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Albinism, Cri Du Chat Syndrome, And Reilly Syndrome (CIPA): Features, Genetics, Diagnosis, And Treatment

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Abstract: This article discusses three rare genetic diseases — albinism, Cri du Chat syndrome, and Reilly syndrome (congenital insensitivity to pain with anhidrosis). Their main clinical manifestations, diagnostic methods, features of genetic transmission, and modern approaches to treatment are described. An overview of the impact of these diseases on the quality of life of patients and the prospects for medical care is presented.

Keywords: Albinism, Cri du Chat syndrome, Reilly syndrome, congenital diseases, genetics, diagnostics, treatment.

Introduction: The study of hereditary diseases such as albinism, Cri du Chat syndrome and Reilly syndrome is of great importance for medicine. These rare pathologies lead to serious disorders in human development and health. Early diagnostics and understanding of genetic mechanisms help to provide timely assistance, improve the quality of life of patients and develop effective methods of treatment and prevention.

Theoretical part

The term "hereditary diseases" refers to diseases that

are transmitted to a child from his parents. The cause of pathological changes transmitted to the next generations is a change in the human genetic code. The human genetic code contains all the information about a person that will accompany him throughout his life. Hidden in its genes are data on the structure of numerous proteins, from which and with the participation of which all human organs and tissues are built, as well as biologically active molecules that regulate such construction. The causes of genetic anomalies are not always precisely clear.

The first observations on the transmission of diseases by inheritance were made in ancient times, but hereditary diseases received a scientific basis with the development of genetics. In the 19th century, Gregor Mendel laid the foundations for the inheritance of traits. In the 20th century, with the discovery of the structure of DNA and chromosomal anomalies, it became possible to identify the causes of many genetic diseases. Gradually, diseases such as albinism, Cri-du-Chat syndrome and Reilly syndrome were described, which made it possible to improve diagnostics and begin developing treatment methods.

Basic concepts

Hereditary diseases are pathologies that arise as a result of mutations in genes or chromosomes that are passed from parents to offspring.

Classification of hereditary diseases

By type of mutation:

Gene (monogenic): caused by a mutation in one gene

Examples: albinism, Reilly syndrome

Chromosomal: caused by a violation of the number or structure of chromosomes

Example: Cri du Chat syndrome (deletion of a region of the 5th chromosome)

Multifactorial: depend on both genes and the external environment

Examples: diabetes, hypertension

By type of inheritance:

Autosomal recessive (e.g. albinism, Reilly syndrome)

Autosomal dominant

Sex-linked (e.g. color blindness)

Albinism is a genetic disease associated with a violation of the production of melanin, a pigment responsible for the color of the skin, hair and eyes. It is transmitted in an autosomal recessive manner.

Cri-du-chat syndrome is a rare genetic disorder caused by a partial deletion of the short arm of chromosome 5. It is named for the child's characteristic cry, reminiscent of a meow, and is accompanied by mental retardation and physical anomalies.

Reilly syndrome (Reilly-Day syndrome) is a rare hereditary disorder belonging to the group of lysosomal storage diseases, most often involving disorders of the nervous system and metabolism. It may be associated with an autosomal recessive type of inheritance.

Modern views

Today, hereditary diseases are considered to be the result of point mutations, chromosomal rearrangements or metabolic disorders. Thanks to the development of molecular genetics, it has become possible to accurately identify genetic defects that underlie diseases such as albinism (mutations in the TYR, OCA2 genes), Cri-du-chat syndrome (deletion of a chromosome region), and Reilly syndrome (defects in metabolic pathways). Research is underway on gene therapy, prenatal diagnostics, and gene editing (CRISPR-Cas9), which opens up new prospects for treatment and prevention. Albinism is a congenital hereditary disease in which the production of melanin, the pigment responsible for the color of the skin, hair, and eyes, is disrupted. People with albinism have fair skin, white or light hair, and very light or blue eyes.

At birth — characteristic external signs are noticeable.

Genetic testing can confirm the type of albinism.

Ophthalmological examination reveals visual impairments characteristic of albinism (for example, nystagmus, photophobia, low vision).

Pros and cons of this "disease"

Pros:

Albinism is not life-threatening in itself.

People with albinism are full-fledged individuals with normal intelligence.

In some cultures, albinos are considered special.

Cons:

Increased risk of skin cancer due to sensitivity to the sun.

Visual impairment, sometimes severe.

Albinism is caused by a mutation in the genes involved in the synthesis of melanin. There are several types of albinism:

OCA1, OCA2, OCA3 and OCA4 are the most common.

Transmitted in an autosomal recessive manner.

If both parents are carriers of the defective gene:

25% chance that the child will have albinism.

50% — will be a carrier.

25% — will not have the mutation.

Symptoms: Very light skin and hair, light or blue eyes,

poor vision (astigmatism, nystagmus, photophobia), sensitivity to light, increased risk of skin cancer.

Cri du chat syndrome (French: Cri-du-chat, ICD-10: Q93.4) is a rare genetic disorder caused by partial loss of the short arm of the fifth chromosome (deletion 5p). The name comes from the fact that newborns with this syndrome emit an unusual high-pitched cry, reminiscent of the meowing of a kitten. This is due to underdevelopment of the larynx and impaired innervation. The disease was first described in 1963 by the French geneticist Jerome Lejeune, who also discovered Down syndrome.

Characteristics of the disease

The work of a number of genes located in the 5p15.2–5p15.3 region is disrupted. Frequently encountered are facial developmental abnormalities, microcephaly, growth and weight retardation, mental retardation, motor and speech delays. The disease is not gender-specific — it occurs in both boys and girls. In most cases (up to 85%), the deletion occurs spontaneously (de novo), without being inherited from parents.

