

Down Syndrome

What is Down syndrome?

Down syndrome is a chromosomal disorder that includes a combination of birth defects. Affected individuals have some degree of mental retardation, characteristic facial features and, often, heart defects and other health problems. The severity of these problems varies greatly among affected individuals.

How common is Down syndrome?

Down syndrome is one of the most common genetic birth defects, affecting about 1 in 800 babies (1). According to the National Down Syndrome Society, there are approximately 350,000 individuals with Down syndrome in the United States (2).

What causes Down syndrome?

Down syndrome is caused by extra genetic material from chromosome 21. Chromosomes are the structures in cells that contain the genes.

Each person normally has 23 pairs of chromosomes, or 46 in all. An individual inherits one chromosome per pair from the mother's egg and one from the father's sperm. When an egg and sperm cell join together, they normally form a fertilized egg with 46 chromosomes.

Sometimes something goes wrong before fertilization. A developing egg or sperm cell may divide incorrectly, sometimes resulting in an egg or sperm cell with an extra chromosome number 21. When this cell joins with a normal egg or sperm cell, the resulting embryo has 47 chromosomes instead of 46. Down syndrome also is called trisomy 21 because affected individuals have three number 21 chromosomes, instead of two. This type of error in cell division causes about 95 percent of the cases of Down syndrome (3).

Occasionally, before fertilization, a part of chromosome 21 breaks off during cell division and becomes attached to another chromosome in the egg or sperm cell. The resulting embryo may have what is called translocation Down syndrome. Affected individuals have two normal copies of chromosome 21 plus extra chromosome 21 material attached to another chromosome. This type of error in cell division causes about 3 to 4 percent of the cases of Down syndrome (3). In some cases, the parent has a rearrangement of chromosome 21, called a balanced translocation, which does not affect his or her health.

About 1 to 2 percent of individuals with Down syndrome have a form called mosaicism (3). In this form, the error in cell division occurs after fertilization. Affected individuals have some cells with an extra chromosome 21 and others with the normal number.

What health problems might a child or adult with Down syndrome have?

The outlook for individuals with Down syndrome is far brighter than it once was. Most of the health problems associated with Down syndrome can be treated, and life expectancy is now about 55 years (2). Individuals with Down syndrome are more likely than unaffected individuals to have one or more of the following health conditions:

- **Heart defects.** Almost half of babies with Down syndrome have heart defects (3). Some defects are minor and may be treated with medications, while others require surgery. All babies with Down syndrome should be examined by a pediatric cardiologist, a doctor who specializes in heart diseases of children, and have an echocardiogram (a special ultrasound examination of the heart) in the first two months of life so that heart defects can be treated (2, 3).
- **Intestinal defects.** About 12 percent of babies with Down syndrome are born with intestinal malformations that require surgery (3).
- **Vision problems.** More than 60 percent of children with Down syndrome have vision problems, including crossed eyes (esotropia), near- or far-sightedness and cataracts

- (3). Glasses, surgery or other treatments usually can improve vision. A child with Down syndrome should be examined by a pediatric ophthalmologist (eye doctor) within the first six months of life and have regular vision exams (3).
- **Hearing loss.** About 75 percent of children with Down syndrome have some [hearing loss](#) (3). Hearing loss may be due to fluid in the middle ear (which may be temporary), a nerve or both. Babies with Down syndrome should be screened for hearing loss at birth or by 3 months of age. They also should have regular hearing exams so any problems can be treated before they hinder development of language and other skills (3).
 - **Infections.** Children with Down syndrome tend to have many colds and ear infections, as well as bronchitis and pneumonia. Children with Down syndrome should receive all the standard [childhood immunizations](#), which help prevent some of these infections.
 - **Thyroid problems, leukemia and seizures (3)**
 - **Memory loss.** Individuals with Down syndrome are more likely than unaffected individuals to develop Alzheimer's disease (characterized by progressive memory loss, personality changes and other problems). Adults with Down syndrome tend to develop Alzheimer's disease at an earlier age than unaffected individuals. Studies suggest that about 25 percent of adults with Down syndrome over age 35 have symptoms of Alzheimer's disease (2).

Some individuals with Down syndrome may have a number of these problems, while others may have none. The severity of these conditions varies greatly.

[What does a child with Down syndrome look like?](#)

A child with Down syndrome may have eyes that slant upward and small ears that may fold over a little at the top. The child's mouth may be small, making the tongue appear large. The nose also may be small, with a flattened nasal bridge. Some babies with Down syndrome have short necks and small hands with short fingers. Having less muscle tone, they may appear somewhat "floppy."

The child or adult with Down syndrome is often short and has unusual looseness of the joints. Most children with Down syndrome have some, but not all, of these features.

[How serious is the mental retardation?](#)

The degree of mental retardation varies widely. Most fall within the mild to moderate range. With proper intervention, few will have severe mental retardation (3). There is no way to predict the mental development of a child with Down syndrome based upon physical features.

[What can a child with Down syndrome do?](#)

Children with Down syndrome usually can do most things that any young child can do, such as walking, talking, dressing and being toilet-trained. However, they generally start learning these things later than other children.

The exact age that these developmental milestones will be achieved cannot be predicted. However, early intervention programs beginning in infancy can help these children achieve their developmental milestones sooner.

