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The Incidence of Down syndrome in Newborns

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Abstract: Down syndrome is the most common chromosomal pathology. The human body consists of millions of cells, each of which has 46 chromosomes. As a result of a genetic mutation, children with Down syndrome have 47 rather than 46 chromosomes. The clinical presentation of the disease is different: from clear defects in the structure and function of organs to permanent mental disorders and acquired immune deficiency. People with this syndrome need qualified help from doctors of various specialties. The main goal of treatment is to correct physical and mental developmental disorders associated with the disease and to improve the patient's quality of life.

Keywords: Down syndrome treatment, Prevention, Rehabilitation, Down syndrome.

About the disease

According to statistics, Down's syndrome is recorded in one in 500-800 babies. About 2,500 children are born with this pathology in the Russian Federation every year, and 8 out of 10 parents leave them immediately after birth.

In recent years, the number of newborns with Down syndrome has increased, which is probably due to the negative impact of environmental factors on the health of future parents and the increase in the number of older women giving birth. depends.

Boys and girls get sick with the same frequency.

Types

Today, geneticists identify three main variants of gene mutation in Down syndrome:

Trisomy (the most common form - in more than 90% of cases);

Translocation (4-5% of people with this diagnosis suffer from it);

Mosaic (registered in less than 1% of cases).

Down syndrome symptoms

During pregnancy, a woman may experience obstetric manifestations of Down syndrome in the fetus. There is a high risk of miscarriage with this anomaly - about 30% of pregnancies in which the fetus has an incorrect set of chromosomes end spontaneously in 6-8 weeks.

A newborn baby is small compared to healthy children. Many children have clear clinical signs, as a result of which the neonatologist can easily identify the chromosomal pathology during the initial examination of the baby.

A child with Down syndrome has the following symptoms:

flat round face;

flat back of the head;

small nose;

wide shortened neck;

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deformed ear structure;

the presence of a characteristic skin fold on the neck;

narrow, wide eyes;

the presence of epicanthus - a vertical fold of skin that covers the corner of the eye from the inside;

slightly opening the mouth with an enlarged tongue;

shortened limbs and fingers;

crooked little fingers;

eye abnormalities (cloudy lenses, white spots on the iris, strabismus);

deformed breast structure;

malocclusion.

Such children are sick and often develop infectious diseases. They tolerate any disease, they often suffer from acute respiratory viral infections, otitis, pneumonia, adenoids, tonsillitis and other pathologies. The inability of the immune system to fight various infections is the most common cause of death in children with Down syndrome in the first 5 years of life.

The syndrome is often accompanied by defects of internal organs (heart, lungs), abnormal structure of genital organs, and pathologies in the structure and function of the esophagus. A growing child lags behind in development compared to healthy children - later he begins to sit, crawl, walk and talk.

Adult children have learning difficulties, poor concentration, poor vocabulary, and impaired speech.

Causes of Down syndrome

As mentioned above, the cells of the human body usually contain 23 pairs of chromosomes - the first half from the father, the second from the mother. As a result of a genetic mutation, additional genetic material is attached to the 21st pair of chromosomes. At this stage of medical development, doctors cannot give a clear answer as to why this happens.

It is generally accepted that a chromosomal abnormality can occur for the following reasons:

accidental genetic diseases;

abnormal division of cells immediately after fertilization of the egg;

genetic mutations inherited from the child's parents.

Neither the lifestyle of the child's mother, her ethnic origin, nor the environmental situation affect the probability of being born with Down syndrome. The only reliably confirmed fact: the older the child's mother, the higher the risk of having a baby with this disease.

Thus, if a pregnant woman under the age of 25 has a 1 in 1,400 chance of giving birth to a child with Down syndrome, by the age of 40, this rate increases to 1 in 100, and to 1 in a 45-year-old woman. 1 in 35. A number of studies have also proven the relationship between the age of the child's father and the risk of developing the disease (the risk increases when over 45).

Other factors that increase the likelihood of an abnormality in the fetus:

young age of the mother (up to 18);

conception from close relatives;

the presence of a translocation in one or both spouses (one of the variants of a chromosomal mutation).

Diagnosis of Down syndrome

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Timely diagnosis of Down's syndrome became available to all pregnant women after the introduction of mandatory ultrasound examination. Modern ultrasound equipment makes it possible to suspect an anomaly in the 12-13th obstetric week.

In addition, there is a mandatory blood test for biochemical markers. This study allows the geneticist to assess the likelihood of chromosomal abnormalities in the fetus.

Older pregnant women, as well as women with poor screening results, need invasive diagnostics - chorionic villus biopsy (examination of placental tissue). It is also possible to perform amniocentesis - examination of amniotic fluid.

The high accuracy of the methods used to study pathology allows future parents to make an independent decision whether to take responsibility for the birth and upbringing of a child with Down syndrome or whether to resort to termination of pregnancy for medical reasons. .

If for some reason a woman has not passed mandatory screening tests, this genetic syndrome is easily detected by a neonatologist during the first examination of a newborn.

If there are external manifestations of a genetic disease, the child will be sent for additional tests to confirm the diagnosis and identify joint developmental defects.

For this purpose, the following is carried out:

genetic diagnosis of the baby's chromosome set;

echocardiography of the heart;

Ultrasound examination of abdominal organs;

examination by a cardiologist, neurologist, ophthalmologist, orthopedist and other specialists (as indicated).

Expert opinion

In recent years, the life expectancy of people with Down syndrome has increased significantly due to medical advances. If half a century ago it did not exceed 10 years, now its expected duration reaches 50 years.

The correct pedagogical approach ensures a sufficiently high learning ability of such patients: most of them acquire simple household skills needed in everyday life. Sometimes they live independently and start a family.

Today, Down syndrome is not a death sentence. There are many educational methods and rehabilitation tools that allow such patients to feel like a complete part of this world.

Treatment of Down syndrome

The possibilities of modern medicine in the treatment of chromosomal diseases are very limited. Doctors focus their efforts on stabilizing the patient's physical and mental health, improving his quality of life and preventing the development of infection in the body (due to the weak immunity of people with this pathology).

The patient is prescribed nootropic and vascular drugs. Drugs with a symptomatic effect are also used - anti-inflammatory drugs, pain relievers, vitamin complexes. If the thyroid gland is underactive, use hormonal drugs.

Physiotherapy methods and exercise therapy are actively used in the treatment of patients with Down syndrome. Trainings are held with a speech therapist and a psychologist to develop speech and communication skills.

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Prevention

There is no specific prevention for Down syndrome. Every couple has a risk of giving birth to a child with this pathology. To reduce this probability, you should maintain a healthy lifestyle and not delay the birth of a baby until the age of 40.

Rehabilitation

Throughout his life, a person with chromosomal diseases should be monitored by specialists in various fields - ophthalmologist, cardiologist, endocrinologist, neurologist, immunologist, psychotherapist, etc. Timely detection of pathologies of various systems and organs helps doctors to start timely treatment and prevent the development of complications.

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