

# What is Down Syndrome?

Down syndrome is the most commonly occurring chromosomal condition. One in every 691 babies in the United States is born with Down syndrome and it is found in people of all races and economic levels.

A few of the common physical traits of Down syndrome are low muscle tone, small stature, an upward slant to the eyes and a single deep crease across the center of the palm. Every person with Down syndrome is a unique individual and may possess these characteristics to different degrees or not at all.

People with Down syndrome have an increased risk for certain medical conditions such as congenital heart defects, respiratory and hearing problems, Alzheimer's disease, childhood leukemia and thyroid conditions. However, many of these conditions are now treatable, so most people with Down syndrome lead healthy lives. Life expectancy for people with Down syndrome has increased dramatically in recent decades - from 25 in 1983 to 60 today.

People with Down syndrome experience cognitive delays, but the effect is usually mild to moderate and is not indicative of the many strengths and talents that each individual possesses. Children with Down syndrome learn to sit, walk, talk, play and do most other activities, though somewhat later than their peers without Down syndrome.

Quality educational programs, a stimulating home environment, good health care and positive support from family, friends and communities enable people with Down syndrome to realize their aspirations and lead fulfilling lives. People with Down syndrome attend school, work and contribute to society in many wonderful ways.

People with Down syndrome should always be referred to as people first. Instead of "a Down syndrome child," it should be "a child with Down syndrome." Also avoid "Down's child" and describing the condition as "Down's," as in, "He has Down's." People "have" Down syndrome, they do not "suffer from" it and are not "afflicted by" it.

# What Causes Down Syndrome?

In every cell in the human body there is a nucleus where genetic material is stored in genes. Genes carry the codes responsible for all of our inherited traits and are grouped along rod-like structures called chromosomes. Normally, the nucleus of each cell contains 23 pairs of chromosomes, half of which are inherited from each parent.

Down syndrome is usually caused by an error in cell division called nondisjunction. Nondisjunction results in an embryo with three copies of chromosome 21 instead of the usual two. Prior to or at conception, a pair of 21st chromosomes in either the sperm or the egg fails to separate, passing on both copies of the 21st chromosome instead of the typical one. As the embryo develops, the extra chromosome is replicated in every cell of the body. This type of Down syndrome, which accounts for 95% of all cases, is called trisomy 21.

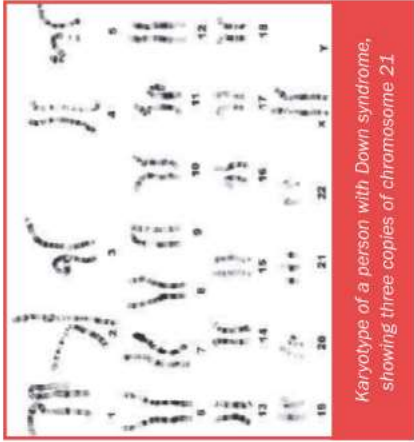
The two other types of Down syndrome are called mosaicism and translocation. Mosaicism (or mosaic Down syndrome) occurs when nondisjunction of chromosome 21 takes place in one (but not all) of the initial cell divisions after fertilization. When this occurs, there is a mixture of two types of cells, some containing the usual 46 chromosomes and others containing 47. Those cells with 47 chromosomes contain an extra chromosome 21. Mosaicism accounts for about 1% of all cases of Down syndrome. Research has indicated that individuals with mosaic Down syndrome may have fewer characteristics of Down syndrome than those with trisomy 21 or translocation Down syndrome. However, broad generalizations are not possible due to the wide range of abilities people with Down syndrome possess.

Translocation accounts for about 4% of all cases of Down syndrome. In translocation, an additional full or partial copy of chromosome 21 breaks off during cell division and attaches to another chromosome, typically chromosome 14. While the total number of chromosomes in the cells remains 46, the presence of an additional full or partial chromosome 21 causes the characteristics of Down syndrome.

Regardless of the type of Down syndrome a person may have, all people with Down syndrome have an extra, critical portion of chromosome 21 present in all or some of their cells. This additional genetic material alters the course of development and causes the characteristics associated with Down syndrome.

While the cause of nondisjunction is currently unknown, research has shown that the likelihood of it occurring increases as a woman ages. However, due to higher birth rates in younger women, 80% of children with Down syndrome are born to women under 35 years of age. Down syndrome is not caused by environmental factors or the parents' activities before or during pregnancy.

Once a woman has given birth to a baby with trisomy 21, it is estimated that her chance of having another baby with trisomy 21 is 1 in 100 (up to age 40). The probability of having a baby with Down syndrome increases substantially after age 40, regardless of whether a mother has already had a baby with Down syndrome. Unlike in trisomy 21 or mosaicism, however, the age of the mother does not seem to be linked to the risk of having a child with translocation Down syndrome..



# How is Down Syndrome Diagnosed?

Down syndrome is usually identified at birth by physical traits. These features may be present in babies who do not have Down syndrome, so a karyotype chromosomal analysis is done to make a diagnosis. To obtain a karyotype, doctors draw blood and photograph the chromosomes within the cells. They group them by size, number, and shape. Down syndrome is diagnosed by examining the karyotype and identifying an additional full or partial copy of chromosome 21. A similar genetic test called fluorescence in situ hybridization (FISH) can also confirm a diagnosis.

There are two types of tests for Down syndrome that can be performed before a baby is born: screening tests and diagnostic tests. Prenatal screens estimate the chance of the fetus having Down syndrome. Diagnostic tests can provide a definitive diagnosis with almost 100% accuracy.

Most screening tests involve a blood test and an ultrasound (sonogram). The blood tests (or serum screening tests) measure quantities of substances in the mother's blood. Together with the mother's age, the tests are used to estimate her chance of having a child with Down syndrome. Serum screening tests are often performed in conjunction with a detailed sonogram to check for markers (characteristics that some researchers feel may have a significant association with Down syndrome). Researchers have developed a maternal serum/ultrasound/age combination that yields higher accuracy at an earlier stage in the pregnancy. New noninvasive prenatal screening tests like MaterniT21 and Harmony involve blood being taken from the expectant mother at as early as 10 weeks of gestation and rely on the detection of cell-free DNA that circulates between the fetus and the expectant mother.

Prenatal screening tests are now routinely offered to women of all ages. If the prenatal screening shows a high chance of the child being born with Down syndrome, doctors will often advise a mother to undergo diagnostic testing to get a definitive diagnosis. The diagnostic procedures available for prenatal diagnosis of Down syndrome are chorionic villus sampling (CVS) and amniocentesis. CVS is usually performed in the first trimester between 9 and 11 weeks, and amniocentesis is usually performed in the second trimester after 15 weeks of gestation. These procedures, which carry up to a 1% risk of causing a miscarriage, are practically 100% accurate in diagnosing Down syndrome.

Maternal Age	Incidence of Down syndrome	Maternal Age	Incidence of Down syndrome	Maternal Age	Incidence of Down syndrome
20	1 in 2,000	30	1 in 900	40	1 in 100
21	1 in 1,700	31	1 in 800	41	1 in 80
22	1 in 1,500	32	1 in 720	42	1 in 70
23	1 in 1,400	33	1 in 600	43	1 in 50
24	1 in 1,300	34	1 in 450	44	1 in 40
25	1 in 1,200	35	1 in 350	45	1 in 30
26	1 in 1,100	36	1 in 300	46	1 in 25
27	1 in 1,050	37	1 in 250	47	1 in 20
28	1 in 1,000	38	1 in 200	48	1 in 15
29	1 in 950	39	1 in 150	49	1 in 10