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Down Syndrome

Also called: Trisomy 21

What is Down syndrome?

Down syndrome is a condition in which a person has an extra chromosome or an extra piece of a chromosome. This extra copy changes how a baby's body and brain develop. It can cause both mental and physical challenges during their lifetime. Even though people with Down syndrome might act and look similar, each person has different abilities.

What causes Down syndrome?

Chromosomes [<https://medlineplus.gov/genetics/understanding/basics/chromosome/>] are tiny "packages" in your cells that contain your genes. Genes carry information, called DNA, that controls what you look like and how your body works. People with Down syndrome have an extra copy of chromosome 21. In some cases, they may have an extra copy of part of the chromosome. Having an extra copy of a chromosome is called trisomy. So sometimes Down syndrome is also called trisomy 21.

Down syndrome is usually not inherited. It happens by chance, as an error when cells are dividing during early development of the fetus [<https://medlineplus.gov/fetalhealthanddevelopment.html>] . It is not known for sure why Down syndrome occurs or how many different factors play a role.

One factor that increases the risk of having a baby with Down syndrome is the age of the mother. Women ages 35 and older are more likely to have a baby with Down syndrome.

What are the symptoms of Down syndrome?

The symptoms of Down syndrome are different in each person. And people with Down syndrome may have different problems at different times of their lives. They usually have mild to moderate intellectual disabilities. Their development is often delayed [<https://medlineplus.gov/developmentaldisabilities.html>] . For example, they may start talking later [<https://medlineplus.gov/speechandlanguageproblemsinchildren.html>] than other children.

Some of the common physical signs of Down syndrome include:

- A flat face
- Eyes that slant up
- A short neck
- Small hands and feet
- Poor muscle tone
- Loose joints

What other problems does Down syndrome cause?

Many people with Down syndrome have the common physical signs and have healthy lives. But some people with Down syndrome might have one or more birth defects or other health problems. Some of the more common ones include:

- Hearing loss [<https://medlineplus.gov/hearingproblemsinchildren.html>]

- Sleep apnea [<https://medlineplus.gov/sleepapnea.html>] (a disorder that causes you to repeatedly stop breathing during sleep)
- Ear infections [<https://medlineplus.gov/earinfections.html>]
- Eye diseases [<https://medlineplus.gov/eyediseases.html>]
- Congenital heart defects [<https://medlineplus.gov/congenitalheartdefects.html>] (heart defects that are present at birth)
- Digestive problems [<https://medlineplus.gov/digestivediseases.html>]
- Problems with the upper part of the spine [<https://medlineplus.gov/spineinjuriesanddisorders.html>]
- Obesity [<https://medlineplus.gov/obesity.html>]

How is Down syndrome diagnosed?

Health care providers can check for Down syndrome during pregnancy or after a child is born.

There are two basic types of tests that help find Down syndrome during pregnancy [<https://medlineplus.gov/lab-tests/down-syndrome-tests/>] :

- **Prenatal screening tests** can show whether your unborn baby has a higher or lower chance of having Down syndrome. If a screening test shows that your baby could have Down syndrome, you'll need another test to find out for sure.
- **Prenatal diagnostic tests** can diagnose or rule out Down syndrome by checking the chromosomes in a sample of cells.

These tests have a small risk of causing a miscarriage, so they're often done after a screening test shows that an unborn baby could have Down syndrome.

After a baby is born, the provider may make an initial diagnosis of Down syndrome based on the physical signs of the syndrome. The provider can use a karyotype genetic test [<https://medlineplus.gov/lab-tests/karyotype-genetic-test/>] to confirm the diagnosis. The test can check for extra chromosomes in a sample of the baby's blood.

What are the treatments for Down syndrome?

There is no single, standard treatment for Down syndrome. Treatments are based on each person's physical and intellectual needs, strengths, and limitations.

Services early in life focus on helping children with Down syndrome develop to their full potential. These services include speech, occupational, and physical therapies. They are typically offered through early intervention programs in each state. Children with Down syndrome may also need extra help or attention in school, although many children are included in regular classes.

Since people with Down syndrome can have birth defects and other health problems, they will need regular medical care. They may need to have certain extra health screenings [<https://medlineplus.gov/healthscreening.html>] to check for problems that happen more often in people with Down syndrome.

