

# **Birth Defects in Infants**

*Information is from the Center of Disease Control (CDC)*

<http://www.cdc.gov/ncbddd/birthdefects/index.html>



## Downs Syndrome

Down syndrome is a condition in which a baby is born with an extra chromosome. Chromosomes are small “packages” of genes in the body. They determine how a baby’s body forms during pregnancy and how, as the baby grows in the womb and after birth, the baby’s body functions. Normally, a baby is born with 46 chromosomes. Babies born with Down syndrome have an extra copy of one of these chromosomes. This extra copy changes the body’s and brain’s normal development and causes mental and physical problems for the baby.

Even though people with Down syndrome might have some physical and mental features in common, symptoms of Down syndrome can range from mild to severe. Usually, mental development and physical development are slower in people with Down syndrome than in those without it.

Some common physical signs of Down syndrome include:

- A flat face with an upward slant to the eye, a short neck, small ears, and a large tongue
- Tiny white spots on the iris (colored part) of the eye
- Small hands and feet
- A single crease across the palm of the hand
- Small pinky fingers that sometimes curve toward the thumb
- Poor muscle tone or loose ligaments

### **What problems do children with Down syndrome have?**

Babies and adults with Down syndrome can have physical problems, as well as intellectual disabilities. Every baby born with Down syndrome is different. In addition to the physical signs, some might have major birth defects or other medical problems. However, many people with Down syndrome live happy, productive lives well into adulthood.

Still, some physical problems associated with Down syndrome include:

- A birth defect of the heart
- Stomach problems, such as a blocked small intestine
- Celiac disease, a digestive disease that damages the small intestine so that nutrients from food are not absorbed well
- Problems with memory, concentration, and judgment, often called dementia
- Hearing problems
- Eye problems, such as cataracts or trouble seeing objects that are close by (far-sighted)
- Thyroid problems
- Skeletal problems

A person with Down syndrome can have an IQ in the mild-to-moderate range of intellectual disabilities. He or she also might have delayed language development and difficulties with physical coordination.

### **What causes Down Syndrome?**

To understand Down syndrome, it is necessary to understand how a baby develops. Each baby starts developing when he or she receives 23 chromosomes from the mother’s egg and 23 chromosomes from the father’s sperm. When a baby has Down syndrome, an error happened when either the egg or the sperm was formed. This error caused an extra chromosome (called chromosome number 21) in the egg or sperm, so that the baby received a total of 24 instead of 23 chromosomes from one of its parents. Therefore, the baby ends up having 47 chromosomes in every cell of his or her body, instead of 46 chromosomes. This extra chromosome causes the physical signs and additional problems that can occur among people with Down syndrome. The causes of the errors that produces the extra chromosome is not known.

The age of the mother is the only factor that has been shown to increase the risk of having a baby with Down syndrome. This risk increases with every year, especially after the mother is 35 years of age. However, because younger women are more likely to have babies than older women, 80% of babies with Down syndrome are born to women younger than 35 years of age.

CDC works with many researchers to study the risk factors that can increase the chance of having a baby with Down syndrome. Following are examples of what this research has found:

- The number of babies with Down syndrome seems to be increasing, especially among mothers older than 35 years of age.<sup>2</sup>
- Certain factors seem to influence how long a person with Down syndrome will live, including ethnicity, low weight at birth, and whether the baby was born with a heart defect.<sup>3</sup>
- Death rates among Black or African-American infants with Down syndrome seem to be higher than death rates among White infants with Down syndrome.<sup>4</sup>

### **Can Down Syndrome be prevented?**

There is no known way to prevent the Down syndrome. However, infants and children with Down syndrome often will benefit from special programs that help to improve their physical and mental limitations. These include speech therapy, occupational therapy, and exercises for physical coordination. Children with Down syndrome usually also need extra help or attention in school.

While there is currently no way to prevent Down syndrome, mothers can take steps before and during pregnancy to have a healthy pregnancy. Steps include taking a daily multivitamin with folic acid (400 micrograms), not smoking, and not drinking alcohol during pregnancy.

## Spina Bifida

Spina bifida is a condition that affects the spine and is usually apparent at birth. It is a type of neural tube defect (NTD).

Spina bifida can happen anywhere along the spine if the neural tube does not close all the way. The backbone that protects the spinal cord does not form and close as it should. This often results in damage to the spinal cord and nerves.

Spina bifida might cause physical and intellectual disabilities that range from mild to severe. The severity depends on:

- The size and location of the opening in the spine.
- Whether part of the spinal cord and nerves are affected.

### **During Pregnancy**

During pregnancy there are screening tests (prenatal tests) to check for spina bifida and other birth defects. Talk with your doctor about any questions or concerns you have about this prenatal testing.

