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Genetic Disorders

What are genetic disorders?

Genetic disorders are health conditions caused by changes (also called mutations or variants) in your genes. Genes are parts of DNA [<https://medlineplus.gov/genetics/understanding/basics/dna/>] found in your cells that carry instructions for how your body grows, develops, and functions. Many genes tell your body how to make proteins, which are needed for your body to work properly.

What causes genetic disorders?

A genetic disorder happens when a **gene variant**

[<https://medlineplus.gov/genetics/understanding/mutationsanddisorders/neutralmutations/>] changes how a protein is made.

The variant may cause the protein to work poorly or not be made at all. If genes don't make the right proteins, or don't make them correctly, it can lead to a genetic disorder.

Not all gene changes cause disease. Often, these changes have no effect. But sometimes, even a small change in DNA can affect how proteins are made.

Gene variants [<https://medlineplus.gov/genetics/understanding/mutationsanddisorders/genemutation/>] can be grouped into two main types:

- **Inherited** variants (also called germline variants) are passed down from your parents. They come from egg or sperm cells.
- **Non-inherited** (also called somatic variants) are not passed down from your parents. They happen during your lifetime. They may be caused by things like harmful chemicals or ultraviolet (UV) rays from the sun.

What are the types of genetic disorders?

Genetic disorders may be caused by:

- **Single gene disorders** are caused by changes in one gene. Examples can include sickle cell anemia [<https://medlineplus.gov/sicklecelldisease.html>] , where a change in a single gene can cause the condition, and Charcot-Marie-Tooth disease [<https://medlineplus.gov/charcotmarietoothdisease.html>] , where a variant in one of many different genes can cause the condition.
- **Chromosomal disorders** are caused by missing, extra, or altered chromosomes. Chromosomes are structures that carry genes. These disorders involve changes in the number [<https://medlineplus.gov/genetics/understanding/mutationsanddisorders/chromosomalconditions/>] of chromosomes people have or changes in the structure [<https://medlineplus.gov/genetics/understanding/mutationsanddisorders/structuralchanges/>] of one or more chromosomes. An example is Down syndrome [<https://medlineplus.gov/downsyndrome.html>] , which is caused by an extra copy of chromosome 21.
- **Complex (multifactorial) disorders** [<https://medlineplus.gov/genetics/understanding/mutationsanddisorders/complexdisorders/>] are caused by changes in many genes. Each change alone may not cause the disease, but together they increase the risk. Lifestyle and environmental factors, such as exercise, diet, or exposure to pollutants, also play a role. Colon cancer [<https://medlineplus.gov/colorectalcancer.html>] is an example.

What are the different ways a genetic disorder can be inherited?

Some genetic conditions are passed down through families (inherited) in one of several patterns, depending on the specific gene involved.

Patterns of inheritance [<https://medlineplus.gov/genetics/understanding/inheritance/inheritancepatterns/>] can include:

- **Dominant** means you only need one changed gene to cause the condition.
- **Recessive** means you need two changed copies of the gene (one from each parent) for the condition to occur.
- **X-linked** conditions involve genes located on the X chromosome. These conditions often affect males more frequently.
- **Mitochondrial** conditions are passed down by the mother.





How are genetic disorders diagnosed?

Your health care provider may check for a genetic condition [<https://medlineplus.gov/genetics/understanding/consult/diagnosis/>] based on:




- A physical examination.
- Your personal medical history.
- Your family health history.
- Laboratory tests, including genetic testing [<https://medlineplus.gov/genetictesting.html>] .

