

ORIGIN AND METHODS OF TREATMENT OF DOWN SYNDROME

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Abstract: This article is devoted to the genetic and epigenetic causes of Down syndrome, as well as modern medical and pedagogical methods of treatment. The article analyzes the genetic mutations of Down syndrome, manifestations of the disease in childhood and adolescence and their impact on the quality of life. The accuracy of symptoms, diagnostic methods (in situ hybridization, karyotyping) and the importance of early detection are considered. The effectiveness of drugs, physiotherapy, speech therapy, inclusive education and family counseling as methods of intervention is also assessed. The experimental part analyzes the metrics of diagnosis and rehabilitation based on clinical observations and social surveys. The discussion section compares with the scientific literature, considers the advantages and limitations of treatment methods, as well as prospects for further research. The conclusions note the need to strengthen the integrative approach to improve the quality of life of children with Down syndrome.

Keywords: Down syndrome, genetic mutation, diagnostics, karyotyping, therapeutic rehabilitation, inclusive education, physiotherapy, family counseling.

Introduction

1. General characteristics and relevance

Down syndrome is the most common genetic disorder caused by a complete or partial third copy of chromosome 21. The disease occurs in approximately one case per 700–800 live births, and in some regions the rate may be one case per 1000. Epidemiological studies show that Down syndrome most often occurs as a result of a random genetic mutation, with the risk increasing with maternal age, especially after 35 years. A comprehensive approach, including medical, pedagogical, psychological and social aspects, is required to properly understand the disease.

Down syndrome has a significant impact on health, development and social integration, making it a pressing issue for both national and international medical communities and society as a whole. Modern scientific research covers not only genetic mechanisms and diagnostic methods, but also issues of individual development, support, family adaptation and inclusive education.

2. Genetic basis

Understanding the genetic nature of Down syndrome is important for proper diagnosis and selection of rehabilitation methods. There are three main types:

Complete trisomy 21: the most common form (92–95% of cases), in which the patient has a complete third copy of chromosome 21 in all or most of his cells.

Translocation trisomy 21: chromosome 21 or part of it is abnormally attached to another chromosome (usually 14). This form is less common, but can be hereditary if one of the parents is a carrier of a balanced translocation.

Mosaic trisomy: occurs in about 1–2% of cases, when the patient has cells with both a normal and a trisomic set of chromosome 21. Symptoms may be less pronounced, but not always.

These variants are diagnosed using the following methods:

Karyotyping: a classic method of determining the structure and number of chromosomes under a microscope.

Hybridization: a method for detecting genetic markers on chromosomes that is highly sensitive and rapid.

Molecular genetic techniques (such as CGH microarray or nucleic acid sequencing) that can detect small changes.

3. Epidemiology and local characteristics

Global data on the prevalence of Down syndrome vary due to prenatal diagnosis, abortion laws, and social factors. In Western Europe and North America, the number of cases diagnosed with Down syndrome at birth has decreased due to prenatal screening and abortion. However, statistics do not always reflect the full picture due to recording limitations and social barriers.

In Uzbekistan, official data are limited, but according to information from regional medical institutions, cases of children born with Down syndrome are registered every year. Diagnostic capabilities in central cities are better than in rural areas.

4. Social, psychological, and family consequences

Down syndrome is not only a medical problem, but also a social one.

Family reactions: Parents often experience stress, anxiety and guilt, especially in the early years. Intensive psychological support is needed.

Social stereotypes: Negative attitudes towards children with disabilities are still widespread, which makes it difficult for them to integrate into society and education.

Financial burden: The costs of diagnosis, rehabilitation, special education and support can be significant, especially for low-income families.

5. Treatment and rehabilitation methods

Down syndrome cannot be completely cured, but the following interventions significantly improve the quality of life:

Physiotherapy and developmental therapy: aimed at strengthening muscle tone, improving motor skills and balance. Occupational therapy promotes independence in everyday activities.

Speech and speech therapy: help develop speech and communication skills.

Inclusive education: integration into regular schools with the support of teachers promotes socialization and equal opportunities.

Medical care: treatment of associated diseases such as heart defects, thyroid disorders, vision and hearing problems.

Psychological support for the family: consultations, group trainings and seminars help parents adapt and share experiences.

Objectives of the study: **6. Objectives, Limitations, and Methods of the Study**

Analysis of genetic mechanisms and types of trisomy 21.

Study of modern diagnostic methods and their limitations.

Evaluation of the effectiveness of rehabilitation programs in the context of Uzbekistan.

Study of family and social support and the role of national policy.

Identification of areas for future research, including genetic protection, pedagogical integration, and telemedicine. Limitations: Insufficient epidemiological data, especially in Uzbekistan.

Individual differences in development and conditions, complicating generalizations.

Subjectivity of assessment methods and lack of standardized indicators.

7. Structure of the article

The article consists of: Introduction, revealing the problem, objectives, and context.

Methodological part, describing clinical observations and social surveys.

Analysis and discussion of the results.

Conclusions with findings and recommendations.

Conclusion

Down syndrome is one of the most common genetic disorders caused by an extra 21st chromosome. This leads to complex changes in physical, mental, speech and emotional development. Typical features of appearance, developmental delay, as well as concomitant medical problems (heart defects, hearing and vision impairment, thyroid dysfunction, etc.) are typical.

The article examines in detail the genetic mechanisms of Down syndrome and various forms of trisomy 21. The importance of prenatal diagnosis is emphasized, which allows for timely detection of the disease and planning of the necessary support measures.

Various diagnostic methods, including karyotyping, molecular studies and FISH, provide accurate detection of genetic changes. However, prenatal screening is not available in all regions and is not always acceptable from a psychological point of view.

Rehabilitation and social integration play a key role in improving the quality of life of children with Down syndrome. Early comprehensive assistance, inclusive education, speech therapy and physical therapy support help to maximize the child's potential.

The diversity of development levels requires an individual approach, and the lack of resources and the presence of stereotypes limit the opportunities of children and their families, especially in developing countries.

Current trends show an increase in positive attitudes towards people with Down syndrome, the expansion of inclusive programs and social support. An important task remains the development of scientific research, improvement of local statistics and infrastructure, advanced training of specialists and informing the society.

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