- **AFP** - AFP stands for alpha-fetoprotein (sounds like: al-fa-fee-toe-pro-teen), a protein the unborn baby produces. This is a simple blood test that measures how much AFP has passed into the mother's bloodstream from the baby. A high level of AFP might mean that the baby has spina bifida. An AFP test might be part of a test called the "triple screen" that looks for neural tube defects and other issues.
- **Ultrasound** - An ultrasound is a type of picture of the baby. In some cases, the doctor can see if the baby has spina bifida or find other reasons that there might be a high level of AFP. Frequently, spina bifida can be seen with this test.
- **Amniocentesis** (sounds like: am-knee-oh-sin-te-sus; [hear how "amniocentesis" sounds](#)) - For this test, the doctor takes a small sample of the amniotic fluid surrounding the baby in the womb. Higher than average levels of AFP in the fluid might mean that the baby has spina bifida.

### **After the Baby Is Born**

In some cases, spina bifida might not be diagnosed until after the baby is born.

Sometimes there is a hairy patch of skin or dimple on the baby's back that is first seen after the baby is born. A doctor can use an image scan, such as an X-ray, MRI, or CT, to get a clearer view of the baby's spine and the bones in the back.

Sometimes spina bifida is not diagnosed until after the baby is born because the mother did not receive prenatal care or an ultrasound did not show clear pictures of the affected part of the spine.

### **Treatments**

Not all people born with spina bifida have the same needs, so treatment will be different for each person. Some people have problems that are more serious than others.

### **Causes and Prevention**

We do not know all of the causes of spina bifida. The role that factors, such as genes and the environment, play in causing spina bifida need to be studied further. However, we do know that there are ways before and during pregnancy for women to reduce the risk of having a baby with spina bifida.

If you are pregnant or could get pregnant, use the following tips to help prevent your baby from having spina bifida:

- Take 400 micrograms (mcg) of [folic acid](#) every day. If you already have had a pregnancy affected by spina bifida, talk with your doctor about a prescription to take 4,000 mcg (4.0 milligrams). Folic acid prevents most, but not all, cases of spina bifida.
- Talk to your doctor or pharmacist about any prescription and over-the-counter drugs, vitamins, and dietary or herbal supplements you are taking. [Learn about medication and pregnancy »](#)
- If you have a medical condition—such as [diabetes](#) or [obesity](#)—be sure it is under control before you become pregnant.
- Avoid overheating your body, as might happen if you use a hot tub or sauna.
- Treat any fever you have right away with Tylenol® (or store brand).

### **Remember!**

Spina bifida happens in the first few weeks of pregnancy, often before a woman knows she's pregnant. Although folic acid is not a guarantee that a woman will have a healthy pregnancy, taking folic acid can help reduce a woman's risk of having a pregnancy affected by spina bifida. Because half of all pregnancies in the United States are unplanned, it is important that all women who can become pregnant take folic acid before and during pregnancy.

### **Living with Spina Bifida**

Spina bifida can range from mild to severe. Some people have little or no noticeable disability. Others are limited in the way they can move or function. They even might be paralyzed (unable to walk or move parts of the body). Even so, with the right care, most people affected by spina bifida will be able to grow up to lead full and productive lives.

## **Anencephaly**

Anencephaly is a serious birth defect in which a baby is born without parts of the brain and skull. It is a type of neural tube defect (NTD). These are birth defects that happen during the first month of pregnancy, usually before a woman knows she is pregnant. As the neural tube forms and closes, it helps form the baby's brain and skull (upper part of the neural tube), spinal cord, and back bones (lower part of the neural tube).

Anencephaly happens if the upper part of the neural tube does not close all the way. This often results in a baby being born without the front part of the brain (forebrain) and the thinking and coordinating part of the brain (cerebrum). The remaining parts of the brain are often not covered by bone or skin. Almost all babies born with anencephaly will die shortly after birth. CDC estimates that each year, about 1 in every 4,859 babies in the United States will be born with anencephaly

### **Diagnosis**

Anencephaly can be diagnosed during pregnancy or after the baby is born.

### **During Pregnancy**

During pregnancy, there are [screening tests \(prenatal tests\)](#) to check for birth defects and other conditions. Anencephaly would result in an abnormal result on a blood or serum screening test or it might be seen during an ultrasound (which creates pictures of the body).

### **After the Baby is Born**

In some cases, anencephaly might not be diagnosed until after the baby is born. Anencephaly is immediately seen at birth.

### **Treatments**

There is no known cure or standard treatment for anencephaly. Almost all babies born with anencephaly will die shortly after birth.

## Ventricular Septal Defect

A ventricular septal defect (VSD) is a birth defect of the heart in which there is a hole in the wall (*septum*) that separates the two lower chambers (*ventricles*) of the heart. This wall also is called the *ventricular septum*. A ventricular septal defect happens during pregnancy if the wall that forms between the two ventricles does not fully develop, leaving a hole. A ventricular septal defect is one type of congenital heart defect. *Congenital* means present at birth.

### **Causes and Risk Factors**

The causes of heart defects (such as a ventricular septal defect) among most babies are unknown. Some babies have heart defects because of changes in their [genes](#) or [chromosomes](#). Heart defects also are thought to be caused by a combination of genes and other risk factors, such as the things the mother comes in contact with in the environment or what the mother eats or drinks or the medicines the mother uses.

### **Diagnosis**

A ventricular septal defect usually is diagnosed after a baby is born.

