

ABCs of DNA

HELPING PATRONS UNRAVEL THE MYSTERY OF GENETIC INFORMATION

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**Genetics
Overview**

**Genomic
Health
Literacy**

**Genetic
Testing**

**Consumer
Health
Resources**

**Societal
Issues**

***All of Us*
Research
Program**



NNLM...?

NIH

- **National Institutes of Health**
- Nation's research agency

NLM

- **National Library of Medicine**
- World's largest biomedical library

NNLM

- **National Network of Libraries of Medicine**
- Program of the NLM comprised of 8 Regional Libraries (RMLs) and 5 offices

PNR

- **Pacific Northwest Region (NNLM PNR)**
- Is one of the 8 RMLs
- Serves Alaska, Idaho, Montana, Oregon, Washington



NNLM



U.S. National Library of Medicine

National Network of Libraries of Medicine
Pacific Northwest Region



The mission of NNLM is to advance the progress of medicine and improve the public health by:

- Providing all U.S. health professionals with equal access to biomedical information
- Improving the public's access to information to enable them to make informed decisions about their health

NNLM

<https://nnlm.gov/>



U.S. National Library of Medicine

National Network of Libraries of Medicine
Pacific Northwest Region

Genetics in the News

CRISPR Gene-Editing May Offer Path To Cure For HIV, First Published Report Shows

As Made-To-Order DNA Gets Cheaper, Keeping It Out Of The Wrong Hands Gets Harder

Opioids: Can a Genetic Test Identify an Addict in the Making?

Afraid of Needles? You May Want to Blame Your Genes

No 'gay gene': Massive study homes in on genetic basis of human sexuality

Signing up for 23andMe? You might be exposing your family to the FBI

Millions of Americans Could Be Identified Using Consumer Genetic Databases—Even If They've Never Taken a DNA Test

Birth of Baby With Three Parents' DNA Marks Success for Banned Technique



U.S. National Library of Medicine

National Network of Libraries of Medicine
Pacific Northwest Region

Genomic Health Literacy

- Lack biology basics
- Lack mathematical concepts
- Low health literacy



Leading causes of death

1. Heart disease: 647,457
2. Cancer: 599,108
3. Accidents (unintentional injuries): 169,936
4. Chronic lower respiratory diseases: 160,201
5. Stroke (cerebrovascular diseases): 146,383
6. Alzheimer's disease: 121,404
7. Diabetes: 83,564
8. Influenza and pneumonia: 55,672
9. Nephritis, nephrotic syndrome, and nephrosis: 50,633
10. Intentional self-harm (suicide): 47,173

[CDC FastStats](#)

The Story of You



[The Story of You](#)

Genetic Testing

including Direct-to-Consumer

Types of Genetic Tests

Diagnostic

Predictive

Carrier

Prenatal

Newborn
Screening

Research

Pharmacogenetic

Jean's Genetic Testing Timeline

- **Age 1 day: newborn** testing for a few serious childhood diseases
- **Age 30: carrier** testing (with her partner) before getting pregnant
- **Age 35: predictive testing** when sister develops breast cancer at a young age
- **Age 45 direct to consumer** genetic testing to investigate ancestry
- **Age 65 pharmacogenomics** testing when Plavix wasn't effective



Genetic Testing Results

What genes and what variants did you test for?

- Different tests offered for the same conditions
- Knowledge is always changing

Might not have enough examples in the database to determine association between specific variants and specific conditions.

Might not have enough examples of people like you in the database.

Possibility of false positives and false negative results.

Genetic Testing- is it necessary?

Before testing:

- You think about your reasons for wanting the test
- You get the right test
- You and your family are prepared for the results
- You have a personalized plan for dealing with the results

Genetic Counselors

- Evaluate family history and medical records
- Assist in making decisions regarding genetic testing
- Identify and interpret risks of inherited disorders, increase the family's understanding of a genetic condition
- Discuss options regarding disease management and the risks and benefits of further testing and other options
- Help the individual and family identify the psychosocial tools required to cope with potential outcomes
- Reduce the family's anxiety

[National Society of Genetic Counselors](#)

Direct to Consumer Testing



Testing for talent



Achieve
your full potential.



