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# DOWN SYNDROME DISEASE AND ITS CAUSES OF ORIGIN

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**Annotation:** This article provides information on the history and causes of Down syndrome, symptoms of the disease, epidemiology, trisomy, diagnosis of the disease, prenatal diagnosis, Robertsonian translocations, the main factors for a child born with Down syndrome to be born with this disease and successful people with Down syndrome.

**Keywords:** Down syndrome, trisomy 21, disease, diagnosis, epidemiology, prenatal diagnosis, mosaicism, Robertsonian translocations

## INTRODUCTION

Down syndrome (trisomy 21) is a form of genomic pathology, in which the karyotype is often represented by 47 chromosomes instead of the normal 46. Because the 21st pair of chromosomes is present in three copies instead of the normal two. Translocation of chromosome 21 to other chromosomes (most often - 15, less often - 14, even less often - to 21, 22 and Y-chromosome) - 4%; mosaic variant of the syndrome - 5%

This syndrome is named after the English doctor John Down, who first described it in 1866. The connection between the occurrence of the congenital syndrome and a change in the number of chromosomes was established only by 1959 by the French geneticist Jerome Lejeune.

The word "syndrome" means a set of signs or characteristic features. When using this term, it is more correct to use the form "Down syndrome" rather than "Down disease".

International Day of People with Down Syndrome was first celebrated on March 21, 2006, at the initiative of the Greek geneticist Stilianos Antonarakis from the University of Geneva. The day and month were chosen depending on the pair number and the number of chromosomes.

HISTORY. The English doctor John Langdon Down was the first to describe the syndrome that later bore his name, described as a form of mental illness in 1862. This concept became widespread after the publication of a report on the subject in 1866. Due to the epicanthus, Down used the term Mongoloid (and the syndrome was called "Mongolism"). The concept of Down syndrome was strongly associated with racism until the 1970s.

Mete Rivolla of the University of Bordeaux discovered the remains of a child who lived about 1,500 years ago in a necropolis near a church in Chalon-sur-Saône and had characteristic features of Down syndrome, which is the oldest known case of this syndrome. Mete notes that the child's burial was not inferior to that of others, meaning that people suffering from this syndrome were not socially stratified.

In the 20th century, Down syndrome became a fairly common diagnosis. There are many people with Down syndrome, but only a few of the symptoms can be prevented. Most people with Down syndrome die in infancy or childhood.

There is currently no known cause of Down syndrome. A number of common factors have been identified that can lead to a baby being born with Down syndrome. These factors include:

Mother's Age. As the mother's age increases, the chance of having a baby with Down syndrome increases. However, women who give birth before the age of 35 can also have babies with Down syndrome. Women in their 40s are 40 times more likely to have a baby with Down syndrome than women in their 30s; Close relationship between the parents; Hereditary predisposition.

**SYMPTOMS OF DOWN SYNDROME**

Psychomotor and physical developmental delay; mental retardation; decreased or impaired muscle tone; short neck, excess skin on the back of the neck; flat face and flattened nasal bridge; small head, ears, and mouth; chest deformity; eyes that are tilted upwards, often with a skin fold extending from the upper eyelid and covering the inner corner of the eye; spots on the edges of the iris (called Brushfield spots); squinting; clouding of the pupil; open mouth (due to low muscle tone and the special structure of the tongue); enlarged tongue; large fingers due to underdevelopment of the middle phalanges; a single deep crease on the palm; defects in the interventricular septum of the heart; Girshprung's disease; congenital hypothyroidism. In addition, physical development in children with Down syndrome is often slower than in healthy children. For example, due to poor muscle tone, a child with Down syndrome may be very slow to learn to roll over, sit, stand, and walk. Despite these delays, children with Down syndrome can learn to do physical activities like other children. Children with Down syndrome may take longer than other children to complete all developmental milestones, but they eventually reach most of these milestones.

**DIAGNOSIS**

In most countries, prenatal screening is performed on pregnant women of all ages to detect trisomy 21. Screening is performed between 11 and 13 weeks of pregnancy. However, this method does not allow for an accurate diagnosis, and screening identifies pregnant women at high risk of having a baby with Down syndrome. The most effective way to detect this syndrome is through noninvasive prenatal testing, which involves analyzing fetal DNA from the mother's blood. This test can be performed at 9 weeks of pregnancy.