[Can a child with Down syndrome go to school?](#)

Yes. There are special programs beginning in the preschool years to help children with Down syndrome develop skills as fully as possible. Along with benefiting from early intervention and special education, many children are integrated into the regular classroom. Many affected children learn to read and write, and some graduate from high school and go on to post-

secondary programs or college. Individuals with Down syndrome participate in diverse childhood activities both at school and in their neighborhoods.

While there are special work programs designed for adults with Down syndrome, many people with the disorder hold regular jobs. Today, an increasing number of adults with Down syndrome live semi-independently in community group homes where they take care of themselves, participate in household chores, develop friendships, partake in leisure activities and work in their communities.

Can Down syndrome be cured or prevented?

There is no cure for Down syndrome, nor is there any way to prevent it. However, some studies suggest that women who have certain variant genes that affect how their bodies metabolize (process) the B vitamin folic acid may be at increased risk for having a baby with Down syndrome (4, 5). If confirmed, this finding may provide yet another reason why all women who might become pregnant should take a daily multivitamin containing 400 micrograms of [folic acid](#) (which has been shown to reduce the risk of certain birth defects of the brain and spinal cord).

Does the risk of Down syndrome increase with the mother's age? Yes. The risk of Down syndrome increases from about 1 in 1,250 at age 25, to 1 in 1,000 at age 30, 1 in 400 at age 35, 1 in 100 at age 40 and 1 in 30 at age 45 (6). Women over age 35 have been traditionally considered most likely to have a baby with Down syndrome. However, about 80 percent of babies with Down syndrome are born to women who are under age 35, as younger women have far more babies (2).

What is the risk that parents of a child with Down syndrome will have another affected child?

In general, in each subsequent pregnancy, the chance of having another baby with Down syndrome is 1 percent plus whatever additional risk a mother has, based upon her age (2, 7). If, however, the first child has translocation Down syndrome, the chance of having another child with Down syndrome may be greatly increased.

After birth, the provider takes a blood sample from a baby suspected of having Down syndrome and sends it to a laboratory. The lab examines the chromosomes (called a karyotype) to determine if the baby has Down syndrome and what genetic form of Down syndrome the baby has. This information is important in determining the risk in future pregnancies. The doctor may refer parents to a [genetic counselor](#) who can explain the results of chromosomal tests in detail, including what the recurrence risks may be in another pregnancy.

Can Down syndrome be diagnosed before the child is born?

Yes. The American College of Obstetricians and Gynecologists (ACOG) recommends that all pregnant women be offered a screening test for Down syndrome, regardless of the woman's age. Screening may consist of a [maternal blood test](#) done in the first trimester (at 11 to 13 weeks of pregnancy), along with a special ultrasound examination of the back of the baby's neck (called nuchal translucency), or a maternal blood test done in the second trimester (at 15 to 20 weeks) (8). A screening test helps identify pregnancies that are at higher-than-average risk of Down syndrome. However, a screening test cannot diagnose Down syndrome or other birth defects.

Women who have an abnormal screening test result are offered a diagnostic test, such as [amniocentesis](#) or [chorionic villus sampling \(CVS\)](#). These tests are highly accurate at diagnosing, or more likely, ruling out Down syndrome.

ACOG also recommends that pregnant women of all ages have the option of bypassing the screening test and choosing a diagnostic test for Down syndrome instead (8). Until recently, only women over age 35 and others considered at increased risk for having a baby with Down

syndrome were offered diagnostic testing because amniocentesis and CVS pose a very small risk of miscarriage.

Most parents-to-be receive reassuring news from a screening or diagnostic test for Down syndrome. However, if a prenatal diagnostic test shows that the baby has Down syndrome, parents have an opportunity to prepare medically, emotionally and financially for the birth of a child with special needs, such as arranging for delivery in a medically appropriate setting.

Can people with Down syndrome have children?

Some people with Down syndrome marry. With rare exceptions, men with Down syndrome cannot father a child (3). In any pregnancy, a woman with Down syndrome has a 50-50 chance of conceiving a child with Down syndrome, but many affected fetuses are miscarried.

Is the March of Dimes conducting research on Down syndrome?

Some March of Dimes grantees are investigating why errors in chromosome division occur, in the hope of someday preventing Down syndrome and other birth defects caused by abnormalities in the number or structure of chromosomes. Other grantees are investigating the role of specific genes in causing the brain abnormalities associated with Down syndrome, with the goal of treating the mental retardation associated with the disorder. An international team of scientists has mapped all the genes of chromosome 21. This information eventually may pave the way for treatment of many features of this disorder.

Where can families affected by Down syndrome get additional information?

There are organizations across the country that provide information and support for families with children affected by Down syndrome. Two are:

National Down Syndrome Society

666 Broadway
New York, NY 10012
(800) 221-4602 or (212) 460-9330

National Down Syndrome Congress 1370 Center Drive, Suite 102

Atlanta, GA 30338
800 (232)-NDSC or (770) 604-9500

References

1. *Centers for Disease Control and Prevention (CDC). Birth Defects: Frequently Asked Questions. Updated 12/12/06.*
2. *National Down Syndrome Society. Information Topics. Accessed 1/11/07..*
3. *American Academy of Pediatrics Committee on Genetics. Health Supervision for Children with Down Syndrome. Pediatrics, volume 107, number 2, February 2001, pages 442-449.*
4. *O'Leary, V.B., et al. MTRR and MTHFR Polymorphism: Link to Down Syndrome? American Journal of Medical Genetics, January 15, 2002, volume 107, number 2, pages 151-155*