Increase
your athletic performance.



Harness
your natural ability through
personalized genomics.


www.GenomicExpress.com

DNA dating



Pheramor



American College of Medical Genetics and Genomics

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ACMG STATEMENT | Genetics
in Medicine

Direct-to-consumer genetic testing: a revised position statement of the American College of Medical Genetics and Genomics

ACMG Board of Directors¹

Disclaimer: These recommendations are designed primarily as an educational resource for medical geneticists and other health-care providers to help them provide quality medical genetics services. Adherence to these recommendations does not necessarily assure a successful medical outcome. These recommendations should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, geneticists and other

clinicians should apply their own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from the recommendations.

Genet Med advance online publication 17 December 2015

Key Words: consumer; direct-to-consumer; genetic testing; self-testing; OTC

With ongoing genetic discoveries and improvements in technology, more genetic tests are available than ever before. Along with greater availability has come increased consumer demand for genetic tests and expansion of direct-to-consumer testing. The American College of Medical Genetics and Genomics (ACMG) has revised its 2008 e-publication regarding this issue (ACMG Statement on Direct-to-Consumer Genetic Testing, retired; available by request to acmg@acmg.net) and believes that it is critical for the public to realize that genetic testing is only one part of a complex process that includes genetic risk

- A genetics expert such as a certified medical geneticist or genetic counselor should be available to help the consumer determine, for example, whether a genetic test should be performed and how to interpret test results in light of personal and family history. A board-certified genetic counselor can help facilitate this process by providing information about the test and helping to explain test results. A number of risks can be reduced if a board-certified genetics professional is involved in genetic testing, including inadequate or lack of informed consent.

ACMG



MedlinePlus

NIH U.S. National Library of Medicine

MedlinePlus
Trusted Health Information for You

Search MedlinePlus

About MedlinePlus Site Map FAQs Customer Support

Health Topics Drugs & Supplements Videos & Tools

Home → Health Topics → Genetic Testing

Genetic Testing

On this page

Basics <ul style="list-style-type: none">SummaryStart Here	Learn More <ul style="list-style-type: none">Related IssuesSpecifics	See, Play and Learn <ul style="list-style-type: none">No links available
Research <ul style="list-style-type: none">Statistics and ResearchClinical TrialsJournal Articles	Resources <ul style="list-style-type: none">Reference DeskFind an Expert	For You <ul style="list-style-type: none">Patient Handouts

Summary

Genetic tests are tests on blood and other tissue to find **genetic disorders**. Over 2000 tests are available. Doctors use genetic tests for several reasons. These include

- Finding genetic diseases in unborn babies
- Finding out if people carry a gene for a disease and might pass it on to their children
- Screening embryos for disease
- Testing for genetic diseases in adults before they cause symptoms
- Making a diagnosis in a person who has disease symptoms
- Figuring out the type or dose of a medicine that is best for a certain person

People have many different reasons for being tested or not being tested. For some, it is important to know whether a disease can be prevented or treated if a test is positive. In some cases, there is no treatment. But test results might help a person make life decisions, such as family planning or insurance coverage. A **genetic counselor** can provide information about the pros and cons of testing.

NIH: National Human Genome Research Institute

Start Here

Get Genetic Testing updates email

MEDICAL ENCYCLOPEDIA

- BRCA1 and BRCA2 gene tests
- Buccal smear
- Genetic testing and your cancer
- Karyotyping

Related Health Topics

FEDERAL TRADE COMMISSION

CONSUMER INFORMATION

Search

ESPAÑOL

Veá esta página en español

Direct-to-Consumer Genetic Tests

Could a simple medical test tell you if you are likely to get a particular disease? Could it evaluate your health risks and even suggest a specific treatment? Could you take this test in the privacy of your home, without a doctor's prescription or guidance?

