

DOWN SYNDROME



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What is Down syndrome?

Down syndrome is a genetic condition where people are born with an extra chromosome. Most people have 23 pairs of chromosomes within each cell in their body, for a total of 46. A person diagnosed with Down syndrome has an extra copy of chromosome 21, which means their cells contain 47 total chromosomes instead of 46. This changes the way their brain and body develop.



Who does Down syndrome affect?

Down syndrome can affect anyone. It's a genetic condition, and it doesn't happen as a result of something that the parents did before or during pregnancy. The majority of Down syndrome cases happen randomly (sporadically). People don't usually inherit Down syndrome in an autosomal dominant or recessive pattern during conception when the egg and sperm meet.

What are the risk factors for Down syndrome?

Research is ongoing to learn more about the risk factors of Down syndrome. Studies suggest that the risk of giving birth to a child with Down syndrome increases as the birthing parent ages. Women or people assigned female at birth (AFAB) who are 35 or older are more likely to have a child diagnosed with Down syndrome or another type of genetic condition. Since people AFAB younger than 35 have higher fertility rates, most babies with Down syndrome are born to people under 35.

Symptoms and Causes

What are the symptoms of Down syndrome?

Down syndrome causes physical, cognitive and behavioral symptoms. Not all people with Down syndrome have all of these symptoms. Symptoms and their severity are different from person to person.

Physical signs of Down syndrome



Physical signs of Down syndrome are usually present at birth and become more apparent as your baby grows. They can include:

- A flat nose bridge
- Slanted eyes that point upward
- A short neck
- Small ears, hands and feet
- Weak muscle tone at birth
- Small pinky fingers those points inward towards the thumb
- One crease in the palm of their hand (palmar crease)
- Shorter-than-average height

As child grows, additional symptoms can arise because of the way that their body developed in the uterus, including

- Ear infections or hearing loss
- Vision problems or eye diseases
- Dental problems
- Being more prone to infections or illnesses
- Obstructive sleep apnea
- Congenital heart disease

Cognitive symptoms of Down syndrome

Children with Down syndrome may have cognitive development challenges as a result of their extra chromosome. This can cause intellectual or developmental disabilities. Your child's ability to meet developmental milestones, or things that your child can do at a certain age, may differ from other children, including how they

- Walk and move (gross and fine motor skills)
- Speak (language development skills)
- Learn (cognitive skills)
- Play (social and emotional skills)

As a result, it may take your child longer to do the following things

- Toilet training
- Speaking their first words
- Taking their first steps
- Eating food independently

Behavioral symptoms of Down syndrome

Child diagnosed with Down syndrome may exhibit behavioral symptoms. This can be the result of your child not being able to communicate their needs to you or their caregivers effectively.

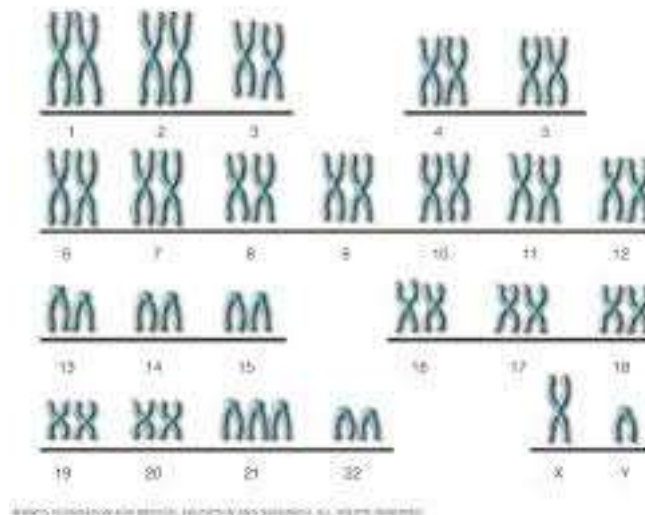
- Stubbornness and tantrums
- Difficulty paying attention
- Obsessive or compulsive behaviors

What causes Down syndrome?

An extra chromosome causes Down syndrome. Each human cell typically contains 23 pairs of chromosomes, which totals 46. Down syndrome occurs because of changes in the way cells in chromosome 21 divide. Every person with Down syndrome has an extra chromosome 21 in some or all of their cells

There are three types of Down syndrome with different causes, including

- Trisomy 21
- Translocation
- Mosaicism



What is trisomy 21?

