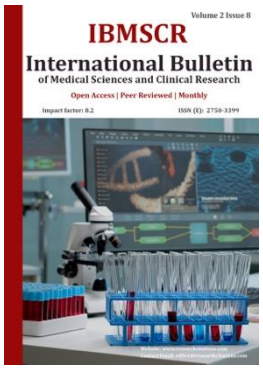

THE FREQUENCY OF OCCURRENCE OF PATIENTS IN THIS COHORT AMONG PATIENTS WITH DOWN SYNDROME TREATED WITH VARIOUS DISEASES IN ADOLESCENTS WITH DOWN SYNDROME

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https://doi.org/10.5281/zenodo.10682422

ABSTRACT. This article focuses on a comparative description of anthropometric indicators in children with Down syndrome existing adolescence. In the Departments of Reproductive Center of Bukhara region, Mother-Child Health, and hereditary diseases, the degree of occurrence of Down syndrome in existing children in adolescence, as well as a specific Clinic of Down syndrome and modern research methods are presented.

Keywords. Down syndrome, immune status, reproductive function, blood system, metabolism, respiratory infection, anthropometric indicator.

Down syndrome is the most common chromosomal pathology. The human body consists of millions of cells, each of which contains 46 chromosomes. As a result of a genetic mutation, not 46, but 47 chromosomes are formed in children with Down syndrome. The clinical picture of the disease is diverse: from pronounced defects in the structure and functioning of organs to persistent mental disorders and acquired immunodeficiency. People with this syndrome need qualified help from doctors of various profiles. The main task of treatment is to correct concomitant disorders of physical and mental development and to achieve an improvement in the patient's quality of life. 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 expectant parents and growth.

To date, geneticists have identified 3 main variants of the gene mutation in Down syndrome:

- trisomy (the most common form is more than 90% of cases);
- translocation (4-5% of people with this diagnosis suffer from it);
- mosaic (less than 1% of cases are registered).

Symptoms of Down syndrome

During pregnancy, a woman may encounter obstetric manifestations of fetal Down syndrome. With this anomaly, high risks of miscarriages are determined – about 30% of pregnancies in which the fetus has an incorrect set of chromosomes are spontaneously interrupted at 6-8 weeks.

A newborn baby is small in comparison with healthy children. Most children have vivid clinical signs, due to which chromosomal pathology is easily determined by a neonatologist already at the initial examination of the baby.

A child with Down syndrome is characterized by:

- * a flat round face;
- flat nape;
- small nose;

• Wide shortened neck;
• deformed structure of the ears;
• the presence of a characteristic skin fold on the neck;
• narrow, wide-set eyes;
• the presence of epicanthus – a vertical skin fold covering the corner of the eye from the inside;
• slightly open mouth with enlarged tongue;
• shortened limbs and fingers;
• crooked little fingers;
• eye abnormalities (clouding of the lens, white spots on the iris, strabismus);
• deformed structure of the chest;
• malocclusion.

Such children are painful – they often have infectious diseases. They tolerate any diseases more difficult, they often suffer from acute respiratory infections, otitis media, 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 the genitals, pathologies in the structure and functioning of the esophagus. Growing up, the child lags behind in development in comparison with healthy children – later begins to sit, crawl, walk, talk. Grown-up children have learning difficulties, they have reduced concentration, poor vocabulary, slurred speech.

Causes of Down syndrome

As already mentioned, the cells of the human body normally 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 the development of medicine, doctors cannot give an exact answer why this is happening.

It is generally believed that a chromosomal abnormality can be caused by:
• accidental genetic disorders;
• abnormal cell division immediately after fertilization of the egg;
• genetic mutations inherited from the child's parents.

Neither the lifestyle of the child's mother, nor her ethnicity, nor the environmental situation affect the likelihood of having a baby with Down syndrome. The only reliably established fact is that the higher the age of the child's mother, the greater the risk of having a baby with this disease.

So, if a pregnant woman under the age of 25 has a probability of having a child with Down syndrome of 1 in 1400, then by the age of 40 this indicator increases to 1 in 100, and for a 45-year-old woman to 1 in 35. Also, a number of studies have proven the relationship between the age of the child's father and the risk of developing the disease (at the age of over 45 years, the risk increases).

