



Down Syndrome

What is Down syndrome?

Down syndrome is a genetic disorder caused when abnormal cell division results in an extra full or partial copy of chromosome 21. This extra genetic material causes the developmental changes and physical features of Down syndrome.

Down syndrome varies in severity among individuals, causing lifelong intellectual disability and developmental delays. It's the most common genetic chromosomal disorder and cause of learning disabilities in children. It also commonly causes other medical abnormalities, including heart and gastrointestinal disorders.

What are the symptoms of Down syndrome?

Each person with Down syndrome is an individual — intellectual and developmental problems may be mild, moderate or severe. Some people are healthy while others have significant health problems such as serious heart defects.

Children and adults with Down syndrome have distinct facial features. Though not all people with Down syndrome have the same features, some of the more common features include:

- Flattened face
- Small head
- Short neck
- Protruding tongue
- Upward slanting eye lids (palpebral fissures)
- 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's 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.

How is Down syndrome diagnosed?

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 or chances that a mother is carrying a baby with Down syndrome. But these tests can't tell for sure or diagnose whether the baby has Down syndrome.
- Diagnostic tests can identify or diagnose 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.

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 to confirm diagnosis. Using a sample of blood, this test analyzes your child's chromosomes. If there's an extra chromosome 21 in all or some cells, the diagnosis is Down syndrome.

Treatment options for Down syndrome

Early intervention for infants and children with Down syndrome can make a major difference in improving their quality of life. Because each child with Down syndrome is unique, treatment will depend on individual needs. Also, different stages of life may require different services.

If your child has Down syndrome, you'll likely rely on a team of specialists that can provide medical care and help him or her develop skills as fully as possible. Depending on your child's particular needs, 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
- Speech pathologist
- Physical therapist
- Occupational therapist

You'll need to make important decisions about your child's treatment and education. 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 and adults with disabilities.

Long-term complications of Down syndrome

Life spans have increased dramatically for people with Down syndrome. Today, someone with Down syndrome can expect to live more than 60 years, depending on the severity of health problems.

Resources

- **National Down Syndrome Society** - www.ndss.org/Down-Syndrome/What-Is-Down-Syndrome
- **Mayo Foundation for Medical Education and Research** - www.mayoclinic.org/diseases-conditions/down-syndrome/home/ovc-20337339