



Diseases and Conditions

Down syndrome

By Mayo Clinic Staff

Down syndrome is a genetic disorder caused when abnormal cell division results in extra genetic material from chromosome 21. This genetic disorder, which varies in severity, causes lifelong intellectual disability and developmental delays, and in some people it causes health problems.

Down syndrome is the most common genetic chromosomal disorder and cause of learning disabilities in children.

Better understanding of Down syndrome and early interventions can greatly increase the quality of life for children and adults with this disorder and help them live fulfilling lives.

Each person with Down syndrome is an individual — intellectual and developmental problems range from mild to moderate, and some people are healthy while others have severe health problems such as serious heart defects.

Children with Down syndrome have a distinct facial appearance. Though not all children with Down syndrome have the same features, some of the more common features are:

- Flattened facial features
- Small head
- Short neck
- Protruding tongue
- Upward slanting eyes, unusual for the child's ethnic group
- Unusually shaped or small ears
- Poor muscle tone
- Broad, short hands with a single crease in the palm
- Relatively short fingers and small hands and feet

- Excessive flexibility
- Tiny white spots on the colored part (iris) of the eye called Brushfield spots
- Short height

Infants with Down syndrome may be average size, but typically they grow slowly and remain shorter than other children the same age. In general, developmental milestones, such as sitting and crawling, occur at about twice the age of children without impairment.

Human cells normally contain 23 pairs of chromosomes. One chromosome in each pair comes from your father, the other from your mother.

Down syndrome results when abnormal cell division involving chromosome 21 occurs. These cell division abnormalities result in extra genetic material from chromosome 21, which is responsible for the characteristic features and developmental problems of Down syndrome. Any one of three genetic variations can cause Down syndrome:

- **Trisomy 21.** About 95 percent of the time, Down syndrome is caused by trisomy 21 — the child has three copies of chromosome 21 (instead of the usual two copies) in all cells. This is caused by abnormal cell division during the development of the sperm cell or the egg cell.
- **Mosaic Down syndrome.** In this rare form of Down syndrome, children have some cells with an extra copy of chromosome 21. This mosaic of normal and abnormal cells is caused by abnormal cell division after fertilization.
- **Translocation Down syndrome.** Down syndrome can also occur when part of chromosome 21 becomes attached (translocated) onto another chromosome, before or at conception. These children have the usual two copies of chromosome 21, but they also have additional material from chromosome 21 attached to the translocated chromosome.

There are no known behavioral or environmental factors that cause Down syndrome.

Is it inherited?

Most of the time, Down syndrome isn't inherited. It's caused by a mistake in cell division during the development of the egg, sperm or embryo.

Translocation Down syndrome is the only form of the disorder that can be passed from parent to child. However, only about 4 percent of children with Down syndrome have translocation. And only about one-third of these children inherited it from one of their parents.

When translocations are inherited, the mother or father has some rearranged genetic material, but no extra genetic material — this means he or she is a balanced carrier. A

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balanced carrier has no signs or symptoms of Down syndrome, but he or she can pass the translocation on to children, causing extra genetic material from chromosome 21.

The chance of passing on the translocation depends on the sex of the parent who carries the rearranged chromosome 21:

- If the father is the carrier, the risk is about 3 percent.
- If the mother is the carrier, the risk is between 10 and 15 percent.

Some parents have a greater risk of having a baby with Down syndrome. Risk factors include:

- **Advancing maternal age.** A woman's chances of giving birth to a child with Down syndrome increase with age because older eggs have a greater risk of improper chromosome division. By age 35, a woman's risk of conceiving a child with Down syndrome is about 1 in 350. By age 40, the risk is about 1 in 100, and by age 45, the risk is about 1 in 30. However, most children with Down syndrome are born to women under age 35 because younger women have far more babies.
- **Having had one child with Down syndrome.** Typically, a woman who has one child with Down syndrome has about a 1 in 100 chance of having another child with Down syndrome.
- **Being carriers of the genetic translocation for Down syndrome.** Both men and women can pass the genetic translocation for Down syndrome on to their children.

