NATIONAL DOWN SYNDROME SOCIETY

down syndrome

national down syndrome society

ndss

Education. Research. Advocacy.

Down syndrome affects people of all

ages, races and economic levels.

It is one of the most

frequently occurring chromosomal

abnormalities, occurring once in every

800 to 1,000 live births. More than

350,000 people in the United States

alone have Down syndrome.

When was Down Syndrome Discovered?

For centuries, people with Down syndrome have been alluded to in art, literature and science. It wasn't until the late nineteenth century, however, that John Langdon Down, an English physician, published an accurate description of a person with Down syndrome. It was this scholarly work, published in 1866, that earned Down the recognition as the "father" of the syndrome. Although other people had previously recognized the characteristics of the syndrome, it was Down who described the condition as a distinct and separate entity.

In recent history, advances in medicine and science have enabled researchers to investigate the characteristics of people with Down syndrome. In 1959, the French physician Jerome Lejeune identified Down syndrome as a chromosomal anomaly. Instead of the usual 46 chromosomes present in each cell, Lejeune observed 47 in the cells of individuals with Down syndrome. It was later determined that an extra partial or complete chromosome 21 results in the characteristics associated with Down syndrome. In May of 2000, an international team of scientists successfully identified and catalogued each of the approximately 329 genes on chromosome 21. This accomplishment opened the door to great advances in Down syndrome research.

Why is Down Syndrome Referred to as a Genetic Condition?

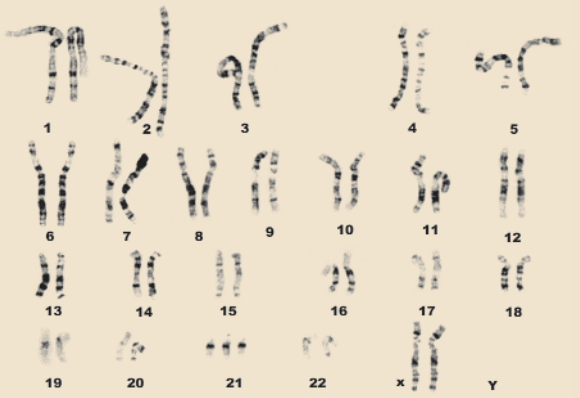
The human body is made of cells; all cells contain a center, called a nucleus, in which genetic material is stored. This genetic material, known as genes, carries the codes responsible for all our inherited characteristics. Genes are grouped along rod-like structures called chromosomes. Normally, the nucleus of each cell contains 23 pairs of chromosomes, half of which are inherited from each parent.

In individuals with Down syndrome, however, the cells usually contain 47, not 46, chromosomes; the extra chromosome is the 21st. This excess genetic material, in the form of additional genes along chromosome 21, results in Down syndrome. The extra 21st chromosome is detected by using a procedure called a karyotype. A karyotype is a visual display of the chromosomes grouped by size, number and shape. Chromosomes may be studied by examining blood or tissue cells. Individual chromosomes are identified, stained and numbered from largest to smallest. Ninety-five percent of occurrences of Down syndrome result from the presence of an extra (third) chromosome, a condition described as Trisomy 21.

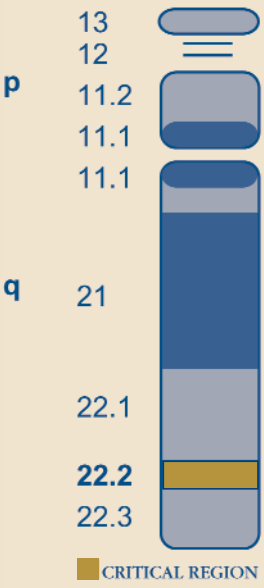
What Causes Down Syndrome?

Down syndrome is usually caused by an error in cell division called nondisjunction. However, two other types of chromosomal abnormalities, mosaicism and translocation, are also implicated in Down syndrome — although to a much lesser extent. 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 the syndrome.

TRISOMY 21 KARYOTYPE (FEMALE)



CHROMOSOME 21 MAP

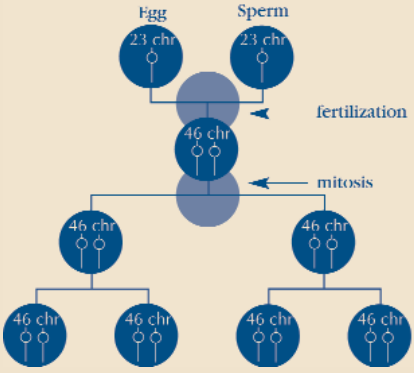


The indicated region of chromosome 21 appears to be associated with many of the characteristics linked to Down syndrome.

