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Genetic Testing

What is genetic testing?

Genetic testing is a type of medical test that looks for changes in your DNA. DNA is short for deoxyribonucleic acid. It contains the genetic instructions in all living things. Genetic tests analyze your cells or tissue to look for any changes in:

- **Genes**, which are parts of DNA that carry the information needed to make a protein
- **Chromosomes**, which are thread-like structures in your cells. They contain DNA and proteins.
- **Proteins**, which do most of the work in your cells. Testing can look for changes in the amount and activity level of proteins. If it finds changes, it might be due to changes in your DNA.

Why is genetic testing done?

Genetic testing may be done for many different reasons, including to:

- Find genetic diseases [<https://medlineplus.gov/geneticdisorders.html>] in unborn babies. This is one type of prenatal testing [<https://medlineplus.gov/prenataltesting.html>] .
- Screen newborn babies [<https://medlineplus.gov/newbornscreening.html>] for certain treatable conditions
- Lower the risk of genetic diseases in embryos that were created using assisted reproductive technology [<https://medlineplus.gov/assistedreproductivetechnology.html>]
- Find out if you carry a gene for a certain disease that could be passed on to your children. This is called carrier testing.
- See whether you are at increased risk of developing a specific disease. This may be done for a disease that runs in your family.
- Diagnose certain diseases
- Identify genetic changes that may be causing or contributing to a disease that you were already diagnosed with
- Figure out how severe a disease is
- Help guide your doctor in deciding the best medicine and dosage for you. This is called pharmacogenomic testing [<https://medlineplus.gov/lab-tests/pharmacogenetic-tests/>] .

How is genetic testing done?

Genetic tests are often done on a blood or cheek swab sample. But they may also be done on samples of hair, saliva, skin, amniotic fluid (the fluid that surrounds a fetus during pregnancy), or other tissue. The sample is sent to a laboratory. There, a lab technician will use one of several different techniques to look for genetic changes.

What are the benefits of genetic testing?

The benefits of genetic testing include:

- Helping doctors make recommendations for treatment or monitoring
- Giving you more information for making decisions about your health and your family's health:
 - If you find out that you are at risk for a certain disease, you might take steps to lower that risk. For example, you may find out that you should be screened for a disease earlier and more often. Or you might decide to make healthy lifestyle changes [<https://medlineplus.gov/healthyliving.html>] .
 - If you find out that you are not at risk for a certain disease, then you can skip unnecessary checkups or screenings

- A test could give you information that helps you make decisions about having children
- Identifying genetic disorders early in life so treatment can be started as soon as possible

What are the drawbacks of genetic testing?




The physical risks of the different types of genetic testing are small. But there can be emotional, social, or financial drawbacks:

- Depending on the results, you may feel angry, depressed, anxious, or guilty. This can be especially true if you are diagnosed with a disease that does not have effective treatments.
- You may be worried about genetic discrimination in employment or insurance
- Genetic testing may give you limited information about a genetic disease. For example, it cannot tell you whether you will have symptoms, how severe a disease might be, or whether a disease will get worse over time.
- Some genetic tests are expensive, and health insurance might only cover part of the cost. Or they may not cover it at all.






How do I decide whether to be tested?





The decision about whether to have genetic testing is complex. In addition to discussing the test with your health care provider, you can meet with a **genetic counselor** [<https://medlineplus.gov/geneticcounseling.html>]. Genetic counselors have specialized degrees and experience in genetics and counseling. They can help you understand the tests and weigh the risks and benefits. If you do get a test, they can explain the results and make sure that you have the support that you need.