Centers for Disease Control and Prevention

Start Here

- About Down Syndrome [<https://ndss.org/about>] (National Down Syndrome Society)
- About Down Syndrome [<https://www.nichd.nih.gov/health/topics/down/conditioninfo>]
 (Eunice Kennedy Shriver National Institute of Child Health and Human Development)
- Down Syndrome (For Parents) [<https://kidshealth.org/en/parents/down-syndrome.html>] (Nemours Foundation)
 Also in Spanish [<https://kidshealth.org/es/parents/down-syndrome.html>]
- Down Syndrome in Adults: Staying Healthy [<https://familydoctor.org/down-syndrome-in-adults-staying-healthy/?adfree=true>]
 (American Academy of Family Physicians)
 Also in Spanish [<https://es.familydoctor.org/sindrome-de-down-el-sindrome-de-down-en-adultos-como-mantenerse-saludable/?adfree=true>]

- Facts about Down Syndrome [<https://www.cdc.gov/ncbddd/birthdefects/DownSyndrome.html>] (Centers for Disease Control and Prevention)

Symptoms

- What Are the Symptoms of Down Syndrome? [<https://www.nichd.nih.gov/health/topics/down/conditioninfo/symptoms>]  (Eunice Kennedy Shriver National Institute of Child Health and Human Development)
Also in Spanish [<https://espanol.nichd.nih.gov/salud/temas/down/informacion/sintomas>]

Diagnosis and Tests

- Alpha-Fetoprotein (AFP) Test [<https://medlineplus.gov/lab-tests/alpha-fetoprotein-afp-test/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-afp-alfafetoproteina>]
- Down Syndrome Tests [<https://medlineplus.gov/lab-tests/down-syndrome-tests/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-sindrome-de-down>]
- How Do Health Care Providers Diagnose Down Syndrome?
[<https://www.nichd.nih.gov/health/topics/down/conditioninfo/diagnosis>] 
(Eunice Kennedy Shriver National Institute of Child Health and Human Development)
Also in Spanish [<https://espanol.nichd.nih.gov/salud/temas/down/informacion/diagnostica>]

Treatments and Therapies

- What Are Common Treatments for Down Syndrome?
[<https://www.nichd.nih.gov/health/topics/down/conditioninfo/treatments>] 
(Eunice Kennedy Shriver National Institute of Child Health and Human Development)
Also in Spanish [<https://espanol.nichd.nih.gov/salud/temas/down/informacion/tratamientos>]

Related Issues

- ADHD and Down Syndrome [<https://ndss.org/resources/adhd-down-syndrome>] (National Down Syndrome Society)
- Alzheimer's Disease in People with Down Syndrome [<https://www.nia.nih.gov/health/alzheimers-causes-and-risk-factors/alzheimers-disease-people-down-syndrome>]  (National Institute on Aging)
- Blood Diseases and Down Syndrome [<https://ndss.org/resources/blood-diseases-syndrome>] (National Down Syndrome Society)
- Dentistry and Down Syndrome [<https://ndss.org/resources/dentistry-down-syndrome>] (National Down Syndrome Society)
- Down Syndrome: Eye Problems [<https://www.aapos.org/glossary/down-syndrome>] (American Association for Pediatric Ophthalmology and Strabismus)
- Dual Diagnosis of Down Syndrome and Autism [<https://ndss.org/resources/dual-diagnosis-syndrome-autism>] (National Down Syndrome Society)
- Ear, Nose, and Throat (ENT) and Down Syndrome [<https://ndss.org/resources/ear-nose-throat-ent-down-syndrome>] (National Down Syndrome Society)
- Endocrine Conditions and Down Syndrome [<https://ndss.org/resources/endocrine-conditions-syndrome>] (National Down Syndrome Society)
- Gastrointestinal System and Down Syndrome [<https://ndss.org/resources/gastrointestinal-tract-syndrome>] (National Down Syndrome Society)
- Mental Health and Down Syndrome [<https://ndss.org/resources/mental-health-down-syndrome>] (National Down Syndrome Society)
- Relationships & Sexuality [<https://ndss.org/resources/relationships-sexuality>] (National Down Syndrome Society)
- Sleep and Down Syndrome [<https://ndss.org/resources/sleep-down-syndrome>] (National Down Syndrome Society)
- What Causes Down Syndrome? [<https://www.nichd.nih.gov/health/topics/down/conditioninfo/causes>] 
(Eunice Kennedy Shriver National Institute of Child Health and Human Development)
Also in Spanish [<https://espanol.nichd.nih.gov/salud/temas/down/informacion/causas>]

- What Conditions or Disorders Are Commonly Associated with Down Syndrome?