Nondisjunction

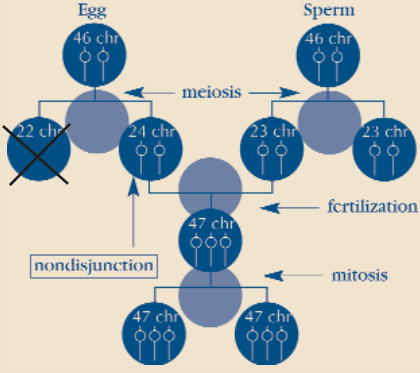
Nondisjunction is a faulty cell division that results in an embryo with three copies of chromosome 21 instead of the usual two. Prior to or at conception, a pair of 21st chromosomes in either the sperm or the egg fails to separate. As the embryo develops, the extra chromosome is replicated in every cell of the body. This error in cell division is responsible for 95 percent of all cases of Down syndrome.

NORMAL CELL DIVISION



During fertilization, the 23 chromosomes from the egg and sperm combine. The resulting fertilized egg has 46 chromosomes. During mitosis, the cell replicates itself and divides into two cells with 46 chromosomes in each.

NONDISJUNCTION



Nondisjunction is the failure of the pair of chromosomes to separate during meiosis, which is the process by which egg and sperm cells replicate themselves and divide. Nondisjunction results in both 21st chromosomes being carried to one cell and none to the other.

Why nondisjunction occurs is currently unknown, although research has shown that it increases in frequency as a woman ages. However, many people are surprised to find out that 80 percent of children born with Down syndrome are born to women under 35 years of age. This occurs because younger women have higher fertility rates. It does not contradict the fact that the incidence of births of children with Down syndrome dramatically increases with the age of the mother.

Once a woman has given birth to a baby with Down syndrome, it is estimated that the risk of having a second child with Down syndrome is about one in 100. However, the age of the mother may also be a risk factor.

INCIDENCE OF DOWN SYNDROME AND MATERNAL AGE

Maternal Age	Incidence of Down Syndrome
20	1 in 2000
24	1 in 1300
27	1 in 1050
30	1 in 900
33	1 in 600
36	1 in 300
40	1 in 100
42	1 in 70
45	1 in 30
47	1 in 20
48	1 in 15
49	1 in 10

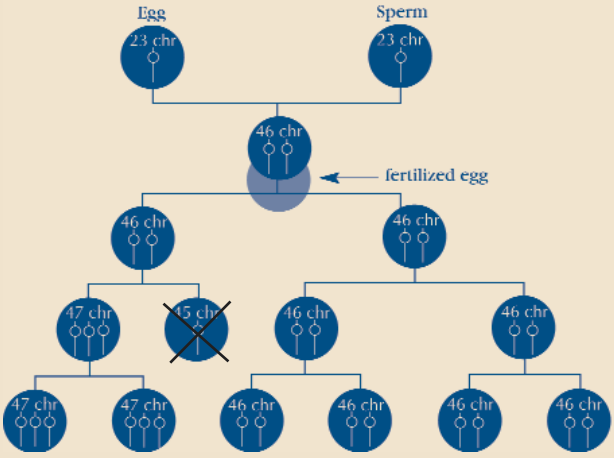
Although nondisjunction can be of paternal origin, it is much less common. It has been suggested that environmental factors may cause nondisjunction. However, despite years of research, the cause (or causes) of nondisjunction is still unknown. There is no scientific evidence that environmental factors or the parents' activities before or during pregnancy have an effect on any of the three types of Down syndrome.

Mosaicism

Mosaicism occurs when nondisjunction of chromosome 21 takes place in one of the initial cell divisions after fertilization. When this occurs, there is a mixture of two types of cells, some containing 46 chromosomes and some containing 47. Those cells with 47 chromosomes contain an extra chromosome 21. Because of the "mosaic" pattern of the cells, the term mosaicism is used. Mosaicism is rare, responsible for only one to two percent of all cases of Down syndrome.

