

# DOWN SYNDROME AND ITS AGE-RELATED CHARACTERISTICS

Khudjanova Muattar Absalamovna  
Senior Lecturer at the KIUT  
Department of Fundamental Medical Sciences

Shukhrat Pulodov,  
Khadichabonu Jurakulova,  
KIUT MED-S Students

## Abstract

This article provides information on the origins, history, causes, clinical presentation, etiology, age-related characteristics, and developmental stages of Down syndrome.

**Keywords:** Down syndrome, leukemia, Alzheimer's, chromosome, autosome, anomaly, trisomy, mosaicism, translocation, cognitive, hippocampus, cerebellum.

## Introduction

**Objective:** The objective of this article is to provide a comprehensive overview of Down syndrome, including its genetic origins, clinical features, and age-specific characteristics. It aims to deepen the understanding of Down syndrome's developmental trajectory, the physiological and cognitive challenges associated with the condition, and its prevalence. By exploring these aspects, the article seeks to highlight the significance of genetic factors in the manifestation of Down syndrome and its impact on individuals across different stages of life.

**Relevance of the Topic:** Approximately 20% of severe disorders of the central nervous system are linked to genetic anomalies. Down syndrome is one of the most common chromosomal anomalies known today, characterized by delayed intellectual development and unique physical traits. This genetic anomaly, a congenital chromosomal disorder associated with an extra chromosome, is relatively widespread, occurring in approximately one out of every 650–800 newborns.

Human cells typically contain 46 chromosomes that carry hereditary traits passed down from parents, arranged in equal pairs. English physician L. Down first described Down syndrome in 1866. Children born with Down syndrome have an additional chromosome in the 21st pair, totaling 47 chromosomes. This extra chromosome is the primary cause of Down syndrome. It is associated with mild to moderate developmental delays. Most individuals with Down syndrome exhibit distinctive facial features and low muscle tone in early childhood. Many children with Down syndrome also experience heart defects, a heightened risk of leukemia, early onset of Alzheimer's disease, gastrointestinal issues, and other pathological conditions. The severity of Down syndrome symptoms can vary from mild to severe.



### Types of Down Syndrome

**Trisomy 21** – This is a condition in which there are three chromosomes in the 21st autosomal pair instead of two. Studies confirm that the presence of an extra chromosome in the autosome group leads to developmental abnormalities in the embryo. Sometimes, the occurrence of Down syndrome may be linked to "aging" in the female body and reproductive health issues.

In the form known as Translocation Down Syndrome, part or all of a chromosome is transferred to another chromosome. Typically, chromosomes 13-15 and 21-22 are involved, yet the total chromosome count remains 46. In this case, the likelihood of a child being born with this chromosomal abnormality is associated with the father (who carries the mutation), and the mother's age plays a lesser role. Children with Translocation Down Syndrome represent approximately 5% of all cases.

**Mosaic Down Syndrome** – This third type of chromosomal anomaly occurs when some cells have 46 chromosomes, while others have 47 (with trisomy of the 21st chromosome). This form occurs in about 1% of cases. In some children, the characteristics of the syndrome may be mild or more pronounced, and their intellectual abilities may be higher than those with Translocation Down Syndrome. This abnormal chromosome set is linked to errors occurring in specific stages of meiosis.

After age 35, women experience hormonal changes and alterations in the formation of egg cells. It is still unknown which of the extra genes on chromosome 21 contribute to the various symptoms. Scientists suggest that the increase in gene number disrupts their interactions, causing some genes to become overactive while others become underactive. This imbalance negatively impacts differentiation, development, and psycho-emotional and intellectual growth in the body. Chromosome 21 contains approximately 400 genes, most of which are not fully understood.

### Intellectual Abilities in Down Syndrome

Individuals with Down syndrome often exhibit significant individual differences. They inherit many traits from their parents and often resemble their siblings. Alongside unique personal characteristics, individuals with Down syndrome also share common physiological traits. In addition to physical characteristics associated with this syndrome, individuals often face challenges in learning and acquiring new skills. Cognitive impairments, another common issue with Down syndrome, especially impact communication skills. Individuals with Down syndrome experience delayed cognitive development and face lifelong learning difficulties.