Some companies say genetic testing can do all this and more. They claim that direct-to-consumer (DTC) genetic testing can screen for diseases and provide a basis for choosing a particular diet, dietary supplement, lifestyle change, or medication. These companies primarily sell their tests online and through multi-level marketing networks.

The Federal Trade Commission (FTC) wants you to know the facts about the DTC marketing of genetic tests.

- **Genes and Genetic Tests**
- **Interpreting the Results**
- **Company Claims**
- **If You're Considering a DTC Genetic Test**
- **For More Information**

Related Items

Anatomy of a Cancer Treatment Scam

- **Dietary Supplements**
- **Miracle Health Claims**
- **Cancer Treatment Scams**

MONEY & CREDIT

HOMES & MORTGAGES

HEALTH & FITNESS

Healthy Living

Treatments & Cures

Weight Loss & Fitness

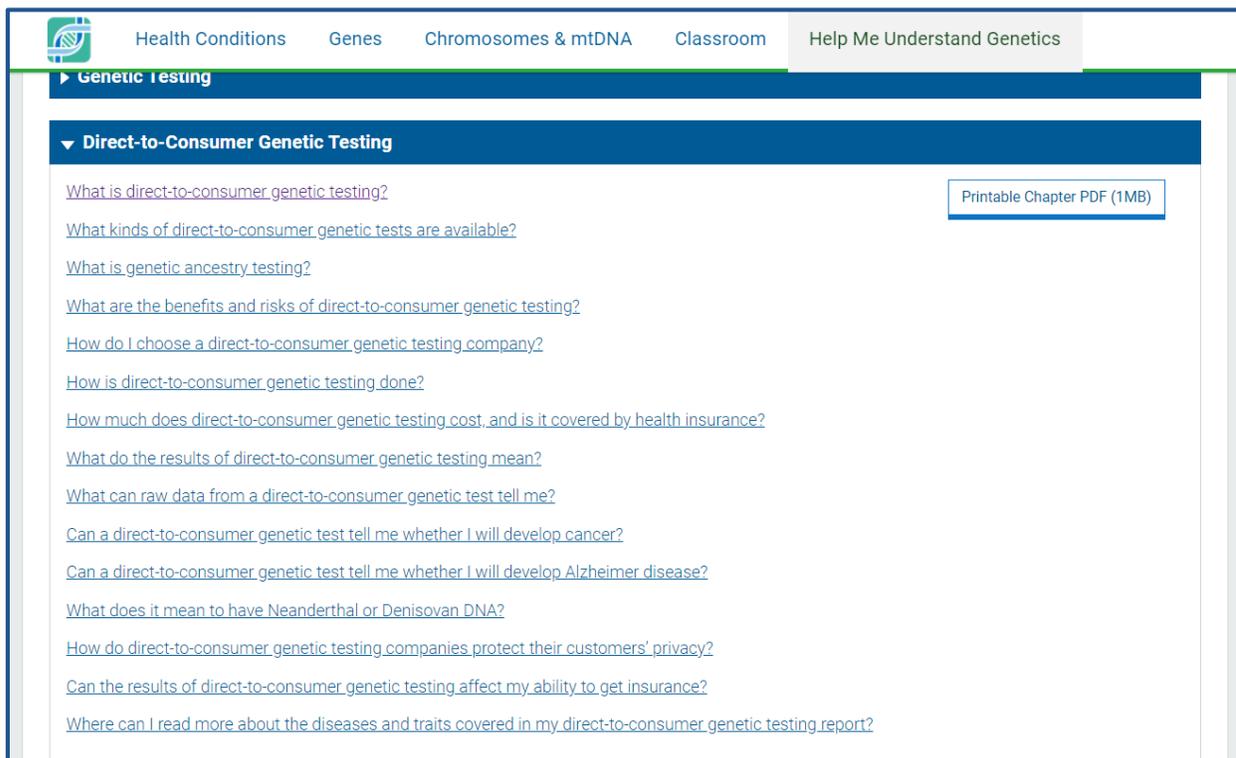
JOB & MAKING MONEY

PRIVACY, IDENTITY & ONLINE SECURITY

BLOG

VIDEO & MEDIA

Genetics Home Reference - DTC



Health Conditions Genes Chromosomes & mtDNA Classroom Help Me Understand Genetics