Start Here











- Genetic Testing [<https://www.cdc.gov/genomics-and-health/about/genetic-testing.html>]
(Centers for Disease Control and Prevention)
- Genetic Testing (For Parents) [<https://kidshealth.org/en/parents/genetics.html>]  (Nemours Foundation)
Also in Spanish [<https://kidshealth.org/es/parents/genetics.html>]
- Genetic Testing FAQ [<https://www.genome.gov/FAQ/Genetic-Testing>]  (National Human Genome Research Institute)
Also in Spanish [<https://www.genome.gov/es/FAQ/Preguntas-frecuentes-sobre-las-pruebas-geneticas>]
- Genetic Testing: What You Should Know [<https://familydoctor.org/genetic-testing-what-you-should-know/?adfree=true>]
(American Academy of Family Physicians)
Also in Spanish [<https://es.familydoctor.org/pruebas-geneticas-lo-que-usted-deberia-saber/?adfree=true>]
- Regulation of Genetic Tests [<https://www.genome.gov/about-genomics/policy-issues/Regulation-of-Genetic-Tests>]
 (National Human Genome Research Institute)
Also in Spanish [<https://www.genome.gov/es/about-genomics/policy-issues/Regulation-of-Genetic-Tests>]

Related Issues


- At-Home Medical Tests [<https://medlineplus.gov/lab-tests/at-home-medical-tests/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/pruebas-medicinas-en-el-hogar/>]
- Genetics: MedlinePlus Genetics [<https://medlineplus.gov/genetics/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/>]
- How can I be sure a genetic test is valid and useful?: MedlinePlus Genetics
[<https://medlineplus.gov/genetics/understanding/testing/validtest/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/entender/pruebas/pruebasvalidas/>]
- How is genetic testing done?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/testing/procedure/>]
 (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/entender/pruebas/procedimiento/>]
- Privacy in Genomics [<https://www.genome.gov/about-genomics/policy-issues/Privacy>]
 (National Human Genome Research Institute)
- Taking a Genetic Test on Your Own: What You Need to Know [<https://www.cdc.gov/breast-ovarian-cancer-hereditary/testing/taking-a-genetic-test-on-your-own.html>] (Centers for Disease Control and Prevention)

- Talking with Family Members about Genetic Testing [<https://www.cancer.org/cancer/risk-prevention/genetics/genetic-testing-for-cancer-risk/talking-with-family-members-about-genetic-testing.html>] (American Cancer Society)
- What are the risks and limitations of genetic testing?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/testing/riskslimitations/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/entender/pruebas/riesgoslimitaciones/>]
- What do the results of genetic tests mean?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/testing/interpretingresults/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/entender/pruebas/significadorestados/>]
- 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>]
- What is direct-to-consumer genetic testing?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/dtcgenetictesting/directtoconsumer/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/entender/pruebasdirectasalconsumidor/queson/>]
- What Is Genetics? [<https://biobeat.nigms.nih.gov/2024/04/what-is-genetics/>]  (National Institute of General Medical Sciences)

Specifics


- About Lynch Syndrome [<https://www.cdc.gov/colorectal-cancer-hereditary/about/lynch-syndrome.html>] (Centers for Disease Control and Prevention)
- BRCA Gene Changes: Cancer Risk and Genetic Testing [<https://www.cancer.gov/about-cancer/causes-prevention/genetics/brca-fact-sheet>]  (National Cancer Institute)
Also in Spanish [<https://www.cancer.gov/espanol/cancer/causas-prevencion/genetica/hoja-informativa-brca>]
- BRCA Genetic Test [<https://medlineplus.gov/lab-tests/brca-test/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-brca/>]
- Carrier Screening [<https://www.acog.org/womens-health/faqs/carrier-screening>] (American College of Obstetricians and Gynecologists)
- Genetic Testing for Inherited Cancer Risk [<https://www.cancer.gov/about-cancer/causes-prevention/genetics/genetic-testing-fact-sheet>]  (National Cancer Institute)
- Karyotype Genetic Test [<https://medlineplus.gov/lab-tests/karyotype-genetic-test/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-cariotipo/>]
- MTHFR Mutation Test [<https://medlineplus.gov/lab-tests/mthfr-mutation-test/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-mutaciones-de-mthfr/>]
- Pharmacogenetic Tests [<https://medlineplus.gov/lab-tests/pharmacogenetic-tests/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/pruebas-farmacogeneticas/>]
- PTEN Genetic Test [<https://medlineplus.gov/lab-tests/pten-genetic-test/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-genetica-del-pten/>]
- TP53 Genetic Test [<https://medlineplus.gov/lab-tests/tp53-genetic-test/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-genetica-tp-53-proteina-tumoral-53/>]
- What are the uses of genetic testing?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/testing/uses/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/entender/pruebas/usos/>]
- What is genetic ancestry testing?: MedlinePlus Genetics [<https://medlineplus.gov/genetics/understanding/dtcgenetictesting/ancestrytesting/>]  (National Library of Medicine)
Also in Spanish [<https://medlineplus.gov/spanish/genetica/entender/pruebasdirectasalconsumidor/pruebasdeascendencia/>]

