

# Down Syndrome (Trisomy 21)

Down syndrome is a relatively common chromosomal disorder. Most children with Down syndrome have mild to moderate mental impairment. About half have heart defects or other medical problems such as hearing and vision problems. Prenatal testing may be able to recognize Down syndrome before the baby is born. Your doctor can help to arrange for appropriate medical care for your child with Down syndrome, along with services and support for your family.

## What is Down syndrome?

Down syndrome is a genetic disease caused by a chromosomal abnormality. The child usually has a certain appearance that is common to children with Down syndrome. These children also have differing degrees of mental deficiency, heart disease, and other problems.

Down syndrome usually results from an extra copy of chromosome 21, although other chromosomal abnormalities are possible. It occurs in about 1 of every 600 to 800 live-born babies.

Your child will have screening tests to look for medical problems associated with Down syndrome. Visits to some medical specialists may be recommended as well. We can help you access appropriate medical care for your child as well as support for your family, including early intervention and special education services.

## What kinds of problems occur with Down syndrome?

*Physical appearance.* Children with Down syndrome have a certain typical appearance:

- Flat, broad head.
- Slanted eyes, pointing upward.
- Small mouth; tongue tends to stick out.
- Short, broad hands with a single crease across the palm. The little finger is short and curves inward. There is a wide space between the first and second toes.
- Poor muscle tone during the newborn period (“floppy”).
- As they grow, children with Down syndrome have shorter than average height.

*Medical problems.* Children with Down syndrome are at increased risk of many different medical problems:

- Heart problems. About half are born with heart defects, most commonly atrial septal defect (ASD). In this con-

dition, a hole is present between two chambers of the heart. As children get older, problems with the heart valves may develop.

- Mental impairment. Most children with Down syndrome have mild to moderate mental retardation. Some have severe mental impairment.
- Hearing loss and ear infections are frequent.
- Vision problems, including cataracts and strabismus.
- Snoring and difficulty breathing during sleep (obstructive sleep apnea [OSA]) occur in about half of children.
- Thyroid problems. Usually low thyroid function (hypothyroidism), but sometimes high thyroid function (hyperthyroidism).
- Infections are more frequent than in the average child.
- Gastrointestinal problems, especially obstruction (blockage) of the small intestine during the newborn period. There is also an increased risk of celiac disease: sensitivity to gluten, a substance present in cereals and breads.
- Obesity.
- Leukemia. Risk is higher than average, although only about 1% of Down syndrome children are affected.
- Instability of the cervical spine (neck). This can affect the spinal nerves, causing weakness and other neurologic symptoms.
- Females with Down syndrome are fertile and able to bear children. Almost all males are infertile.
- People with Down syndrome have a shortened lifespan. They are also at increased risk of early Alzheimer’s disease.

## What causes Down syndrome?

In over 90% of children with Down syndrome, the disease results from the presence of an extra chromosome. Normally, people have 46 chromosomes—23 from each parent. Most children with Down syndrome have 47 chromosomes, with an extra copy of chromosome 21; that’s why this condition is also called “trisomy 21.” A small number of children have the right number of chromosomes, but an extra piece of chromosome 21 is attached to one of the other chromosomes. This is called a *translocation*, and affected children have the same problems as those with typical Down syndrome.

In 1% to 2% of children with Down syndrome, some cells contain the normal 46 chromosomes, while others contain 47 chromosomes. This is called *mosaicism*. These children have fewer problems than those with other chromosomal abnormalities.

## What increases the risk of Down syndrome?

- Older age of the mother. The rate of Down syndrome pregnancies is increased for mothers over 35.
- Having another family member with Down syndrome.

## How is Down syndrome diagnosed?

- After the baby is born, a simple chromosome test can confirm whether or not Down syndrome is present.
- Women over 35 and other pregnant women at high risk of having a Down syndrome child can undergo prenatal (before birth) testing. This may include procedures to obtain samples of the amniotic fluid in the womb (called amniocentesis) or the placenta (called chorionic villus sampling).
- New strategies to detect Down syndrome before birth are now being used. A combination of blood tests and ultrasound scans is performed during the first and second trimesters of pregnancy. The results help to determine if amniocentesis or other additional tests are needed. If not, the mother can avoid the risks of these tests.
- If Down syndrome is diagnosed before birth, the parents face the difficult decision of whether to terminate the pregnancy.

## How is Down syndrome managed?

*Medical issues.* During regular checkups, your child will be screened for various health problems that may occur:

- Thyroid function is checked as part of regular newborn screening tests and rechecked frequently.
- A test called an echocardiogram (“echo”), which uses sound waves to take pictures of your child’s heart, will be done to check for heart defects. If any are found, your doctor will recommend a visit to a heart specialist (cardiologist). Some heart defects require surgery.
- By age 6 months, your child should be checked by an eye specialist (ophthalmologist) for eye problems.
- Hearing is checked at birth and rechecked at certain times. We may recommend visits to an ear, nose, and

throat specialist (ENT or otorhinolaryngologist) and an audiologist (a professional trained to evaluate hearing problems).

- The ENT specialist can also evaluate and treat snoring and sleep-related breathing problems.
- Screening for celiac disease (gluten intolerance) may be recommended at around age 2 or 3.

*Developmental issues.* Education is an important aspect of care for Down syndrome:

- *Early intervention* should start as soon as your child’s condition is diagnosed. Every state has an early intervention program; our office can put you in touch with resources to get you started. Early intervention experts can assess your child and develop an Individualized Family Support Plan (IFSP) based on your child’s development, need for support, and goals for independence.
- *Special education* services are also available in every state (although children with less severe impairment may not qualify for services). You are entitled to expert evaluation of your child. Based on the results, an Individualized Education Program (IEP) can be developed to meet your child’s educational needs.
- Speech therapy can be helpful.
- Having a child with Down syndrome is a life-changing event for your family. We can help to put you in touch with community resources to help children with Down syndrome and their families.

## When should I call your office?

Your child will receive regular checkups to look for medical problems associated with Down syndrome. Call our office if problems come up between visits or if you have any questions about your child’s testing, treatment, or educational intervention.

## Where can I get more information about Down syndrome?

A good place to start is the National Down Syndrome Society. On the Internet at [www.ndss.org](http://www.ndss.org), or call 1-800-221-4602.