Genetic Testing

Direct-to-Consumer Genetic Testing

[What is direct-to-consumer genetic testing?](#) Printable Chapter PDF (1MB)

[What kinds of direct-to-consumer genetic tests are available?](#)

[What is genetic ancestry testing?](#)

[What are the benefits and risks of direct-to-consumer genetic testing?](#)

[How do I choose a direct-to-consumer genetic testing company?](#)

[How is direct-to-consumer genetic testing done?](#)

[How much does direct-to-consumer genetic testing cost, and is it covered by health insurance?](#)

[What do the results of direct-to-consumer genetic testing mean?](#)

[What can raw data from a direct-to-consumer genetic test tell me?](#)

[Can a direct-to-consumer genetic test tell me whether I will develop cancer?](#)

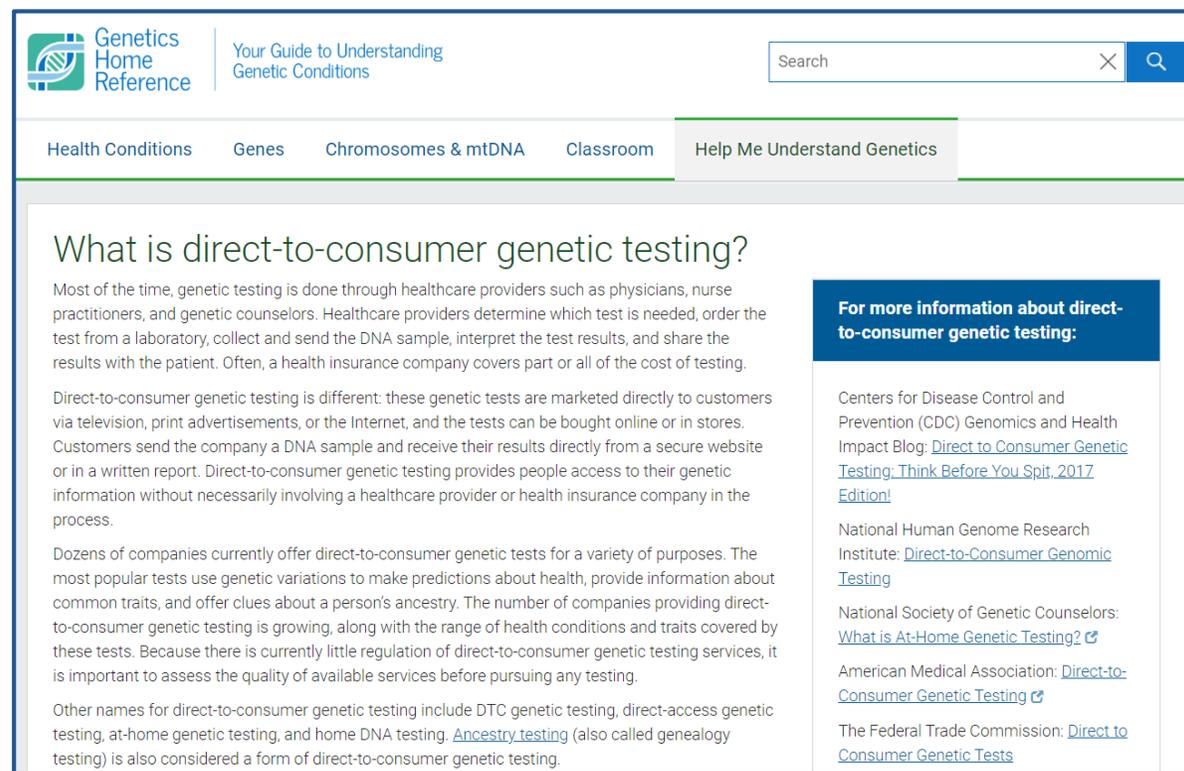
[Can a direct-to-consumer genetic test tell me whether I will develop Alzheimer disease?](#)