[<https://www.nichd.nih.gov/health/topics/down/conditioninfo/associated>]

 (Eunice Kennedy Shriver National Institute of Child Health and Human Development)

Also in Spanish [<https://espanol.nichd.nih.gov/salud/temas/down/informacion/asociarse>]

Genetics

- About Down Syndrome [<https://www.genome.gov/Genetic-Disorders/Down-Syndrome>]

 (National Human Genome Research Institute)

- Down syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/down-syndrome>]

 (National Library of Medicine)

Statistics and Research

- DS-Connect: The Down Syndrome Registry [<https://ds-connect.org/>]

(Eunice Kennedy Shriver National Institute of Child Health and Human Development)

- Welcome to the Down Syndrome Consortium [<https://downsyndrome.nih.gov/>]

 (Eunice Kennedy Shriver National Institute of Child Health and Human Development)

- Who Is at Risk for Down Syndrome? [<https://www.nichd.nih.gov/health/topics/down/conditioninfo/Risks>]

 (Eunice Kennedy Shriver National Institute of Child Health and Human Development)

Also in Spanish [<https://espanol.nichd.nih.gov/salud/temas/down/informacion/riesgo>]

Clinical Trials

- ClinicalTrials.gov: Down Syndrome [<https://clinicaltrials.gov/search?cond=%22Down+Syndrome%22&aggFilters=status:not%20rec>]

 (National Institutes of Health)

Journal Articles

References and abstracts from MEDLINE/PubMed (National Library of Medicine)

- Article: Surgical management strategies for atlantoaxial instability/dislocation in down syndrome. [<https://www.ncbi.nlm.nih.gov/pubmed/40745318>]

- Article: Lifestyle Intervention Improves Metabolic Dysfunction-Associated Steatotic Liver Disease in Children with... [<https://www.ncbi.nlm.nih.gov/pubmed/40732956>]

- Article: A meta-analysis and systematic review of interventions to prevent or treat... [<https://www.ncbi.nlm.nih.gov/pubmed/40709486>]

- Down Syndrome -- see more articles [<https://pubmed.ncbi.nlm.nih.gov/?term=%22Down+Syndrome%22%5Bmajr%3Aexp%5D+AND+humans%5Bmh%5D+AND+english%5Bla%5D+AND+%22last+1+Year%22+%5Bdat%5D+NOT+%28letter%5Bpt%5D+OR+case+reports%5Bpt%5D+OR+editorial%5Bpt%5D+OR+comment%5Bpt%5D%29+AND+free+full+text%5Bsb%5D+>]

Find an Expert

- Centers for Disease Control and Prevention [<https://www.cdc.gov/>]

Also in Spanish [<https://www.cdc.gov/spanish/>]

- Eunice Kennedy Shriver National Institute of Child Health and Human Development [<https://www.nichd.nih.gov/>]

 Also in Spanish [<https://espanol.nichd.nih.gov/>]

- March of Dimes Foundation [<https://www.marchofdimes.org/>]

Also in Spanish [<https://nacersano.marchofdimes.org/>]

- National Down Syndrome Society [<https://ndss.org/>]

Children

- Down Syndrome (For Kids) [<https://kidshealth.org/en/kids/down-syndrome.html>] (Nemours Foundation)

Also in Spanish [<https://kidshealth.org/es/kids/down-syndrome.html>]

Patient Handouts

- Down syndrome [<https://medlineplus.gov/ency/article/000997.htm>] (Medical Encyclopedia)
Also in Spanish [<https://medlineplus.gov/spanish/ency/article/000997.htm>]
- Nuchal translucency test [<https://medlineplus.gov/ency/article/007561.htm>] (Medical Encyclopedia)
Also in Spanish [<https://medlineplus.gov/spanish/ency/article/007561.htm>]



MEDICAL ENCYCLOPEDIA

[Down syndrome](https://medlineplus.gov/ency/article/000997.htm) [<https://medlineplus.gov/ency/article/000997.htm>]

[Duodenal atresia](https://medlineplus.gov/ency/article/001131.htm) [<https://medlineplus.gov/ency/article/001131.htm>]

[Karyotyping](https://medlineplus.gov/ency/article/003935.htm) [<https://medlineplus.gov/ency/article/003935.htm>]

[Nuchal translucency test](https://medlineplus.gov/ency/article/007561.htm) [<https://medlineplus.gov/ency/article/007561.htm>]

National Institutes of Health

The primary NIH organization for research on *Down Syndrome* is the Eunice Kennedy Shriver National Institute of Child Health and Human Development [<https://www.nichd.nih.gov/Pages/index.aspx>]

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