At birth, attention is paid to the characteristic “cat” cry, low weight, and atypical head shape. External signs raise suspicion of genetic pathology

Confirmed by genetic analysis:

Karyotyping — allows you to see the loss of a chromosome region.

FISH analysis (fluorescent in situ hybridization) is more accurate.

CGH array (molecular karyotyping) is a modern method for diagnosing microdeletions.

Symptoms and signs Early manifestations (newborns):

High-pitched, shrill cry

Low birth weight and height

Problems with sucking, swallowing

Later (children and adolescents):

Microcephaly (small head size)

Mental retardation of varying degrees

Facial anomalies: flat face, wide bridge of the nose, hypertelorism (wide-set eyes), low-set ears

Scoliosis, hypotonia (decreased muscle tone)

Congenital heart defects, kidney defects and other anomalies of internal organs are possible

Diagnostics

Physical examination and symptom analysis are the first basis for suspicion.

Genetic tests:

Karyotype is a standard method, reveals large deletions

FISH reveals small losses.

CGH is the most sensitive method, used when microdeletions are suspected.

Additional studies:

ECG, ultrasound of the heart - when defects are suspected

MRI of the brain

Neuropsychological examination

Treatment

There is no specific therapy, treatment is symptomatic and aimed at improving the quality of life:

Early intervention (from infancy): Drug treatment - if necessary (for example, anticonvulsants, if there is epilepsy) Surgery - in the presence of heart defects, larynx, etc. Long-term rehabilitation and support in education, everyday adaptation are necessary.

Probability of the disease Occurs in approximately 1 in 20,000–50,000 newborns. In about 85% of cases, the deletion occurs by chance (de novo).

In the remaining 10-15% of cases, it is inherited from parents who had a balanced translocation (without symptoms, but with a risk of passing on the defect). The risk of having a sick child again depends on the genetic profile of the parents and can be from 0.5% to 50% in the hereditary form.

Life expectancy

In the absence of severe malformations of internal organs, children can live to adulthood. The average life expectancy is reduced, but with good care and medical supervision, life expectancy of over 50 years is possible.

What is Reilly syndrome?

CIPA (Congenital Insensitivity to Pain with Anhidrosis) is a rare genetic disorder in which a person does not feel physical pain, does not sweat (anhidrosis) and often has problems with body temperature regulation. In common parlance, it is also called “Reilly syndrome”, but this is a less accurate term. The syndrome was described in 1932 by doctors Reilly and Meev, hence the name.

Cause of the disease

CIPA is caused by a mutation in the NTRK1 gene, which is responsible for the development and function of nerve cells that transmit pain and temperature signals. As a result:

Nociceptors - pain receptors - are not formed.

The functioning of the autonomic nervous system is disrupted.

Main symptoms: complete insensitivity to pain from birth, lack of sweating (anhidrosis), increased body

temperature (hyperthermia) in the heat, under stress, self-harm (for example, biting their fingers, tongue), injuries, burns, fractures go unnoticed.

Anomalies are noticeable from birth: the child does not cry when given injections, blows, does not react to burns. Genetic testing (NTRK1 analysis) confirms the diagnosis. Additionally, a neurological examination and skin biopsy (checking sensory nerves) are performed. Diagnostics includes: anamnesis (absence of pain, sweat, injury), physical examination, phenetic analysis, skin or nerve biopsy, sweat test.

Treatment: There is no complete cure, treatment is aimed at maintaining life and preventing complications:

Constant examination of the body for wounds, fractures

Cooling the body in the heat (fans, cool clothing)

Physiotherapy, orthopedic care

Practical significance, significance for science and medicine, practical application and possible research

The study of hereditary diseases such as albinism, Cri-du-chat syndrome and Reilly syndrome is of great practical importance for both medicine and society. These pathologies, despite their relative rarity, require an integrated approach to diagnosis, treatment and patient care, which is especially important in the context of increasing birth rates and the emergence of modern methods of gene diagnostics.

The scientific significance of such studies lies in a deeper understanding of the mechanisms of inheritance, molecular and cellular processes occurring during genetic mutations. Using albinism as an example, one can trace how a mutation of one gene (for example, TYR) completely changes the metabolism of melanin in the body. Cri du Chat syndrome provides a clear example of the consequences of a chromosomal deletion. And Reilly syndrome is of interest from the point of view of metabolic disorders and neurogenetics. Medical significance For medicine, knowledge of the nature of these diseases opens the way to the development of methods of genetic diagnostics, prenatal screening, family counseling, and, in the future, to gene therapy, which allows eliminating the cause of the disease at the molecular level. This is especially relevant in neonatology and pediatrics, where it is important to identify and correct possible developmental deviations as early as possible.

Personal opinion

In my opinion, the study of hereditary diseases is not just a scientific necessity, but an important step towards the humanization of medicine. It is especially important that future doctors, including students, understand not only the biological mechanisms of such diseases from their first years, but also feel responsible for the fate of patients. For me personally, the topic of albinism and other rare genetic disorders has become a reason to think about the importance of early diagnosis and careful attitude towards people living with such features, and these diseases seem to me to be related. I believe that the development of genetic research is the key to future medicine, in which every disease can be prevented, and not just treated.

"Understanding the underlying causes of hereditary diseases is the key to the future, where medicine does not just treat diseases, but prevents their occurrence, opening the way to the health of a new generation!"

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