[What does it mean to have Neanderthal or Denisovan DNA?](#)

[How do direct-to-consumer genetic testing companies protect their customers' privacy?](#)

[Can the results of direct-to-consumer genetic testing affect my ability to get insurance?](#)

[Where can I read more about the diseases and traits covered in my direct-to-consumer genetic testing report?](#)



Genetics Home Reference Your Guide to Understanding Genetic Conditions

Health Conditions Genes Chromosomes & mtDNA Classroom Help Me Understand Genetics

What is direct-to-consumer genetic testing?

Most of the time, genetic testing is done through healthcare providers such as physicians, nurse practitioners, and genetic counselors. Healthcare providers determine which test is needed, order the test from a laboratory, collect and send the DNA sample, interpret the test results, and share the results with the patient. Often, a health insurance company covers part or all of the cost of testing.

Direct-to-consumer genetic testing is different: these genetic tests are marketed directly to customers via television, print advertisements, or the Internet, and the tests can be bought online or in stores. Customers send the company a DNA sample and receive their results directly from a secure website or in a written report. Direct-to-consumer genetic testing provides people access to their genetic information without necessarily involving a healthcare provider or health insurance company in the process.

Dozens of companies currently offer direct-to-consumer genetic tests for a variety of purposes. The most popular tests use genetic variations to make predictions about health, provide information about common traits, and offer clues about a person's ancestry. The number of companies providing direct-to-consumer genetic testing is growing, along with the range of health conditions and traits covered by these tests. Because there is currently little regulation of direct-to-consumer genetic testing services, it is important to assess the quality of available services before pursuing any testing.

Other names for direct-to-consumer genetic testing include DTC genetic testing, direct-access genetic testing, at-home genetic testing, and home DNA testing. [Ancestry testing](#) (also called genealogy testing) is also considered a form of direct-to-consumer genetic testing.

For more information about direct-to-consumer genetic testing:

- Centers for Disease Control and Prevention (CDC) Genomics and Health Impact Blog: [Direct to Consumer Genetic Testing: Think Before You Spit, 2017 Edition!](#)
- National Human Genome Research Institute: [Direct-to-Consumer Genomic Testing](#)
- National Society of Genetic Counselors: [What is At-Home Genetic Testing?](#)
- American Medical Association: [Direct-to-Consumer Genetic Testing](#)
- The Federal Trade Commission: [Direct to Consumer Genetic Tests](#)

Concerns

- Privacy and legality
- Who has access?
- What all is being done now and in the future with the information?
- Unexpected surprises?
- Test results can vary among companies
- Validity of tests
- No counseling provided

Benefits

- Learn more about own health
- Learn more about ethnicity and family history
- Bring awareness to family health issues for future generations
- Motivation to work on health habits
- Encourages patient engagement
- Contributing to advancement of healthcare and science
- Moral obligation

Questions to ask before using a Direct to Consumer Genetic Test

- Is the test right for me?
- What are the company claims?
- What do I hope to find out?
- Am I ready to hear something unexpected?
- Who will the results affect besides me?
- What happens to my genetic information?



[Think Before You Spit – CDC](#)

Think After You Spit

- Have a healthy dose of skepticism
- Discuss and share the results of tests with health care providers
- Seek, collect and validate as much as possible family health history
- There are general disease prevention and health promotion messages that are important (stop smoking, exercise, etc.)
- Learn about health and disease and become involved in both family and patient-provider interactions

[Think After You Spit- CDC](#)