NIH: National Library of Medicine


Start Here

- Genetic and Rare Diseases Information Center [<https://rarediseases.info.nih.gov/>] 
- Genetic Disorders [<https://www.genome.gov/For-Patients-and-Families/Genetic-Disorders>]  (National Human Genome Research Institute)
- Genetics: MedlinePlus Genetics [<https://medlineplus.gov/genetics/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/>]
- How can gene variants affect health and development?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/mutationsanddisorders/mutationscausedisease/>]  (National Library of Medicine)


Diagnosis and Tests

- Genetic Testing: MedlinePlus Health Topic [<https://medlineplus.gov/genetictesting.html>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetictesting.html>]
- How are genetic conditions diagnosed?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/consult/diagnosis/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/entender/consulta/diagnostico/>]
- Newborn Screening: MedlinePlus Health Topic [<https://medlineplus.gov/newbornscreening.html>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/newbornscreening.html>]
- Progeria Research Foundation Diagnostic Testing Program [<https://www.progeriaresearch.org/the-prf-diagnostic-testing-program/>] (Progeria Research Foundation, Inc.)













Treatments and Therapies

- How are genetic conditions treated or managed?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/consult/treatment/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/entender/consulta/tratamiento/>]


Related Issues

- What Is Genetics? [<https://biobeat.nigms.nih.gov/2024/04/what-is-genetics/>]  (National Institute of General Medical Sciences)


Specifics

- About Poland Anomaly [<https://www.genome.gov/Genetic-Disorders/Poland-Anomaly>]  (National Human Genome Research Institute)
- About Progeria [<https://www.genome.gov/Genetic-Disorders/Progeria>]  (National Human Genome Research Institute)
- Cryopyrin-Associated Autoinflammatory Syndromes (CAPS) [<https://rheumatology.org/patients/cryopyrin-associated-autoinflammatory-syndromes-caps>] (American College of Rheumatology)
Also in Spanish [<https://rheumatology.org/patients/sindromes-autoinflamatorios-asociados-a-la-criopirina>]
- Frequently Asked Questions about Genetic Disorders [<https://www.genome.gov/For-Patients-and-Families/Health-FAQ>]  (National Human Genome Research Institute)
- Genetic and Chromosomal Conditions [<https://www.marchofdimes.org/find-support/topics/planning-baby/genetic-and-chromosomal-conditions>]  (March of Dimes Foundation)
Also in Spanish [<https://nacersano.marchofdimes.org/embarazo/condiciones-geneticas-y-cromosomicas.aspx>]
- Genetics: MedlinePlus Genetics [<https://medlineplus.gov/genetics/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/>]
- Infantile Neuroaxonal Dystrophy [<https://www.ninds.nih.gov/health-information/disorders/infantile-neuroaxonal-dystrophy>]  (National Institute of Neurological Disorders and Stroke)
- Noonan Syndrome [<https://www.mayoclinic.org/diseases-conditions/noonan-syndrome/symptoms-causes/syc-20354422?p=1>] (Mayo Foundation for Medical Education and Research)
- Triple X Syndrome: Symptoms and Causes [<https://www.mayoclinic.org/diseases-conditions/triple-x-syndrome/symptoms-causes/syc-20350977?p=1>] (Mayo Foundation for Medical Education and Research)
Also in Spanish [<https://www.mayoclinic.org/es/diseases-conditions/triple-x-syndrome/symptoms-causes/syc-20350977?p=1>]
- Undiagnosed Condition in an Adult FAQ [<https://www.genome.gov/FAQ/Learning-About-An-Undiagnosed-Condition-in-an-Adult>]  (National Human Genome Research Institute)
- VEXAS Syndrome [<https://www.niams.nih.gov/labs/grayson-lab/vexas>]  (National Institute of Arthritis and Musculoskeletal and Skin Diseases)
- What are complex or multifactorial disorders?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/mutationsanddisorders/complexdisorders/>]  (National Library of Medicine)
- What are the different ways a genetic condition can be inherited?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/inheritance/inheritancepatterns/>]  (National Library of Medicine)
- What does it mean if a disorder seems to run in my family?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/inheritance/runsinfamily/>]  (National Library of Medicine)
- Williams Syndrome [<https://rarediseases.info.nih.gov/diseases/7891/williams-syndrome>]  (Genetic and Rare Diseases Information Center)
Also in Spanish [<https://rarediseases.info.nih.gov/espanol/13102/sindrome-de-williams>]

Statistics and Research

- Human Genome Project (HGP) [<https://www.genome.gov/human-genome-project>]  (National Human Genome Research Institute)