Research has indicated that individuals with mosaic Down syndrome may be less affected by the physical and mental characteristics of Down syndrome than those with nondisjunction or translocation; however, broad generalizations are not possible due to the wide range of abilities that people with Down syndrome possess.

MOSAICISM

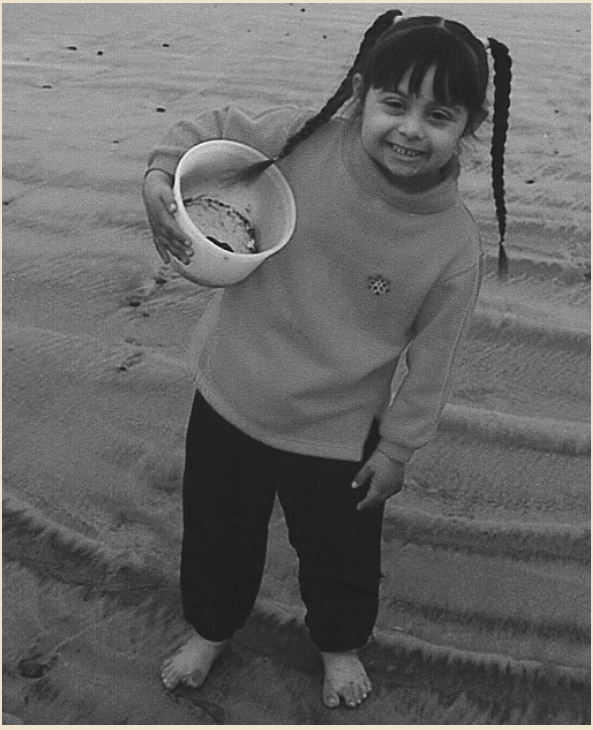


Mosaicism occurs after the fertilized egg begins to divide normally. Nondisjunction occurs in one cell line resulting in an individual with a combination of both typical and Trisomy 21 cell lines.

Translocation

Translocation accounts for only three to four percent of cases of Down syndrome. In translocation, part of chromosome 21 breaks off during cell division and attaches to another chromosome. While the total number of chromosomes in the cells remain 46, the presence of an extra part of chromosome 21 causes the characteristics of Down syndrome.

As with nondisjunction, translocation occurs either prior to or at conception. But, unlike nondisjunction, maternal age is not linked to the risk of translocation. Most cases are sporadic, chance events. However, in about one third of translocation incidents, one parent is a carrier of a translocated chromosome. For this reason, the risk of recurrence of translocation in a subsequent child is higher than that of nondisjunction. Genetic counseling can determine the origin of translocation.



Are Prenatal Tests Available to Detect Down Syndrome?

There are two types of tests available to pregnant women: screening and diagnostic. Screening tests estimate the risk of the fetus having Down syndrome; diagnostic tests tell whether or not the fetus actually has the condition.

Currently, the most common screening test is the Triple Screen, a combination of three tests that measure quantities of various substances in the blood (alpha-fetoprotein, human chorionic gonadotropin and unconjugated estriol) and together with the woman's age, estimate her risk of having a child with Down syndrome. These screening tests are typically offered between 15 and 20 weeks of gestation.

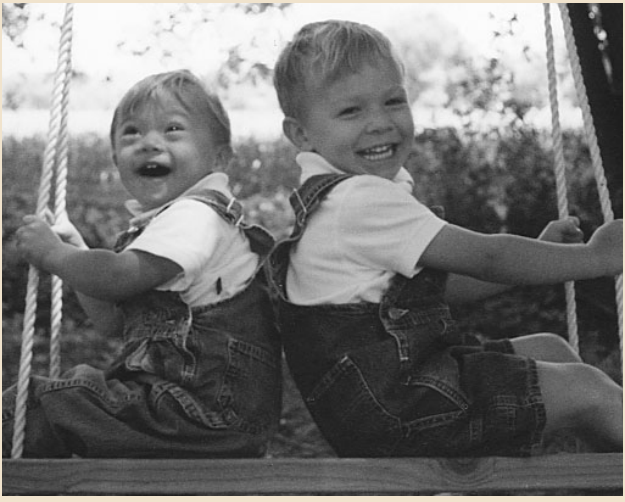
A "quad" test that measures an additional substance, inhibin-A, has recently been introduced in an attempt to provide a more accurate screening.

