatus of the eye - the iris and the pigment layer. This leads to refraction and transparency of the cornea, astigmatism and strabismus, foveolar retinal hypoplasia. There are forms of albinism (called eye types), which are characterized only by damage to the organs of vision. The most common form of ocular albinism is recessive and is linked to the X chromosome. It is caused by a mutation in the GPR143 gene, which encodes the receptor for G-proteins of eye melanocytes. As a result, the formation processes of melanosomes are disturbed, which causes the development of ocular albinism. In 1970, an autosomal recessive form of this disease was also identified, but the pathogenesis of this type was not yet determined - some (14%) of such patients had alb-OCA1 gene mutation, while others (36%) had disorders. P- gene, squirrel. In almost half of patients with autosomal recessive ocular albinism, it was not possible to determine the genetic cause of the disease.

Classification

Previously, all cases of albinism were divided into only phenotypic manifestations - complete and incomplete. The first includes all types of cutaneous-ocular albinism, characterized by severe pigmentation disorders of the eyes, skin and its appendages. Incomplete forms include eye types of the disease, as well as types of pathology that lead to skin staining. Currently, genetic classification is more widely used, among which the following types of albinism are distinguished:

• Cutaneous-ocular albinism type 1A is caused by a nonsense mutation of the alb-OCA1 gene, which simply "turns off" its activity. As a result, the synthesis of tyrosinase in the body stops completely.

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• Ocular albinism type 1B - as in the previous case, it is caused by mutations in the alb-OCA1 gene, but it can be expressed. As a result, a defective tyrosinase enzyme with different levels of activity is synthesized. The severity of such albinism depends on the type of gene mutation.

• Temperature-sensitive oculocutaneous albinism is type 1B characterized by temperature-dependent variable tyrosinase activity. Skin manifestations may be pale, and ophthalmic disturbances may be significant. These characteristics of such albinism depend on the high temperature of the eyes - therefore, tyrosinase in them is less active.

• Type 2 cutaneous-ocular albinism is caused by a mutation of the gene encoding P-protein, which is an element of the membrane of melanosomes inside the cell. As a result, the transport of tyrosine in the cell is disturbed, and melanin synthesis does not occur even with normal tyrosinase activity.

• Type 3 cutaneous-ocular albinism is the result of mutations in the TRP-1 gene, which probably controls the production of eumelanin. It occurs only in Africans, causes the development of brown skin and hair and mild ophthalmic diseases.

• Ocular albinism is recessive and linked to the X chromosome. It is caused by a mutation in the GPR143 gene, which is responsible for some elements of intracellular communication.

• Autosomal recessive ocular albinism - this has not yet been linked to specific genetic diseases. Some cases of this disease are assumed to be eye forms of the types of eye pathology - 1B and 2.

Even within the same genotype of albinism, there can be significant differences in the severity of symptoms. This is due to the fact that different types of mutations have different effects on melanin production.

Signs

The main manifestations of albinism include paleness of the skin, which is especially noticeable at the birth of a sick child. Most often, the skin is pink due to transparent blood vessels, the eyes are blue at birth, but in some corners they can also have a reddish color. Later, in the process of growth, the symptoms of albinism may differ slightly depending on the type of the disease. With the most severe type 1A, the synthesis of melanin in the body does not occur at all, so the patient retains white skin and hair and blue eyes throughout life. Albinism type 1B is characterized by a rapid

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accumulation of yellow pigment in the hair, so they take on a light straw color, often with age, pigmentation appears in the eyelashes and cornea of the eye.

Temperature-sensitive albinism is often manifested in a specific distribution of melanin - normal pigmentation is observed in the limbs, the scalp is pale, and the hair remains white. In such patients, the eyes become blue not in the extremities, but due to the increase in temperature. Type 2 albinism is also characterized by significant variability of symptoms - from almost complete absence of pigmentation to imperceptible lightening of skin and hair. Also, this form of the disease is often characterized by an improvement in the synthesis of melanin with age - the hair begins to darken, freckles appear, and a look at the sun appears. However, it is necessary to be careful when exposed to sunlight - the skin of patients with albinism is very sensitive to ultraviolet radiation, skin burns and photodermatitis appear easily.

A characteristic symptom of albinism is a violation of visual acuity in patients and other ophthalmic changes. The more pronounced the decrease in vision, the more melanin is synthesized in the body, especially in the cornea and the pigment layer of the retina. In addition, strabismus, astigmatism, and nystagmus, which appear immediately after birth or in the first years of life, are frequent companions of albinism. In the eye forms of the disease, similar symptoms appear without disrupting the pigmentation of the skin and hair. Due to the lack of a protective layer of melanocytes, patients with albinism often have photophobia, which sometimes turns into day blindness.

Diagnosis

In most cases, the diagnosis of albinism can be made immediately after the birth of the patient - a dermatologist evaluates the state of pigmentation of the skin and hair, can identify the disease and find out its type. Follow-up by this specialist is necessary to monitor the progress of the pathology and prevent possible complications - for example, skin cancer. An ophthalmologist with albinism often reveals the transparency of the iris, and in adult patients, retinal hypoplasia is often detected in the area of the macula. The foveolar reflex is sharply reduced or absent. In people with incomplete albinism, foci of depigmentation are often found in the fundus of the eye. Other visual disturbances are also found - nystagmus, astigmatism, myopia.

To confirm the diagnosis and determine the type of pathology, a geneticist can determine the sequence of genes associated with it. Also, collection of genetic history

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plays an important role, genetic diagnosis of the patient's relatives can identify carriers of defective genes. A rare and expensive method of diagnosing albinism is the detection of tyrosinase activity in tissues (for example, hair follicles), but this allows to clarify the prognosis of the disease somewhat. The better the activity of this pigment is preserved in the tissues, the lower the severity of other symptoms of this pathology.

Differential diagnosis of albinism should be carried out with other hereditary pathologies accompanied by similar skin and ophthalmological signs. First of all, these are Chediak-Higashi and German-Pudlak syndromes, X-linked ichthyosis, microphthalmia, Kallman's disease.

Treatment and prognosis

To date, there is no specific treatment for albinism, only preventive measures have been developed to improve the patient's quality of life. To maintain the current level of vision, it is necessary to protect the eyes from sunlight - this is achieved by wearing special sunglasses or contact lenses. Avoid exposure to the bright sun or protect the skin with special creams and lotions. If you follow these recommendations, in general, the prognosis for albinism is favorable - patients can live a long and happy life. This requires regular consultations with a dermatologist and ophthalmologist to prevent complications such as skin cancer or retinal detachment.

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