Consumer Resources

Consumer and K-12 Education

MedlinePlus

The screenshot shows the MedlinePlus website homepage. At the top, it features the U.S. National Library of Medicine logo and the MedlinePlus logo with the tagline "Trusted Health Information for You". A search bar is located in the top right corner. Below the header, there are navigation tabs for "Health Topics", "Drugs & Supplements", "Videos & Tools", and "Español". The main content area is divided into several sections: "Health Topics" (with an apple icon), "Drugs & Supplements" (with a pill icon), "Videos & Tools" (with a play button icon), "Medical Tests" (with a flask icon), and "Medical Encyclopedia" (with a book icon). There is also a "Healthy Recipes" button. A central banner features a photo of a doctor and text about "September is National Prostate Cancer Awareness Month". Below this, there are sections for "Tweets by @MedlinePlus" (showing a tweet about the opioid crisis), "Stay Connected" (with a newsletter sign-up form), "NIH MedlinePlus Magazine" (with a link to the latest issue), "All of Us" (with a link to research program participants), and "Clinical Trials" (with a link to search for drug and treatment studies). At the bottom, there are icons for "Easy-to-Read Materials", "Organizations and Directories", "Health Information in Multiple Languages", and "MedlinePlus Connect for EHRs". Social media links for RSS, Twitter, and Facebook are also present.

- Section: Genetics/Birth Defects
- Health Topic pages:
 - Genetics
 - Genetic testing
 - Genetic counseling
 - Genetic disorders
 - Genetic brain disorders
 - Genes and gene therapy
- text word search

[MedlinePlus](https://www.nlm.nih.gov/medlineplus/)



MedlinePlus – Genetics topics

[Health Topics](#) [Drugs & Supplements](#) [Videos & Tools](#)

[Home](#) → [Health Topics](#) → [Genetics/Birth Defects](#)

Genetics/Birth Defects

[Abnormalities](#) *see* [Birth Defects](#)

[Achondroplasia](#) *see* [Dwarfism](#)

[Adrenoleukodystrophy](#) *see* [Leukodystrophies](#)

[Alpha-1 Antitrypsin Deficiency](#)

[Amniocentesis](#) *see* [Prenatal Testing](#)

[Anencephaly](#) *see* [Neural Tube Defects](#)

[Arnold-Chiari Malformation](#) *see* [Chiari Malformation](#)

[Ataxia](#) *see* [Friedreich's Ataxia](#)

[Ataxia Telangiectasia](#)

[Birth Defects](#)

[Blood Coagulation Disorders](#) *see* [Hemophilia](#)

[Brain Disorders, Inborn Genetic](#) *see* [Genetic Brain Disorders](#)

[Brain Malformations](#)

[Canavan Disease](#) *see* [Leukodystrophies](#)

[Cephalic Disorders](#) *see* [Brain Malformations](#)

[Cerebral Palsy](#)

[Charcot-Marie-Tooth Disease](#)

[Chiari Malformation](#)

[Chorionic Villi Sampling](#) *see* [Prenatal Testing](#)

[Cleft Lip and Palate](#)

[Cleft Palate](#) *see* [Cleft Lip and Palate](#)

[Cleft Spine](#) *see* [Spina Bifida](#)

[Cloning](#)

[Color Blindness](#)

[Congenital Heart Defects](#)

MedlinePlus – stroke topic page

NIH U.S. National Library of Medicine

MedlinePlus
Trusted Health Information for You

Search MedlinePlus

About MedlinePlus Site Map FAQs Customer Support

Health Topics Drugs & Supplements Videos & Tools Español

Home → Health Topics → Stroke

Stroke

Also called: Brain attack, CVA

On this page

Basics <ul style="list-style-type: none">SummaryStart HereSymptomsDiagnosis and TestsPrevention and Risk FactorsTreatments and Therapies	Learn More <ul style="list-style-type: none">Related IssuesGenetics	See, Play and Learn <ul style="list-style-type: none">ImagesHealth Check ToolsVideos and Tutorials
Research <ul style="list-style-type: none">Statistics and ResearchClinical TrialsJournal Articles	Resources <ul style="list-style-type: none">Reference DeskFind an Expert	For You <ul style="list-style-type: none">ChildrenWomenSeniorsPatient Handouts

Get Stroke updates by email

Summary

A stroke is a medical emergency. Strokes happen when blood flow to your brain stops. Within minutes, brain cells begin to die. There are two kinds of stroke. The more common kind, called **ischemic stroke**, is caused by a **blood clot** that blocks or plugs a blood vessel in the brain. The other kind, called **hemorrhagic stroke**, is caused by a blood vessel that breaks and bleeds into the brain. "Mini-strokes" or transient ischemic attacks (TIAs), occur when the blood supply to the brain is briefly interrupted.

