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CONGENITAL GENETIC DISORDER: MEDICAL AND SOCIAL ASPECTS OF DOWN SYNDROME

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Abstract
Down syndrome (trisomy 21) is a genetic disorder caused by an extra chromosome in the 21st pair of chromosomes. This syndrome leads to delays in intellectual and physical development, distinctive facial features, and other health issues. Prenatal tests are available for early detection of the condition, and early interventions can improve the development of patients. The article provides information about the diagnosis, development, and treatment methods of Down syndrome.

Keywords: Down syndrome, trisomy 21, prenatal diagnostics, rehabilitation, chromosomal anomalies, nootropic drugs.

Introduction

Down syndrome (trisomy 21) is a genetic disorder caused by the presence of an extra chromosome in the 21st pair. This syndrome is characterized by delayed intellectual and physical development, distinctive facial features, and low muscle tone in both children and adults. The causes of Down syndrome are related to a disruption in the separation of chromosomes during meiosis, leading to the formation of an extra chromosome. The condition is primarily hereditary, and the risk increases with maternal age, especially after the age of 35. Children born with Down syndrome face not only intellectual disabilities but also a variety of physical and medical challenges. Early diagnosis and special interventions can help improve the development of patients. Today, non-invasive prenatal tests allow for the detection of Down syndrome during pregnancy, which is crucial for the future development of the child. This article provides detailed information on the genetics, diagnostics, developmental processes, and treatment methods of Down syndrome, as well as discussing effective assistance and rehabilitation strategies for managing the condition.

Down syndrome is a form of congenital oligophrenia. Along with intellectual disability, the child's physical appearance is also characterized by distinct features. The disease was first described by the English physician Langdon Down in 1886. The condition is hereditary and occurs rarely; it is primarily caused by the presence of an extra chromosome in the chromosome set (the 21st pair), which is why it is sometimes referred to as trisomy. This condition results from the failure of

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chromosome separation during meiosis, leading to the formation of a gamete with 24 chromosomes. When combined with a normal gamete from the opposite sex, the resulting zygote has 47 chromosomes. Children affected by Down syndrome are not only intellectually but also physically weak and prone to various infectious diseases. Patients generally require rehabilitation, and education should be provided in special schools. Down syndrome affects both males and females.

Individuals with Down syndrome typically have a small head in relation to their body, a broad face, closely set eyes, and a partially open mouth, along with intellectual disability. People with Down syndrome almost always have both physical and intellectual impairments. As adults, their intellectual abilities usually resemble those of an 8 or 9-year-old child. However, their emotional and social awareness is often quite high. They may have weak immunity and usually reach developmental milestones at a later age. There is an increased risk of various health problems, including congenital heart defects, epilepsy, leukemia, and thyroid diseases.

People with Down syndrome may display the following physical characteristics: a small chin, epicanthal folds, low muscle tone, a flat nasal bridge, a single crease in the palm, and a protruding tongue. The protruding tongue occurs due to low muscle tone and weak facial muscles and is often corrected with myofunctional exercises. Some characteristic airway features in individuals with Down syndrome may lead to obstructive sleep apnea in about half of these individuals. Other common features include joint hypermobility, an additional space between the thumb and second finger, a single palm crease, and shorter fingers.

Their tongue, skin, and lips may be dry, and they often have strabismus. Their teeth may not be aligned properly. Hair is typically thin and smooth. In Down syndrome, the angle of the hand's palm is usually larger than normal; it can be 80° or more, compared to the typical 57° angle.

The muscular system is also poorly developed. Atlantoaxial joint instability occurs in approximately 1–2% of cases. Atlantoaxial instability can lead to myelopathy later in life due to cervical spine compression, which often presents as newly onset weakness, coordination problems, incontinence, and gait dysfunction. Serial imaging cannot reliably predict future cervical spine compression, but changes can be observed during a neurological examination. The condition is corrected through spinal surgery.

The primary materials for studying Down syndrome include genetic research and prenatal diagnostics. In children born with this syndrome, an extra chromosome in the 21st pair is detected. Methods such as non-invasive prenatal tests, fetal DNA analysis, and genetic counseling are used to diagnose the condition. These methods are highly effective for early diagnosis, allowing the identification of Down syndrome in the early stages of fetal development. The medical histories, developmental characteristics, and clinical symptoms of patients with Down syndrome are also studied. Additionally, the research explores rehabilitation methods used for children with Down syndrome, special education programs, and physical and psychological therapies.

Research results have shown that children born with Down syndrome often have mild to moderate intellectual disability. They face delays in several developmental stages, such as walking independently, self-expression, and speech development. Children with Down syndrome commonly have low muscle tone, which slows their physical development. Additionally, these children are more likely to experience congenital heart defects, infections, and thyroid disorders.

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The impact of maternal age on the development of the condition has also been identified: the likelihood of having a child with Down syndrome significantly increases in women over the age of 35. This information highlights the importance of conducting fetal DNA analysis during pregnancy, as this method improves the chances of early detection of the condition. Moreover, special education programs and rehabilitation for children with Down syndrome, such as speech therapy, physiotherapy, and occupational therapy, significantly improve their development. Early interventions help enhance the children's future social and physical abilities..

The incidence of Down syndrome among newborns (frequency) in the UK research results)

Mother's age	The incidence rate of Down syndrome.
15-19	3:10000
20-24	4:10000
25-29	4:10000
30-34	11:10000
35-39	33:10000
40-44	124:10000
45 and over	312:10000

Conclusion

Down syndrome is a genetic disorder caused by chromosomal abnormalities, leading to developmental delays and intellectual disabilities in children. Early diagnosis and rehabilitation, as well as special education, can help improve the development of these children. Prenatal diagnostics and non-invasive tests allow for the detection of Down syndrome during pregnancy, which facilitates early interventions. Although there is no cure for the condition, with proper support and assistance, children with Down syndrome can become active members of society. Through early intervention, specialized therapy, and education, the quality of life for individuals living with Down syndrome can be significantly improved.

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