Trisomy 21 is the most common type of Down syndrome. The term “trisomy” means having an extra copy of a chromosome. Trisomy 21 occurs when a developing fetus has three copies of chromosome 21 in every cell instead of the typical two copies. This type makes up 95% of all cases of Down syndrome.

What is translocation Down syndrome?

Translocation is a type of Down syndrome where there's a partial or full amount of chromosome 21 attached to another chromosome. Unlike trisomy 21, translocation occurs when chromosome 21 isn't separate, but it relocates to another numbered chromosome. This type of Down syndrome accounts for less than 4% of all cases.

What is mosaic Down syndrome?

Mosaic Down syndrome is the rarest type of Down syndrome and accounts for less than 1% of all cases. Mosaicism occurs when only some cells contain the usual 46 chromosomes and some cells contain 47. The extra chromosome in some cells is chromosome 21.

Diagnosis and Tests

How is Down syndrome diagnosed before birth?

A healthcare provider can suspect Down syndrome during pregnancy with prenatal screening tests. They can also diagnose this condition during pregnancy with diagnostic tests.

Prenatal screening tests

These tests assess your risk of having a child with Down syndrome rather than giving you a confirmation of a diagnosis. Screening tests could be a blood test of the birthing parent's blood to look for indicators of Down syndrome. Another screening test is an ultrasound. During this imaging test, your provider will look for signs of Down syndrome, like extra fluid behind your baby's neck. It's possible that a screening test could be normal and not show signs of Down syndrome when the condition is present.

Diagnostic tests during pregnancy

Diagnostic tests during pregnancy can confirm a Down syndrome diagnosis. They usually take place after a positive screening test.

Diagnostic testing offers more risk for the birthing parent and the developing fetus. Tests could include:

- Amniocentesis
- Chorionic villus sampling (CVS)
- Percutaneous umbilical blood sampling (PUBS)

These tests can detect chromosomal changes that lead to a Down syndrome diagnosis.

How is Down syndrome diagnosed after birth?

After your baby is born, providers look for the physical signs of Down syndrome during a physical exam. To confirm the diagnosis, your baby's provider may perform a blood test called a karyotype test. In this test, your baby's provider will remove a small blood sample to study under a microscope. They'll look for an extra 21st chromosome to diagnose the condition.

Management and Treatment

How is Down syndrome treated?

There's no cure for Down syndrome, but treatment is available to help your child reach their full potential. Treatment focuses on helping your child thrive physically and mentally. Treatment options could include

- Physical or occupational therapy
- Speech therapy
- Participating in special education programs in school
- Treating any underlying medical conditions
- Wearing glasses for vision problems or assisted hearing devices for hearing loss

Who is on my child's care team?

If a child has Down syndrome, they'll likely see a variety of specialists to make sure they're healthy. Their care team may include

- Primary care providers to monitor growth, development and medical concerns and provide vaccinations
- Medical specialists, depending on the needs of the person (for example, cardiologist, endocrinologist)
- Speech therapists to help them communicate
- Physical therapists to help strengthen their muscles and improve motor skills
- Occupational therapists to help refine their motor skills and make daily tasks easier
- Behavioral therapists to help manage emotional challenges that can come with Down syndrome

What other medical conditions are associated with Down syndrome?

People with Down syndrome may have other medical conditions that are either present at birth or develop over time. A healthcare provider can help you and your child manage these conditions. Common conditions of Down syndrome can include

- Heart problems
- Thyroid abnormalities
- Gastrointestinal problems like constipation, gastroesophageal reflux and celiac disease
- Autism, challenges with social skills, communication and repetitive behaviors
- Alzheimer's disease

Prevention

How can I prevent Down syndrome?

You can't prevent Down syndrome since it's a genetic condition. To learn more about your risk of having a child with a genetic condition, talk to your healthcare provider about genetic testing.

What can I expect if I have a child diagnosed with Down syndrome?

Children born with Down syndrome lead happy and healthy lives with supportive care. Treatment and therapies can help them reach developmental milestones, gain an education with their peers, build relationships and have successful careers.

Groups and resources are available for parents, family members and caregivers to help you best care for your child and support them.

Is there a cure for Down syndrome?

No, Down syndrome is a lifelong condition, and there isn't a cure. Symptoms of the condition are manageable, and treatment is available for any associated conditions that may arise.