Children with Down syndrome can have a variety of complications, some of which become more prominent as they get older, such as:

- **Heart defects.** About half the children with Down syndrome are born with some type of heart defect. These heart problems can be life-threatening and may require surgery in early infancy.
- **Leukemia.** Young children with Down syndrome have an increased risk of leukemia.
- **Infectious diseases.** Because of abnormalities in their immune systems, those with Down syndrome are much more at risk of infectious diseases, such as pneumonia.
- **Dementia.** People with Down syndrome have a greatly increased risk of dementia — signs and symptoms may begin around age 50. Those who have dementia also have a higher rate of seizures. Having Down syndrome also increases the risk of developing Alzheimer's disease.
- **Sleep apnea.** Because of soft tissue and skeletal changes that lead to the obstruction of their airways, children and adults with Down syndrome are at greater risk of obstructive sleep apnea.
- **Obesity.** People with Down syndrome have a greater tendency to be obese compared with the general population.

- **Other problems.** Down syndrome may also be associated with other health conditions, including gastrointestinal blockage, thyroid problems, early menopause, seizures, ear infections, hearing loss, skin problems such as psoriasis, skeletal problems and poor vision.

Life expectancy

Life spans have increased dramatically for people with Down syndrome. In 1910, a baby born with Down syndrome often didn't live to age 10. Today, someone with Down syndrome can expect to live to age 60 and beyond, depending on the severity of health problems.

The American College of Obstetricians and Gynecologists recommends offering the option of screening tests and diagnostic tests for Down syndrome to all pregnant women, regardless of age.

- **Screening tests** can indicate the likelihood a mother is carrying a baby with Down syndrome.
- **Diagnostic tests** can identify whether your baby has Down syndrome.

Your health care provider can discuss the types of tests, advantages and disadvantages, benefits and risks, and the meaning of your results. If appropriate, your provider may recommend that you talk to a genetics counselor.

Screening tests during pregnancy

Screening for Down syndrome is offered as a routine part of prenatal care. Although screening tests aren't perfect, they can help you make decisions about more-specific diagnostic tests and the course of the pregnancy.

Various screening tests can help identify whether you have a high risk of carrying a baby with Down syndrome, but they can't identify whether your baby has Down syndrome. Screening tests include the first trimester combined test, the integrated screening test and the cell-free fetal DNA analysis.

The first trimester combined test

The first trimester combined test, which is done in two steps, includes:

- **Blood test.** This blood test measures the levels of pregnancy-associated plasma protein-A (PAPP-A) and the pregnancy hormone known as human chorionic gonadotropin (HCG). Abnormal levels of PAPP-A and HCG may indicate a problem with the baby.

- **Ultrasound.** Ultrasound is used to measure a specific area on the back of your baby's neck. This is known as a nuchal translucency screening test. When abnormalities are present, more fluid than usual tends to collect in this neck tissue.

Using your age and the results of the blood test and the ultrasound, your health care provider can estimate your risk of having a baby with Down syndrome.

Integrated screening test

The integrated screening test is done in two parts during the first and second trimesters of pregnancy. The results are combined to estimate the risk that your baby has Down syndrome. This test can achieve the same level of detection as the first trimester combined test but with a lower false-positive rate, meaning that fewer women are incorrectly identified as carrying a baby with Down syndrome.

- **First trimester.** Part one includes a blood test to measure PAPP-A and an ultrasound to measure nuchal translucency.
- **Second trimester.** The quad screen measures your blood level of four pregnancy-associated substances: alpha fetoprotein, estriol, HCG and inhibin A.

Cell-free fetal DNA analysis

The cell-free fetal DNA test checks for fetal DNA circulating in the mother's blood. This test is usually recommended for women who have a higher risk of having a baby with Down syndrome or in response to risk detected by one of the previous tests. The mother's blood can be tested during pregnancy after 10 weeks gestation.

This test appears to be much more specific than other screening methods for Down syndrome. If this screening test indicates a high risk of Down syndrome, a more invasive diagnostic test may be used to determine whether your baby actually has Down syndrome.

