

# Down Syndrome

## What is it?

**Down syndrome**, also called **trisomy 21**, is a condition caused by an extra copy of chromosome 21. Normally, we have 23 pairs of chromosomes, for a total of 46. We receive one copy of each chromosome from our mothers and one copy of each chromosome from our fathers. People with Down syndrome have an extra copy of chromosome 21, for a total of three copies. **Trisomy** means three copies of a chromosome. The extra copy causes both mental and physical challenges.

Most people with Down syndrome have similar facial features, including a flattened face, almond-shaped eyes that slant up, a short neck, small ears, and tiny white spots on the colored part of the eye. People with Down syndrome also may have small hands and feet, small pinky fingers that curve in, poor muscle tone, loose joints, and short stature.

Some people with Down syndrome have other major conditions such as hearing loss, ear infections, eye diseases, and congenital heart defects. Your baby's doctor should routinely check for any other conditions.

## How common is it?

Down syndrome is the most common chromosomal condition in the United States. About 1 out of every 700 babies born each year has Down syndrome. That's about 6,000 babies each year in the United States.

## What causes it?

The extra chromosome is present due to a random mistake that occurs before pregnancy, during the development of egg or sperm cells. This mistake results in an extra copy of chromosome 21 being present in every cell of the baby's body. It is important for families to know that it is not possible to prevent the extra chromosome from being present. There is also nothing you did to cause the extra chromosome to be present. Pregnant women over the age of 35 years have an increased chance of having a baby with Down syndrome.

## How is it diagnosed?

Screening tests can be performed during pregnancy, such as a blood test and an ultrasound. After a screening test is performed, a diagnostic test is done. There are typically three types of diagnostic tests for Down syndrome that check for the extra chromosome. Your doctor should discuss these options with you.

## How is it treated?

There is no cure for Down syndrome; however, many people with Down syndrome live into adulthood and lead productive lives. Speech, language, and physical therapy services early in life will help a baby with Down syndrome reach his or her full potential.



### For more information:

**National Down Syndrome Society**

<https://www.ndss.org/about-down-syndrome/down-syndrome/>

**Centers for Disease Control and Prevention**

<https://www.cdc.gov/ncbddd/birthdefects/downsyndrome.html>