Clinical Trials





- ClinicalTrials.gov: Genetic Diseases, Inborn [<https://clinicaltrials.gov/search?cond=%22Genetic+Diseases,+Inborn%22&aggFilters=status:not%20rec>]  (National Institutes of Health)

Journal Articles






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

- Article: Advancing genome-based precision medicine: a review on machine learning applications for... [https://www.ncbi.nlm.nih.gov/pubmed/40668553]
- Article: Gene Dosage Sensitivity and Human Genetic Diseases. [https://www.ncbi.nlm.nih.gov/pubmed/40633567]
- Article: Transcriptional Biomarkers in the Diagnosis of Genetic Disorders: Opportunities, Challenges, and... [https://www.ncbi.nlm.nih.gov/pubmed/40609991]
- Genetic Disorders -- see more articles [https://pubmed.ncbi.nlm.nih.gov/?term=%22Genetic+Disorders%2C+Inborn%22%5Bmajr%3A%22%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+]]

Reference Desk


- Chromosome Abnormalities [https://www.genome.gov/about-genomics/fact-sheets/Chromosome-Abnormalities-Fact-Sheet]
 (National Human Genome Research Institute)
Also in Spanish [https://www.genome.gov/es/about-genomics/fact-sheets/Anomal%3%ADas-Cromos%3%B3micas]
- Genetic Testing Registry [https://www.ncbi.nlm.nih.gov/gtr/]  (National Center for Biotechnology Information)
- How are genetic conditions and genes named?: MedlinePlus Genetics [https://medlineplus.gov/genetics/understanding/mutationsanddisorders/naming/]  (National Library of Medicine)
- Talking Glossary of Genomic and Genetic Terms [https://www.genome.gov/genetics-glossary]
 (National Human Genome Research Institute)
Also in Spanish [https://www.genome.gov/es/genetics-glossary]

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/]
- Genetic and Rare Diseases Information Center [https://rarediseases.info.nih.gov/] 
- How can I find a genetics professional in my area?: MedlinePlus Genetics [https://medlineplus.gov/genetics/understanding/consult/findingprofessional/]  (National Library of Medicine)
- National Human Genome Research Institute [https://www.genome.gov/] 
- National Institute of Neurological Disorders and Stroke [https://www.ninds.nih.gov/] 

Children

- Campomelic Dysplasia (For Parents) [https://kidshealth.org/en/parents/campomelic-dysplasia.html] (Nemours Foundation)
Also in Spanish [https://kidshealth.org/es/parents/campomelic-dysplasia.html]
- Cartilage Hair Hypoplasia (For Parents) [https://kidshealth.org/en/parents/cartilage-hair-hypoplasia.html] (Nemours Foundation)
Also in Spanish [https://kidshealth.org/es/parents/cartilage-hair-hypoplasia.html]
- CLOVES Syndrome (For Parents) [https://kidshealth.org/en/parents/cloves-syndrome.html] (Nemours Foundation)
Also in Spanish [https://kidshealth.org/es/parents/cloves-syndrome.html]
- Ellis-Van Creveld Dysplasia (For Parents) [https://kidshealth.org/en/parents/ellis-dysplasia.html] (Nemours Foundation)
Also in Spanish [https://kidshealth.org/es/parents/ellis-dysplasia.html]

