

## DOWN SYNDROME

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named after Abu Ali ibn Sino

### Abstract:

Down syndrome (trisomy of chromosome 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.

**Key words:** down syndrome, cognitive, mother, children

There are two more forms of this syndrome:

Translocation of chromosome 21 to other chromosomes (often — 15, less often — 14, even less often — 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 origin of the congenital syndrome and the change in the number of chromosomes was determined only by 1959 by the French geneticist Jerome Lejeune. In youth slang, "Down" is used to denigrate people who are just stupid.

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

International Down Syndrome Day was first celebrated on March 21, 2006 at the initiative of Stilianos Antonarakisa, a Greek geneticist from the University of Geneva. The day and month were selected based on the pair number and the number of chromosomes (21.03).

### EPIDEMIOLOGY

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

#### Epidemiology of Down syndrome

A graph showing the probability of developing Down syndrome depending on the age of the mother the number of children born with Down syndrome is 1 in every 800 or 1000 babies. In 2006, the Centers for Disease Control and Prevention estimated it to be one case per 733 live births in the United States (5,422 new cases per year). About 95% of them are trisomy of chromosome 21. Down syndrome can occur in all ethnic groups and all economic classes.

The age of the mother affects the probability of conceiving a child with Down syndrome:

If mothers are between the ages of 20 and 24, the odds are 1 in 1,562;

under 30 years old - 1 per 1000;

35 to 39 years old — 214 to 1;

Over the age of 45, the odds are 19 to 1.

Despite the fact that the probability increases with the age of the mother, 80% of children with this syndrome are born to women under the age of 35. This situation is related to the high birth rate in this age group. According to the latest data, the age of the father, especially if he is over 42 years old, increases the risk of developing the syndrome in the child.

A 2008 study found that Down syndrome is also due to random events in gamete formation and/or pregnancy. Behavior of parents and environmental factors do not affect it.

#### TREATMENT

There is no cure for Down syndrome, but there are many ways to help patients learn social and other skills. However, scientists have not left aside the study of medicinal products. Epigallocatechin gallate, a substance found in green tea, is an example of the most recent of such agents.

Cognitive impairment in patients is thought to be related to the product produced by the DYRK1A gene. The above substance has been shown to improve memory and learning by leading to the expression of this gene in mice. But such experiments have not been proven to help.

Children with Down syndrome are usually divided into special classes at school. This is due to their low learning ability and the possibility of falling behind their peers.

In some European countries, such as Germany and Denmark, there is a "two-teacher" system, where the second teacher teaches children with communication difficulties. Importantly, it is all held in one classroom, which makes it easier for children to get along with others.

As an alternative to the "two teachers" system, there are cooperation programs between special and secondary schools. For children who have difficulty learning the essence of these programs, the main lessons are held in other rooms, and meals, walks, physical education, and breaks are done together.

There have been cases of people with Down syndrome graduating from university (Pablo Pineda, Aya Iwamoto).

#### THE END

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

However, life expectancy today is significantly higher than the 25 years in the 1980s. Over time, the causes of death have also changed, and chronic neurodegenerative diseases are more common in relation to the age of the population. At the age of 40-50, most people with this syndrome suffer from Alzheimer's disease - dementia.

## VACCINATION

Down's disease, perinatal encephalopathy, cerebral palsy and other stable neurological conditions are not considered contraindications for vaccination.

## THE ROLE OF THE HIST GENE IN EXPERIMENTAL PREVENTION OF DOWN SYNDROME

In July 2013, a link to an article in the journal "Nature" was published about an in vitro experiment conducted by American researchers from the Faculty of Medicine of the Massachusetts State University, led by Gene Loren.

During the experiment, the Xist gene, which is responsible for the inactivation of the X chromosome, was transferred to chromosome 21 of pluripotent stem cells with trisomy on chromosome 21. In this way, blocking of the third, redundant chromosome was achieved. Blocking it is thought to prevent the development of Down syndrome in the future.

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