**EPIDEMIOLOGY**

Down syndrome is not a rare pathology - it is observed on average in 1 case out of 700 births. Currently, thanks to prenatal diagnosis, the frequency of births of children with Down syndrome has decreased from 1 case out of 1100, since abortion is resorted to when the fetus is diagnosed with the disease. The probability of developing an anomaly in a fetus of both sexes is the same.

trisomy

Trisomy is the presence of three homologous chromosomes instead of the normal pair. This condition occurs when chromosomes do not separate during meiosis, resulting in the formation of a gamete with 24 chromosomes. When fused with a normal gamete of the opposite sex, the zygote becomes 47 chromosomes. Trisomy of chromosome 21 causes Down syndrome in 95%

of cases, in which 88% of cases the non-disjunction of the maternal gamete and 8% of cases the paternal gamete plays a role.

robertson's translocations

The extra material on chromosome 21 that causes Down syndrome can occur due to a Robertsonian translocation in the karyotype of one of the parents. In this case, the long arm of chromosome 21 is attached to the arm of another chromosome (most often 14 [45, XX, der (14; 21) (q10; q10)]). The phenotype of a person with a Robertsonian translocation is normal. During reproduction, normal meiosis increases the likelihood of having a child with trisomy 21 and Down syndrome. Translocation with Down syndrome is usually called "familial Down syndrome". This form

does not depend on the age of the mother. This type of syndrome accounts for 2-3% of all cases.

prenatal diagnosis

triple combination test in the first trimester of pregnancy

At 11-14 weeks of pregnancy, a pregnant woman is referred to a medical institution, where she undergoes a comprehensive prenatal (prenatal) diagnostic examination for developmental disorders of the child. The risk is calculated based on three indicators, taking into account the woman's age:

> Pregnancy associated plasma protein-A (PAPP-A);

> Free B-subunit of human chorionic gonadotropin (B-OXG);

> Ultrasound signs (increased fluid volume in the cervical cavity, short nasal bones, short femurs, changes in brain structure, etc.).

The above methods do not allow for an accurate diagnosis, and as a result of the examination performed, the probability of pregnant women having a fetus with Down syndrome is assessed. At the second stage, an invasive procedure is performed to obtain fetal material necessary for an accurate analysis of Down syndrome in the fetus of women at risk. Depending on the duration of pregnancy, this material can be chorionic villus sampling (8-12 weeks), amniocentesis (14-18 weeks) or cordocentesis (later). The chromosome set is determined in tissue samples taken from the fetus.

The main factor in the birth of a child with Down syndrome is the mother's age. This was reported by the press service of the Medical Genetics Research Center of the Russian Academy of Sciences. Experts say that racial origin or socio-economic conditions do not affect the birth of a fetus with this genetic abnormality. However, traditional and religious characteristics of the region may play a role, since in areas where abortion is not accepted, the number of children with this abnormality increases. By the age of 40, women are 9 times more likely to have a child with Down syndrome than those who are 30.

According to recent studies, the life expectancy of people with Down syndrome has increased significantly in recent decades due to the development of medicine. March 21 is celebrated as International Day of People with Down Syndrome. This disease occurs as a result of a genetic defect. In this case, an extra chromosome appears.

She became the world's first professional model with Down syndrome. In childhood, like other patients with this diagnosis, the girl suffered from excess weight. But Madeleine really wanted to become a model, so she managed to lose 20 kilograms. She refused high-calorie foods, swam in the pool, danced and worked out. At the age of 18, she went with her mother to a fashion show in Brisbane, after which she firmly decided to become a model. The media wrote about her dream. In 2015, Madeleine's mother did everything she could to secure her a modeling contract and opened a company. After some time, she began to receive offers. He has appeared on the catwalk of New York Fashion Week several times, participated in photo shoots. He now lives with his mother in Brisbane.

paul pineda

Pablo is 44 years old, but by this time he has managed to achieve a lot. The boy discovered his uniqueness at the age of 7. He studied at a regular school, but was always outside. Because of his illness, other children avoided him, but he did not despair. After graduating from school, he was able to enter the university and graduated with a bachelor's degree. He received a degree in "Psychopedagogy". Pablo decided to become a teacher, he was the first teacher in Europe with such a diagnosis. In 2009, the film "Me Too" was released, in which Pinedo played the main role, for which he received the "Silver Shell", as well as the "City Shield" from the mayor. Now he lives in his hometown of Malaga (Spain), is engaged in charity work, helps people with Down syndrome find jobs. He gives lectures and teaches.

## CONCLUSION

The fact that the average life expectancy of people with Down syndrome is lower than that of people with a standard set of chromosomes is explained by their high morbidity. A study conducted in the United States in 2002 showed that the average life expectancy of people with Down syndrome is 49 years.

However, life expectancy today is significantly higher than it was in the 1980s, when it was 25 years. Over time, the causes of death have also changed, with chronic neurodegenerative diseases becoming more common as the population ages. Most people with this syndrome suffer from Alzheimer's disease - dementia at the age of 40-50.

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