

DOWN SYNDROME AND ITS CLINICAL AND PSYCHOLOGICAL SIGNS

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Annotation: This article presents a comprehensive study of Down syndrome, a genetic disorder characterized by the presence of an additional copy of the 21st chromosome. The focus is on understanding the clinical and psychological signs associated with this condition. The purpose of the article is to shed light on various manifestations of Down syndrome by analyzing the literature, emphasizing its impact on both physical health and psychological well-being. The Methods section describes the approaches used to collect relevant information, while the results section presents key findings. The Discussion section discusses the implications of these findings, giving insight into potential interventions. Concluding the article with suggestions, it is intended to promote a better understanding of Down syndrome and improve support strategies.

Keywords: Down syndrome, trisomy 21, mental retardation, cognitive impairment, developmental delays, concomitant diseases, psychological impact, intervention strategies.

Down syndrome, also known as trisomy 21, is a genetic disorder resulting from having an extra copy of chromosome 21. This additional genetic material leads to a number of physical and intellectual problems. Despite the fact that advances in medical care have increased the life expectancy and quality of life of people with Down syndrome, it is extremely important to study clinical and psychological signs in order to develop effective intervention strategies.

Numerous studies have examined the clinical manifestations of Down syndrome, highlighting common physical features such as facial features, heart defects and gastrointestinal problems. However, the focus is not only on the physical sphere, but also on intellectual and developmental aspects. People with Down syndrome often exhibit mental retardation, cognitive impairment and developmental delays. Moreover, the syndrome is associated with an increased risk of developing certain diseases, including thyroid dysfunction and breathing problems.

The psychological impact of Down syndrome is equally significant. People may face problems related to social interactions, communication, and emotional well-being. The literature emphasizes the importance of an interdisciplinary approach to meeting

the diverse needs of people with Down syndrome with the involvement of medical professionals, educators and psychologists.

To collect this information, a systematic review of scientific databases such as PubMed, ScienceDirect and PsycINFO was conducted. Search queries included "Down syndrome", "trisomy 21", "mental retardation" and "psychological impact". Articles published over the past decade have been given priority to ensure that the latest research is included.

Down syndrome, also known as trisomy 21, is a genetic disorder caused by the presence of an extra copy of chromosome 21. This additional genetic material affects the normal development of the body and brain, leading to a number of physical and intellectual problems. Here are some clinical and psychological signs associated with Down syndrome:

Clinical signs:

Physical features:

- Facial characteristics: People with Down syndrome often have distinct facial features such as a flattened face, slanted eyes, a small nose and a protruding tongue.

- Low muscle tone: Hypotension, or low muscle tone, is common in people with Down syndrome, which can affect motor skills and coordination.

- Short stature: People with Down syndrome usually have a shorter stature compared to the general population.

- Joint flabbiness: Loose joints are common, and this can promote flexibility, but it can also lead to joint instability.

Health Problems:

- Heart defects: Approximately half of people with Down syndrome are born with congenital heart defects.

- Gastrointestinal problems: There may be an increased risk of gastrointestinal problems, such as blockage of the gastrointestinal tract or malformations.

- Vision and hearing problems: Common problems include nearsightedness, farsightedness and an increased risk of ear infections.

Intellectual and developmental disorders:

- A distinctive feature of Down syndrome is intellectual disabilities, in which a wide range of cognitive abilities is observed.

- Delays in the development of speech and language skills, as well as motor skills are common.

Psychological signs:

Cognitive Abilities:

- Mental retardation: People with Down syndrome usually have mild or moderate mental retardation. However, cognitive abilities vary significantly.

Language and communication:

- Delayed speech and language development: The development of speech and language may be delayed, and speech therapy may be useful to individuals.

Behavioral and Emotional Characteristics:

- Social Skills: Although people with Down syndrome often have strong social skills and enjoy social interaction, they may face difficulties in difficult social situations.

- Emotional expression: Some people may have difficulty expressing and understanding complex emotions.

Memory and Learning:

- Short-term memory: Short-term memory can be a relative strength, while long-term memory and learning can vary greatly.

Adaptive functioning:

- Daily life skills: Independence in everyday activities such as dressing, grooming, and self-care after childbirth may require additional support and training.

It is important to note that each person with Down syndrome is unique, and the severity of symptoms may vary. Early intervention, a supportive environment and individual educational plans can greatly contribute to the well-being and development of people with Down syndrome. In addition, advances in medical care and educational approaches continue to improve the quality of life of people with Down syndrome.

The multifaceted nature of Down syndrome requires a holistic approach to treatment. Early intervention programs, inclusive education and individual therapeutic interventions are important components of a comprehensive strategy. The collaborative efforts of healthcare professionals, educators, and families are critical to meeting the unique needs of people with Down syndrome.

In addition, ongoing research on the genetic and molecular aspects of Down syndrome opens up prospects for the development of targeted therapy. The integration of people with Down syndrome into mainstream society promotes social integration and increases overall well-being.

In conclusion, this article highlights the complex interaction of clinical and psychological manifestations in people with Down syndrome. Understanding these aspects is vital for developing activities that promote optimal development and quality of life. An emphasis on early detection, intervention and the creation of an enabling environment are key strategies in solving problems associated with Down syndrome.

In the future, the continuation of research on the genetic basis of Down syndrome and the improvement of therapeutic approaches will contribute to improving the results of treatment of people with this disease. It is crucial for health care providers, educators, and policy makers to work together to create a more inclusive and supportive society for people with Down syndrome.

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