Other factors that increase the likelihood of an anomaly in the fetus:
• the young age of the mother (under 18 years);
• conception from close relatives;

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

Down syndrome (formerly Down's disease) It was first described by the English physician John Langdon Down in 1866, but its connection with the 21st chromosome was established almost 100 years later by Dr. Jerome Lejeune in Paris. It is the presence of all or part of the third copy of chromosome 21 that causes Down syndrome, the most common chromosomal abnormality found in humans. It has also been found that the most common live-born aneuploidy is trisomy 21, which causes this syndrome.

Most patients with Down syndrome (ICD-10 code: Q90) have an additional copy of chromosome 21. There are various hypotheses related to the genetic basis of Down syndrome and the association of various genotypes with phenotypes. Among them is an imbalance in the dosage of genes, in which there is an increased dosage or number of Hsa21 genes, which leads to increased gene expansion. This also includes the possibility of associating different genes with different phenotypes of Down syndrome. Another popular hypothesis is the hypothesis of increased developmental instability, according to which a genetic imbalance is created by a number of trisomal genes and leads to a greater influence on the expression and regulation of many genes.

The critical domain hypothesis is also well known in this list. The critical regions of Down syndrome (DM) are several chromosomal regions that are associated with partial trisomy for Has21. DSCR 21q21.22 is responsible for many clinical signs of Down syndrome. After careful study of various analyses, it became clear that one gene of a critical region cannot cause all the phenotypic features associated with trisomy 21, rather, it is more obvious that several critical regions or critical genes play a role in this phenomenon.

Prevalence

The frequency of Down syndrome increases with the age of the mother, and its prevalence varies in different populations (from 1 in 319 to 1 in 1000 live births), it is also known that the frequency of conception of fetuses with Down syndrome is quite high during conception, but about 50% to 75% of these fetuses are lost prematurely. The occurrence of another autosomal trisomy is much more common than 21, but postpartum survival is very low compared to Down syndrome. It is believed that such a high survival rate in patients with trisomy 21 is a function of a small number of genes in chromosome 21, called Hsa21, which is the smallest and least dense of the autosomes.

An additional copy of chromosome 21 is associated with Down syndrome, which occurs due to the inability of chromosome 21 to separate during gametogenesis, which leads to the appearance of an additional chromosome in all cells of the body. Robertson translocation and isochromosome or ring chromosome are two other possible causes of trisomy 21. An isochromosome is a condition in which, in Robertson translocation, two long branches separate from each other instead of a long and short branch. This occurs in 2-4% of patients. The long arm of chromosome 21 is attached to another chromosome, mainly chromosome 14. In mosaicism, there are 2 different cell lines due to a division error after fertilization.

Clinical features

Different clinical conditions are associated with Down syndrome because it affects different systems. These patients have a wide range of signs and symptoms, such as intellectual and developmental disorders or neurological features, congenital heart defects, gastrointestinal (GI) disorders, characteristic facial features and abnormalities.

Treatment of people with Down syndrome / management

The management of patients with Down syndrome is interdisciplinary. A newborn with suspected Down syndrome should be karyotyped to confirm the diagnosis. The family should be referred to a clinical geneticist for genetic testing and counseling of both parents.

Educating parents is one of the most important aspects of Down syndrome treatment, as parents need to be aware of the various possible conditions associated with it so that they can be diagnosed and treated appropriately. Treatment is mostly symptomatic, and full recovery is impossible.

These patients should have their hearing and vision assessed, and since they are more prone to developing cataracts, timely surgical intervention is therefore required. Thyroid function tests should be performed annually, and appropriate treatment should be provided in case of a violation.

For optimal growth and weight gain, a balanced diet, regular exercise and physical therapy are necessary, although nutritional problems do improve after heart surgery.

Referral to a cardiologist should be directed to all patients, regardless of the clinical signs of congenital heart disease, which, if present, should be adjusted during the first 6 months of life to ensure optimal growth and development of the child.

Other specialties involved include developmental pediatrician, pediatric pulmonologist, gastroenterologist, neurologist, neurosurgeon, orthopedist, child psychiatrist, physiotherapist and occupational therapist, speech therapist and audiologist.

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