Symptoms of stroke are

- Sudden numbness or weakness of the face, arm or leg (especially on one side of the body)
- Sudden confusion, trouble speaking or understanding speech
- Sudden trouble seeing in one or both eyes
- Sudden trouble walking, dizziness, loss of balance or coordination
- Sudden severe headache with no known cause

Specifics

- Hemorrhagic Stroke: MedlinePlus Health Topic [NIH](#) (National Library of Medicine)
Also in Spanish
- Ischemic Stroke: MedlinePlus Health Topic [NIH](#) (National Library of Medicine)
Also in Spanish
- Spinal Cord Infarction [NIH](#) (National Institute of Neurological Disorders and Stroke)
- Wallenberg's Syndrome [NIH](#) (National Institute of Neurological Disorders and Stroke)

Genetics

- Genetics Home Reference: cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy [NIH](#) (National Library of Medicine)
- Genetics Home Reference: Grange syndrome [NIH](#) (National Library of Medicine)
- Genetics Home Reference: mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes [NIH](#) (National Library of Medicine)
- Genetics Home Reference: moyamoya disease [NIH](#) (National Library of Medicine)

Images

- Craniotomy - slideshow (Medical Encyclopedia)
Also in Spanish

Health Check Tools

- Test Your Stroke Knowledge [NIH](#) (National Institute of Neurological Disorders and Stroke)
- What's Your Stroke I.Q.? (American Heart Association)

Videos and Tutorials

- Know Stroke: Know the Signs, Act in Time Video [NIH](#) (National Institute of Neurological Disorders and Stroke)

Statistics and Research

- FastStats: Cerebrovascular Disease or Stroke (National Center for Health Statistics)
- Heart Disease and Stroke Statistics (American Heart Association)
- Preventing Stroke Deaths (Centers for Disease Control and Prevention)
Also in Spanish

Clinical Trials

- ClinicalTrials.gov: Carotid Stenosis [NIH](#) (National Institutes of Health)
- ClinicalTrials.gov: Cerebrovascular Disorders [NIH](#) (National Institutes of Health)



Get Stroke updates by email

- MEDICAL ENCYCLOPEDIA**
- BAER - brainstem auditory evoked response
 - Brain herniation
 - Brain surgery
 - Brain surgery - discharge
 - Craniotomy - slideshow
 - EFG

MedlinePlus – text search

The screenshot shows the MedlinePlus website interface. At the top, the NIH logo and 'U.S. National Library of Medicine' are visible. The MedlinePlus logo is prominently displayed. A search bar contains the text 'genetics' and a 'GO' button. Below the search bar, there are navigation links for 'About MedlinePlus', 'Site Map', 'FAQs', and 'Customer Support'. The main content area is titled 'Search Results' and features a 'Genetic Disorders' section with a blue header. This section contains text explaining that genes are the building blocks of heredity and that mutations can cause genetic disorders. To the left of the main content, there are two 'Refine by' sections: 'Refine by Type' and 'Refine by Format'. The 'Refine by Type' section lists categories like 'Health Topics (390)', 'External Health Links (4,757)', 'Drugs and Supplements (36)', 'Medical Encyclopedia (418)', 'MedlinePlus Magazine (205)', 'Multiple Languages', and 'National Institutes of Health (2,832)'. The 'Refine by Format' section lists 'PDF (153)', 'Images (36)', and 'Videos (19)'. At the bottom of the page, there are three search results listed, each with a title, a brief description, and a URL.