- **Gene Changes (Mutations)** [<https://kidshealth.org/en/parents/gene-mutations.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/gene-mutations.html>]
- **Genetic Conditions: How Do They Run in Families? (For Parents)** [<https://kidshealth.org/en/parents/genetic-passed-down.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/genetic-passed-down.html>]
- **Genetics (For Parents)** [<https://kidshealth.org/en/parents/about-genetics.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/about-genetics.html>]
- **Multiple Epiphyseal Dysplasia (For Parents)** [<https://kidshealth.org/en/parents/multiple-epiphyseal-dysplasia.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/multiple-epiphyseal-dysplasia.html>]
- **Undiagnosed Condition in a Child FAQ** [<https://www.genome.gov/FAQ/Learning-About-an-Undiagnosed-Condition-in-a-Child>]  (National Human Genome Research Institute)
- **What Is a Gene?** [<https://kidshealth.org/en/kids/what-is-gene.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/kids/what-is-gene.html>]
- **What Is a Pediatric Geneticist?** [<https://www.healthychildren.org/English/family-life/health-management/pediatric-specialists/Pages/What-is-a-Pediatric-Geneticist.aspx>] (American Academy of Pediatrics)
Also in Spanish [<https://www.healthychildren.org/spanish/family-life/health-management/pediatric-specialists/paginas/what-is-a-pediatric-geneticist.aspx>]

Teenagers

- **Genes and Genetics** [<https://kidshealth.org/en/teens/genes-genetic-disorders.html>] (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/teens/genes-genetic-disorders.html>]

Women

- **Genetic Disorders and Pregnancy** [<https://www.acog.org/womens-health/faqs/genetic-disorders>] (American College of Obstetricians and Gynecologists)

Patient Handouts

- **Genetics** [<https://medlineplus.gov/ency/article/002048.htm>] (Medical Encyclopedia)
Also in Spanish [<https://medlineplus.gov/spanish/ency/article/002048.htm>]



MEDICAL ENCYCLOPEDIA

- Alstrom syndrome [<https://medlineplus.gov/ency/article/001665.htm>]
- Beriberi [<https://medlineplus.gov/ency/article/000339.htm>]
- Cystinuria [<https://medlineplus.gov/ency/article/000346.htm>]
- Genetics [<https://medlineplus.gov/ency/article/002048.htm>]

[Hereditary elliptocytosis \[https://medlineplus.gov/ency/article/000563.htm\]](https://medlineplus.gov/ency/article/000563.htm)

[Hereditary ovalocytosis \[https://medlineplus.gov/ency/article/000566.htm\]](https://medlineplus.gov/ency/article/000566.htm)

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

[Lipase test \[https://medlineplus.gov/ency/article/003465.htm\]](https://medlineplus.gov/ency/article/003465.htm)

[Mosaicism \[https://medlineplus.gov/ency/article/001317.htm\]](https://medlineplus.gov/ency/article/001317.htm)

[Nevoid basal cell carcinoma syndrome \[https://medlineplus.gov/ency/article/001452.htm\]](https://medlineplus.gov/ency/article/001452.htm)

[Noonan syndrome \[https://medlineplus.gov/ency/article/001656.htm\]](https://medlineplus.gov/ency/article/001656.htm)

[Noonan syndrome with multiple lentigines \[https://medlineplus.gov/ency/article/001473.htm\]](https://medlineplus.gov/ency/article/001473.htm)

[Peutz-Jeghers syndrome \[https://medlineplus.gov/ency/article/000244.htm\]](https://medlineplus.gov/ency/article/000244.htm)

[Progeria \[https://medlineplus.gov/ency/article/001657.htm\]](https://medlineplus.gov/ency/article/001657.htm)

[Williams syndrome \[https://medlineplus.gov/ency/article/001116.htm\]](https://medlineplus.gov/ency/article/001116.htm)

Related Health Topics

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[Genetic Testing \[https://medlineplus.gov/genetictesting.html\]](https://medlineplus.gov/genetictesting.html)

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[Osteogenesis Imperfecta \[https://medlineplus.gov/osteogenesisimperfecta.html\]](https://medlineplus.gov/osteogenesisimperfecta.html)

[Prader-Willi Syndrome \[https://medlineplus.gov/praderwillisyndrome.html\]](https://medlineplus.gov/praderwillisyndrome.html)

[Rare Diseases \[https://medlineplus.gov/rarediseases.html\]](https://medlineplus.gov/rarediseases.html)

[Sickle Cell Disease \[https://medlineplus.gov/sicklecelldisease.html\]](https://medlineplus.gov/sicklecelldisease.html)

National Institutes of Health

The primary NIH organization for research on *Genetic Disorders* 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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