Statistics and Research

- Genetic Mapping [<https://www.genome.gov/about-genomics/fact-sheets/Genetic-Mapping-Fact-Sheet>]  (National Human Genome Research Institute)

Also in Spanish [<https://www.genome.gov/es/about-genomics/fact-sheets/Hoja-de-datos-del-mapeo-gen%C3%A9tico>]

Clinical Trials




- ClinicalTrials.gov: Genetic Testing [<https://clinicaltrials.gov/search?intr=%22genetic+testing%22&aggFilters=status:not%20rec>]  (National Institutes of Health)

Journal Articles

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




- Article: Homologous recombination deficiency (HRD) tests for ovarian cancer: a multicenter French... [<https://www.ncbi.nlm.nih.gov/pubmed/40597907>]
- Article: Breast cancer germline multigene panel testing in mainstream oncology based on... [<https://www.ncbi.nlm.nih.gov/pubmed/40523834>]
- Article: Utilizing peer educators to increase genetic testing for prostate cancer among... [<https://www.ncbi.nlm.nih.gov/pubmed/40447525>]
- Genetic Testing -- see more articles [<https://pubmed.ncbi.nlm.nih.gov/?term=%22Genetic+Testing%22%5Bmajr%3Anoexp%5D+AND+humans%5Bmh%5D+AND+english%5Bla%5D+AND+%22last+1+Year%22+%5Bedat%5D+AND+%28patient+education+handout%5Bpt%5D+OR+guideline%5Bpt%5D+OR+clinical+trial%5Bpt%5D%29++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

- GeneReviews Glossary [<https://www.ncbi.nlm.nih.gov/books/NBK5191/>]  (National Center for Biotechnology Information)
- Genetic Testing Registry [<https://www.ncbi.nlm.nih.gov/gtr/>]  (National Center for Biotechnology Information)
- 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/>]
- 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/>] 

Patient Handouts

- BRCA1 and BRCA2 gene testing [<https://medlineplus.gov/ency/patientinstructions/000690.htm>] (Medical Encyclopedia)
Also in Spanish [<https://medlineplus.gov/spanish/ency/patientinstructions/000690.htm>]
- Genetic testing and your cancer risk [<https://medlineplus.gov/ency/patientinstructions/000842.htm>] (Medical Encyclopedia)
Also in Spanish [<https://medlineplus.gov/spanish/ency/patientinstructions/000842.htm>]



MEDICAL ENCYCLOPEDIA

BRCA1 and BRCA2 gene testing [<https://medlineplus.gov/ency/patientinstructions/000690.htm>]

Buccal smear [<https://medlineplus.gov/ency/article/003414.htm>]

Genetic testing and your cancer risk [<https://medlineplus.gov/ency/patientinstructions/000842.htm>]

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

Related Health Topics

Birth Defects [<https://medlineplus.gov/birthdefects.html>]

Genetic Counseling [<https://medlineplus.gov/geneticcounseling.html>]

Genetic Disorders [<https://medlineplus.gov/geneticdisorders.html>]

Newborn Screening [<https://medlineplus.gov/newbornscreening.html>]

Prenatal Testing [<https://medlineplus.gov/prenataltesting.html>]

National Institutes of Health

The primary NIH organization for research on *Genetic Testing* is the National Human Genome Research Institute [<http://www.genome.gov/>]

NIH MedlinePlus Magazine

Do You Know Your Family Health History? [<https://magazine.medlineplus.gov/article/do-you-know-your-family-health-history>]

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