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GENERAL INFORMATION ABOUT DOWN'S DISEASE AND CHANGES THAT OCCUR IN THE BODY

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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 (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.

Congenital heart defects (CHD)

Congenital heart defects are by far the most common and leading cause of morbidity and mortality in patients with Down syndrome, especially in the first 2 years of life. Although various assumptions have been made about geographical as well as seasonal differences in the occurrence of different types of congenital heart defects in trisomy 21, so far none of the results have been conclusive.

The incidence of coronary heart disease in children born with Down syndrome reaches 50%. The most common heart defect associated with Down syndrome is an atrioventricular septal defect (AVSD), and this defect accounts for up to 40% of congenital heart defects in Down syndrome, it is believed that this is due to a

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mutation of a gene unrelated to Hsa21 CRELD1, and the second most common heart defect in Down syndrome is an interventricular septal defect (VSD), which is observed in about 32% of patients with Down syndrome. They account for more than 50% of congenital heart defects in patients with Down syndrome.

Other heart defects associated with trisomy 21 are a defect of the second atrium (10%), tetrad of Fallot (6%) and isolated CPC (4%), while about 30% of patients have more than one heart defect. There are geographical differences in the prevalence of heart disease in Down syndrome, with VSD being the most common in Asia, and type II ASD in Latin America. The reason for this difference in the prevalence of different types of coronary heart disease in different regions is still unclear, and it has been found that many factors contribute to this, such as the proximity of regions.

Due to such a high prevalence of coronary heart disease in patients with Down syndrome, it was recommended that all patients undergo an echocardiogram during the first few weeks of life.

Mental disorders

Trisomy Hsa21 is associated with a decrease in brain volume, especially the hippocampus and cerebellum. Hypotension is a hallmark of children with Down syndrome and is present in almost all of them. It is defined as reduced resistance to passive muscle stretching and is the cause of delayed motor development in these patients. Due to hypotension, joint weakness is observed in patients with Down syndrome, which leads to a decrease in gait stability and an increase in energy requirements for physical exertion. These patients tend to have a decrease in bone mass and an increased risk of fractures due to low levels of physical activity, while ligament weakness predisposes these patients to atlantoaxial subluxation.

From five to 13% of children with Down syndrome suffer from seizures, of which 40% will have seizures before their first birthday, and in these cases, seizures are usually childhood spasms. Children with Down syndrome with infantile spasm actually respond better to antiepileptic drugs compared to other children with the same diseases, and therefore early intervention and treatment improve developmental outcomes.

Lennox-Gesto syndrome is also considered more common in children with Down syndrome when it does occur, has a late onset and is associated with reflex seizures along with an increased frequency of EEG disorders.

Forty percent of patients with Down syndrome develop tonic-clonic or myoclonic seizures during the first three decades of life. Dementia is more common in patients over the age of 45 with Down syndrome, and about 84% of them are more likely to develop seizures. Seizures in these patients are associated with a rapid decrease in their cognitive functions.

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The risk of developing early-stage Alzheimer's disease is significantly high in patients with Down syndrome: 50-70% of patients develop dementia by the age of 60. It has been established that the amyloid precursor protein (APP), which is known to be associated with an increased risk of developing Alzheimer's disease, is encoded by Hsa21, and trisomy of this protein is probably responsible for the increased incidence of dementia in people with Down syndrome. Recent studies have shown that APP tripling is associated with an increased risk of early development of Alzheimer's disease, even in a normal population.

Almost all patients with Down syndrome have mild to moderate learning disabilities. Trisomy of multiple genes, including DYRK1A, synaptoyanin 1, and target homologue 2 (SIM2), has been found to cause learning and memory defects in mice, suggesting the possibility that overexpression of these genes may likely cause learning disabilities in people with Down syndrome.

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.

Differential diagnosis

• Congenital hypothyroidism;

- Mosaic trisomy syndrome 21;
- Partial trisomy 21 (or duplication of 21q);
- Robertson's trisomy 21;
- Trisomy 18;
- Zellweger syndrome or other peroxisomal disorders.

Due to recent advances in medical practice, the development of surgical methods for correcting congenital disorders and improved general care, there has been a significant increase in infant survival and life expectancy of patients with Down syndrome. A study conducted in Birmingham (United Kingdom) almost 60 years ago showed that 45% of infants survived the first year of life, and only 40% will be alive after 5 years. A later study, conducted about 50 years after that, showed that 78% of patients with Down syndrome plus congenital heart disease survived for 1 year, while in patients without abnormalities, this number increased to 96%. This increase in the life expectancy of these patients should continue to increase significantly due to advances in medical science. Medical institutions strive to ensure proper and timely treatment of these patients and help them lead a full and productive life.

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