Screenings are often performed in conjunction with a detailed sonogram. Sonograms can show some physical or developmental traits that are helpful in calculating the risk of Down syndrome. However, these tests alone cannot provide a sufficient diagnosis of Down syndrome. It is important to note that screening tests are of limited value. Some women who undergo these tests will be given false positive readings, and some will be given false negative readings. The Triple Screen test has a five to eight percent false positive rate and also has a false negative rate of 35 to 40 percent, detecting about 60 to 65 percent of all incidences of Down syndrome.

The procedures available for prenatal diagnosis of Down syndrome are amniocentesis, chorionic villus sampling (CVS) and percutaneous umbilical blood sampling (PUBS). Each of these procedures carries

a small risk of miscarriage because tissue must be extracted from the placenta or the umbilical cord to examine the fetal chromosomes. The procedures are 98 to 99 percent accurate in the detection of Down syndrome. Amniocentesis is usually performed between 12 and 20 weeks of gestation, CVS between eight and 12 weeks and PUBS after 20 weeks.

Improvements in the sensitivity and predictive value of prenatal tests are ongoing. It is important to consult a medical professional or genetic counselor for the most up-to-date and appropriate information.



How is Down Syndrome Diagnosed in a Newborn?

The diagnosis of Down syndrome is usually suspected after birth as a result of the baby's appearance. There are many physical characteristics that form the basis for suspecting an infant has Down syndrome. Many of these characteristics are found to some extent in the general population. Therefore, if Down syndrome is suspected, a karyotype will be performed to determine the diagnosis.

Some infants with Down syndrome have only a few of these traits, while others have many. Among the most common traits are:

- Muscle hypotonia – low muscle tone
- Flat facial profile – a somewhat depressed nasal bridge and a small nose
- Oblique palpebral fissures – an upward slant to the eyes
- Dysplastic ear – an abnormal shape of the ear
- Single palmar crease – a single deep crease across the center of the palm
- Hyper-flexibility – an excessive ability to extend the joints
- Curvature of the fifth finger, caused by under development of the middle phalanx (bone)
- Epicanthal folds – small skin folds on the inner corner of the eyes
- Excessive space between first and second toe
- Large tongue in relation to size of mouth

Coupled with the natural stresses of childbirth, awaiting a diagnosis can be a particularly difficult time for parents. Although there is no easy way to be informed, most families agree that the best way to proceed includes having the baby and family members present, and being told as soon as possible. Local Down syndrome support groups can be a valuable resource for families during this time. Please contact NDSS to find the closest support group.

Do People with Down Syndrome have Health Problems?

People with Down syndrome are at increased risk for certain health problems. While there is an increased risk for certain medical conditions compared to the general population, advances in medicine have rendered most of these health problems treatable and most people with Down syndrome lead healthy lives.

Congenital heart defects, increased susceptibility to infection, respiratory and hearing problems, obstructed digestive tracts, sleep apnea and childhood leukemia occur with greater frequency in children with Down syndrome. Adults with Down syndrome are also at increased risk for Alzheimer's disease, thyroid conditions and sleep apnea. The majority of people born with Down syndrome today have an average life expectancy of 55 years, with some living into their seventies.

Resources such as the Down Syndrome Health Care Guidelines and specialized growth charts can further assist families and medical professionals in providing appropriate medical and preventative care. These documents can be obtained from NDSS.

How Does Down Syndrome Affect Development?

All people with Down syndrome experience cognitive delays, however, 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, toilet train and do most other activities — only somewhat later than their peers without Down syndrome.