Although the brains of individuals with Down syndrome are generally average-sized, scientists have identified structural and functional changes in specific areas, such as the hippocampus and cerebellum. The hippocampus, crucial for learning and memory, is particularly affected. Currently, researchers are using animal models of Down syndrome to study how certain genes on chromosome 21 might impact cognitive impairments. Development of Children with Down Syndrome

Individuals with Down syndrome have an increased susceptibility to a range of medical conditions, including hearing impairments, ear infections (otitis), thyroid disorders (hypothyroidism), cervical spine instability, vision problems, sleep apnea, obesity, constipation, infantile spasms, seizures,



dementia, and early-onset Alzheimer's disease. Approximately 18-38% of individuals with Down syndrome also experience mental health or behavioral disorders, such as autism spectrum disorders, attention deficit hyperactivity disorder (ADHD), depression, stereotypic movement disorders, and obsessive-compulsive disorders. Intellectual development often features significant speech delays; for example, articulation difficulties and stuttering can sometimes give the impression of lower cognitive abilities. However, in certain cases, they may perform at peer level in non-verbal tasks, such as object classification or basic math problems. Logical reasoning and transferring knowledge from one context to another may be challenging. Understanding abstract concepts in academic subjects and solving practical problems can also be difficult, as limited comprehension and reasoning skills complicate mastery of some subjects.

Memory in individuals with Down syndrome is characterized by hypomnesia (weakened memory), requiring more time to learn and retain new skills and information. Short-term auditory memory and auditory information processing are typically impaired. Attention is often unstable, and they may experience fatigue or a lack of energy, making it difficult to concentrate—they may become easily distracted. Their imaginative capacity is limited, and they perceive images more effectively through visual aids, yet may struggle with forming mental images. They may recognize individual parts of an image but find it challenging to integrate these parts into a complete picture.

Behaviorally, children with Down syndrome are often characterized by compliance, friendliness, and sometimes gentleness; they are usually eager to fulfill requests. Typically, they are sociable and engage with others readily, although a range of behavioral disorders may also be present.

### **Emotional Characteristics**

Children with Down syndrome generally retain the ability to express basic emotions. Many exhibit affection and form strong emotional bonds. Some display positive feelings and communicate with all adults, while others show a stronger interest in individuals they frequently interact with. They generally exhibit positive emotional states more often than negative ones. Typically, they do not become frustrated when facing difficulties and may lack an accurate self-assessment of their performance. Often, they enjoy the completion of a task regardless of its accuracy. They are capable of feeling fear, joy, and sadness, but their emotional responses may not always align with the stimulus, sometimes appearing exaggerated or understated.

Children with Down syndrome are often impressionable and tend to imitate others. Some may display epileptoid personality traits, such as egocentrism and excessive precision. However, most possess positive personality characteristics, often being friendly, balanced, and affectionate.

### **Development of Motor Coordination**

When comparing motor development stages between children with Down syndrome and typically developing children, recent studies indicate that children with Down syndrome are closer to typical developmental milestones than previously thought. Although some delays are present, the differences are not as pronounced as once believed.



Developmental Stages	Down Syndrome (Age in Months)	Typical (Age in Months)
Smiling	2 (1.5-3)	1 (0.5-3)
Rolling	6 (2-12)	5 (2-10)
Sitting	9 (6-18)	7 (5-9)
Crawling	11 (7-21)	8 (6-11)
Walking on all fours	13 (8-25)	10 (7-13)
Standing	10 (10-32)	11 (8-16)
Walking	20 (12-45)	13 (8-18)
Pronouncing Words	14 (9-30)	10 (6-14)
Forming Sentences	24 (18-46)	21 (14-32)

In the past, intellectual abilities of children with Down syndrome were underestimated. Recent studies contradict earlier beliefs that these children exhibit severe intellectual impairments. Research shows that most children with Down syndrome experience mild to moderate delays. Some display borderline or low-average cognitive functions, with only a small percentage having severe intellectual delays.

There is a misconception that the cognitive abilities of individuals with Down syndrome inevitably decline with age. However, long-term studies reveal that cognitive decline is not inevitable, offering a more hopeful outlook for their future.

### Conclusion

In recent decades, life expectancy for individuals with Down syndrome has significantly increased, due to advancements in medical care and social adaptation. Under favorable conditions, individuals with Down syndrome can live for 55 years or more. With timely educational intervention and support from specialists, children with Down syndrome can achieve substantial progress, realize their potential, learn, work, and lead active lives.

### References

1. СИНДРОМ ДАУНА. Текст научной статьи по специальности «Науки о здоровье». Григорьев К.И. Выхристюк О.Ф. Егоренков А.М.2010.
2. СЕСТРИНСКИЙ ПРОЦЕСС ПРИ СИНДРОМЕ ДАУНА У НОВОРОЖДЕННЫХ. Текст научной статьи по специальности «Науки о здоровье» Мерзлова Н.Б. Серова И.А. Ягодина А.Ю. 2013.
3. Респираторные заболевания у детей с синдромом Дауна. Текст научной статьи по специальности «Клиническая медицина» Беляшова М.А., Овсянников Дмитрий Юрьевич., Колтунов И.Е. 2013.
4. Особенности организации сестринского процесса при синдроме Дауна у новорожденных. Текст научной статьи по специальности «Науки о здоровье» Мерзлова Нина Борисовна. Серова И.А.Ягодина А.Ю. 2012.