The size of the ventricular septal defect will influence what symptoms, if any, are present, and whether a doctor hears a heart murmur during a physical examination. Signs of a ventricular septal defect might be present at birth or might not appear until well after birth. If the hole is small, it usually will close on its own and the baby might not show any signs of the defect. However, if the hole is large, the baby might have symptoms, including:

- Shortness of breath,
- Fast or heavy breathing,
- Sweating,
- Tiredness while feeding, or
- Poor weight gain.

During a physical examination the doctor might hear a distinct whooshing sound, called a heart murmur. If the doctor hears a [heart murmur](#) or other signs are present, the doctor can request one or more tests to confirm the diagnosis. The most common test is an *echocardiogram*, which is an ultrasound of the heart that can show problems with the structure of the heart, show how large the hole is, and show how much blood is flowing through the hole.

### **Treatments**

Treatments for symptoms associated with ventricular septal defect can include:

**-Medicines:** Some children will need medicines to help strengthen the heart muscle, lower their blood pressure, and help the body get rid of extra fluid.

**-Nutrition:** Some babies with a ventricular septal defect become tired while feeding and do not eat enough to gain weight. To make sure babies have a healthy weight gain, a special high-calorie formula might be prescribed. Some babies become extremely tired while feeding and might need to be fed through a [feeding tube](#).

## **Cleft Lip**

Cleft lip and cleft palate are birth defects that occur when a baby's lip or mouth do not form properly. Together, these birth defects commonly are called "orofacial clefts". These birth defects happen early during pregnancy. A baby can have a cleft lip, a cleft palate, or both.

Children with a cleft lip with or without a cleft palate or a cleft palate alone often have problems with feeding and talking. They also might have ear infections, hearing loss, and problems with their teeth.

The Centers for Disease Control and Prevention (CDC) recently estimated that each year 2,651 babies in the United States are born with a cleft palate and 4,437 babies are born with a cleft lip with or without a cleft palate.<sup>1</sup> Cleft lip is more common than cleft palate. Isolated orofacial clefts, or clefts that occur with no other birth defects, are one of the most common birth defects in the United States. About 70% of all orofacial clefts are isolated clefts.

### **Cleft Lip**

The lip forms between the fourth and seventh weeks of pregnancy. A cleft lip happens if the tissue that makes up the lip does not join completely before birth. This results in an opening in the upper lip. The opening in the lip can be a small slit or it can be a large opening that goes through the lip into the nose. A cleft lip can be on one or both sides of the lip or in the middle of the lip, which occurs very rarely. Children with a cleft lip also can have a cleft palate.

### **Cleft Palate**

The roof of the mouth is called the "palate." It is formed between the sixth and ninth weeks of pregnancy. A cleft palate happens if the tissue that makes up the roof of the mouth does not join correctly. Among some babies, both the front and back parts of the palate are open. Among other babies, only part of the palate is open.

### **Causes and Risk Factors**

Just like the many families affected by birth defects, CDC wants to find out what causes them. Understanding the risk factors that can increase the chance of having a baby with a birth defect will help us learn more about the causes. CDC currently is working on one of the largest studies in the United States—the [National Birth Defects Prevention Study](#)<sup>2</sup>—to understand the causes of and risk factors for birth defects. This study is looking at many possible risk factors for birth defects, such as orofacial clefts.

The causes of orofacial clefts among most infants are unknown. Some children have a cleft lip or cleft palate because of changes in their [genes](#). Cleft lip and cleft palate are thought to be caused by a combination of genes and other factors, such as exposures in the environment, maternal diet, and medication use.

Recently, CDC reported on important findings about some factors that increase the risk of orofacial clefts:

- Smoking—Women who smoke during pregnancy are more likely to have a baby with an orofacial cleft than women who do not smoke.<sup>2,3</sup>
- Diabetes—Women with diabetes diagnosed before pregnancy have been shown to be an increased risk of having a child with a cleft lip with or without cleft palate.<sup>4</sup>

CDC continues to study birth defects, such as orofacial clefts and how to prevent them. If you smoke or have diabetes, and you are pregnant or thinking about becoming pregnant, talk with your doctor about ways to increase your chances of having a healthy baby.

### **Diagnosis**

Orofacial clefts sometimes can be diagnosed during pregnancy, usually by a routine ultrasound. Most often, orofacial clefts are diagnosed after the baby is born. However, sometimes minor clefts (e.g., submucous cleft palate and bifid uvula) might not be diagnosed until later in life.

### **Treatments**

Services and treatment for children with orofacial clefts can vary depending on the severity of the cleft; the presence of associated syndromes or other birth defects, or both; and the child's age and needs. Surgery to repair a cleft lip usually occurs in the first few months of life and is recommended within the first 12 months of life. Surgery to repair a cleft palate is recommended within the first 18 months of life.<sup>5</sup> Many children will need additional surgeries as they get older. Although surgical repair can improve the look and appearance of a child's face, it also may improve breathing, hearing, speech, and language. Children born with orofacial clefts also might need different types of treatments and services, such as special dental or orthodontic care or speech therapy.<sup>6</sup>