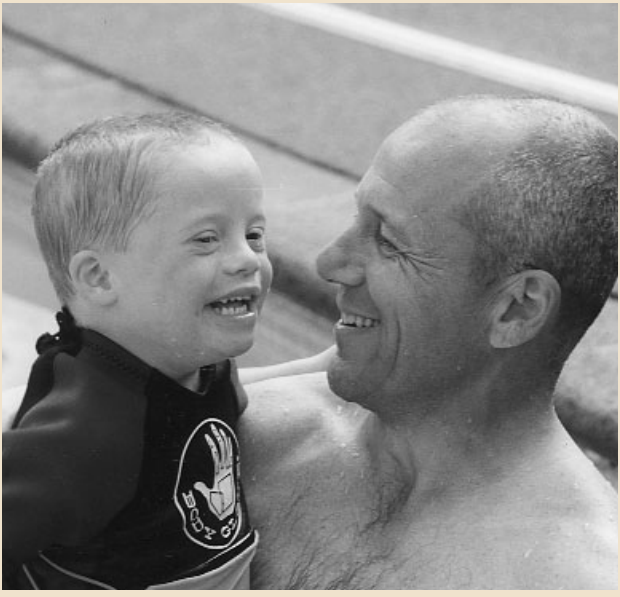
DEVELOPMENTAL MILESTONES

Milestone	Down Syndrome Range	Typical Range
Gross Motor		
Sits Alone	6-30 Months	5-9 Months
Crawls	8-22 Months	6-12 Months
Stands	12-39 Months	8-17 Months
Walks Alone	12-48 Months	9-18 Months
Language		
First Word	12-48 Months	8-23 Months
Two-Word Phrases	2-7 1/2 Years	15-32 Months
Personal/Social		
Responsive Smile	1 1/2 -5 Months	1-3 Months
Finger Feeds	10-24 Months	7-14 Months
Drinks From Cup	12-32 Months	9-17 Months
Uses Spoon	13-39 Months	12-20 Months
Bowel Control	2-7 Years	16-42 Months
Dresses Self*	3 1/2 -8 1/4 Years	3 1/4 -5 Years

**unassisted*

Early intervention services, which begin shortly after birth, help children with Down syndrome develop to their full potential. The physical, speech and occupational therapies that early intervention programs provide can enhance a child's development and provide a way for parents to track their child's progress. Quality educational programs, along with a stimulating home environment and good medical care, enable people with Down syndrome to lead fulfilling lives.

People with Down syndrome are highly responsive to their physical and social environments. A person with Down syndrome has the same emotions and needs as any other person and deserves the same opportunities. Those who receive positive support and are included in the activities of the community can be expected to adapt successfully — to attend school, make friends, find work, participate in decisions that affect them and contribute to society.



What Does the Future Hold for People with Down Syndrome?

People with Down syndrome are people first. They may have some of the characteristics generally associated with this condition, but they are overwhelmingly unique and must be treated as individuals. Over the past few decades, progressive legislation such as Section 504 of the Rehabilitation Act of 1973, the Individuals with Disabilities Education Act and the Americans with Disabilities Act has recognized that people with Down syndrome and other disabilities are individuals and citizens, and are entitled to equal protection and opportunity under the law.

Ensuring equal treatment and access to services is a struggle that every family of a child with Down syndrome faces. Daily, these individuals strive to accomplish the same goals as everyone else: self-fulfillment, pride in achievement, inclusion in the activities of the community and the opportunity and challenge of reaching one's full potential. With the emergence of the self-advocacy and self-determination movements, people with Down syndrome are becoming actively involved in defining their own lives and negotiating their roles within community and family.

People with Down syndrome regularly participate in community activities: in schools, jobs, places of worship and leisure activities. Some live with family, some with friends or independently. They form interpersonal relationships and some marry. While rare, some people with Down syndrome have become parents.

The opportunities available to people with Down syndrome today have never been greater. However, it is only through the collective efforts of parents and family members, people with Down syndrome, professionals and concerned citizens that acceptance is becoming widespread.

About the National Down Syndrome Society

Our Vision: The National Down Syndrome Society envisions a world in which all people with Down syndrome have the opportunity to realize their life aspirations. NDSS is committed to being the national leader in supporting and enhancing the quality of life, and realizing the potential, of all people with Down syndrome.

Our Constituents: Today, there are 350,000 people in the United States living with Down syndrome. Each of these individuals touches the lives of family members, educators, employers, health care professionals, peers and others. This network of people who are affected by Down syndrome numbers in the millions.

Our Programs: **Education**. Through our model programs, award-winning Web site, national conferences, Information and Referral Center and more, NDSS provides targeted, in-depth information that meets the diverse needs of our constituents. **Research.** The Society hosts international research conferences and funds research both independently and through a partnership with the NIH. NDSS is the largest private funder of Down syndrome research in the U.S. **Advocacy.** In cooperation with our more than 165 affiliate groups, NDSS influences important local and federal legislation, established October as National Down Syndrome Awareness Month, organizes the national Buddy Walk program and much more.

NDSS thanks Basic Biomedical Research at the Eleanor Roosevelt Institute in Denver, CO, for providing the karyotypes and map of chromosome 21 in this brochure.

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