Diagnostic tests during pregnancy

If your screening test results are positive or worrisome, or you're at high risk of having a baby with Down syndrome, you might consider more testing to confirm the diagnosis. Your health care provider can help you weigh the pros and cons of these tests.

Diagnostic tests that can identify Down syndrome include:

- **Amniocentesis.** A sample of the amniotic fluid surrounding the fetus is withdrawn through a needle inserted into the mother's uterus. This sample is then used to analyze the chromosomes of the fetus. Doctors usually perform this test in the second trimester, after 15 weeks of pregnancy. The test carries a slight risk of miscarriage, but risk increases if it's done before 15 weeks.

- **Chorionic villus sampling (CVS).** In CVS, cells are taken from the placenta and used to analyze the fetal chromosomes. Typically performed in the first trimester, after 10 weeks of pregnancy, this test appears to carry a somewhat higher risk of miscarriage than second trimester amniocentesis.
- **Cordocentesis.** In this test, also known as percutaneous umbilical blood sampling or PUBS, fetal blood is taken from a vein in the umbilical cord and examined for chromosomal defects. Doctors can perform this test between 18 and 22 weeks of pregnancy. This test carries a significantly greater risk of miscarriage than does amniocentesis or CVS, so it's only offered when results of other tests are unclear and the desired information can't be obtained any other way.

Preimplantation genetic diagnosis is one option available for couples undergoing in vitro fertilization who are at increased risk of passing along certain genetic conditions. The embryo is tested for genetic abnormalities before it's implanted in the womb.

Diagnostic tests for newborns

After birth, the initial diagnosis of Down syndrome is often based on the baby's appearance. But the features associated with Down syndrome can be found in babies without Down syndrome, so your health care provider will likely order a test called a chromosomal karyotype. Using a sample of blood, this test analyzes your child's chromosomes. If there's an extra chromosome 21 present in all or some cells, the diagnosis is Down syndrome.

Early intervention for infants and children with Down syndrome can make a major difference in realizing their potential abilities and in their quality of life.

Early intervention programs

Ask your health care provider about early intervention programs in your area. Available in most states, these special programs offer children with Down syndrome stimulation at an early age with appropriate sensory, motor and cognitive activities.

Programs may vary, but they usually involve therapists and special educators whose goal is to help your baby develop motor skills, language, social skills and self-help skills.

Team care

If your child has Down syndrome, you'll likely rely on a team of specialists that, depending on your child's particular needs, will provide your child's medical care and help him or her develop skills as fully as possible. Your team may include some of these experts:

- Primary care pediatrician to coordinate and provide routine childhood care

- Pediatric cardiologist
- Pediatric gastroenterologist
- Pediatric endocrinologist
- Developmental pediatrician
- Pediatric neurologist
- Pediatric ear, nose and throat (ENT) specialist
- Pediatric eye doctor (ophthalmologist)
- Audiologist
- Physical therapist
- Speech pathologist
- Occupational therapist

When you learn your child has Down syndrome, you may experience a range of emotions, including anger, fear, worry, sorrow and guilt. You may not know what to expect, and you may worry about your ability to care for a baby with a disability. The best antidote for fear and worry is information and support.

Consider these steps to prepare yourself and to care for your child:

- **Find a team of trusted professionals.** You'll need to make important decisions about your child's education and treatment. Build a team of health care providers, teachers and therapists you trust. These professionals can help evaluate the resources in your area and explain state and federal programs for children with disabilities.
- **Seek out other families who are dealing with the same issues.** Most communities have support groups for parents of children with Down syndrome. You can also find Internet support groups. Family and friends can also be a source of understanding and support.
- **Expect a bright future.** Most people with Down syndrome live with their families or independently, go to mainstream schools, read and write, and have jobs. People with Down syndrome can live fulfilling lives.

There's no way to prevent Down syndrome. If you're at high risk of having a child with Down syndrome or you already have one child with Down syndrome, you may want to consult a genetic counselor before becoming pregnant.

A genetic counselor can help you understand your chances of having a child with Down syndrome. He or she can also explain the prenatal tests that are available and help explain the pros and cons of testing.

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