Text word search 'genetics'

Genetics Home Reference

The screenshot shows the homepage of the Genetics Home Reference website. At the top left is the NIH logo and the text "U.S. National Library of Medicine". To the right is a "Share This Page" button. Below this is the "Genetics Home Reference" logo and the tagline "Your Guide to Understanding Genetic Conditions". A search bar is located to the right of the tagline. Below the search bar is a navigation menu with links for "Health Conditions", "Genes", "Chromosomes & mtDNA", "Classroom", and "Help Me Understand Genetics". The main content area features a large banner with a DNA double helix and the text: "Genetics Home Reference provides consumer-friendly information about the effects of genetic variation on human health." Below the banner are two columns. The left column is titled "Health Conditions" and includes a circular icon with a DNA helix, a description: "Learn about the signs and symptoms, causes, and inheritance of more than 1,200 health conditions with a genetic basis.", and a "Browse A-Z" button. The right column is titled "Recently Added Pages" and lists "New Pages" including "SETBP1 disorder", "Dystonia 16", and "Attention-deficit/hyperactivity disorder", with a "More New Pages..." link. A photograph of a family is partially visible in the background of the "Health Conditions" section.

- Health conditions
- Genes
- Chromosomes and DNA
- Classroom Resources
- Help Me Understand Genetics (a handbook)

[Genetics Home Reference](#)

Genetics Home Reference- health conditions

The screenshot displays the Genetics Home Reference website. At the top left is the logo for Genetics Home Reference, with the tagline "Your Guide to Understanding Genetic Conditions". To the right is a search bar with a magnifying glass icon. Below the header is a navigation menu with tabs for "Health Conditions", "Genes", "Chromosomes & mtDNA", "Classroom", and "Help Me Understand Genetics". The "Health Conditions" tab is selected and highlighted. The main content area features a circular icon with a DNA helix and the heading "Health Conditions". Below this is a paragraph: "Explore the signs and symptoms, genetic cause, and inheritance pattern of various health conditions." Underneath is a grid of letter buttons from 0-9 to Z, with the letter 'A' highlighted in blue. Below the grid is a list of links for various conditions starting with 'A', such as "A-alpha1ipoprotein Neuropathy, see [Tangier disease](#)". On the right side of the page, there is a blue box with the heading "Learn More about Health Conditions" and three questions: "What does it mean if a disorder seems to run in my family?", "What are the different ways in which a genetic condition can be inherited?", and "What are complex or multifactorial disorders?".

Genetics Home Reference Williams syndrome

Genetics Home Reference
Your Guide to Understanding Genetic Conditions

Search

Health Conditions Genes Chromosomes & mtDNA Classroom Help Me Understand Genetics

Williams syndrome

Printable PDF Open All Close All

Description

Williams syndrome is a developmental disorder that affects many parts of the body. This condition is characterized by mild to moderate intellectual disability or learning problems, unique personality characteristics, distinctive facial features, and heart and blood vessel (cardiovascular) problems.

People with Williams syndrome typically have difficulty with visual-spatial tasks such as drawing and assembling puzzles, but they tend to do well on tasks that involve spoken language, music, and learning by repetition (rote memorization). Affected individuals have outgoing, engaging personalities and tend to take an extreme interest in other people. Attention deficit disorder (ADD), problems with anxiety, and phobias are common among people with this disorder.

Young children with Williams syndrome have distinctive facial features including a [broad forehead](#), a [short nose](#) with a broad tip, [full cheeks](#), and a [wide mouth](#) with full lips. Many affected people have dental problems such as teeth that are small, widely spaced, crooked, or missing. In older children and adults, the face appears longer and more gaunt.

A form of cardiovascular disease called [supravalvular aortic stenosis](#) (SVAS) occurs frequently in

Related Information

- [What does it mean if a disorder seems to run in my family?](#)
- [What is the prognosis of a genetic condition?](#)
- [Genetic and Rare Diseases Information Center](#)

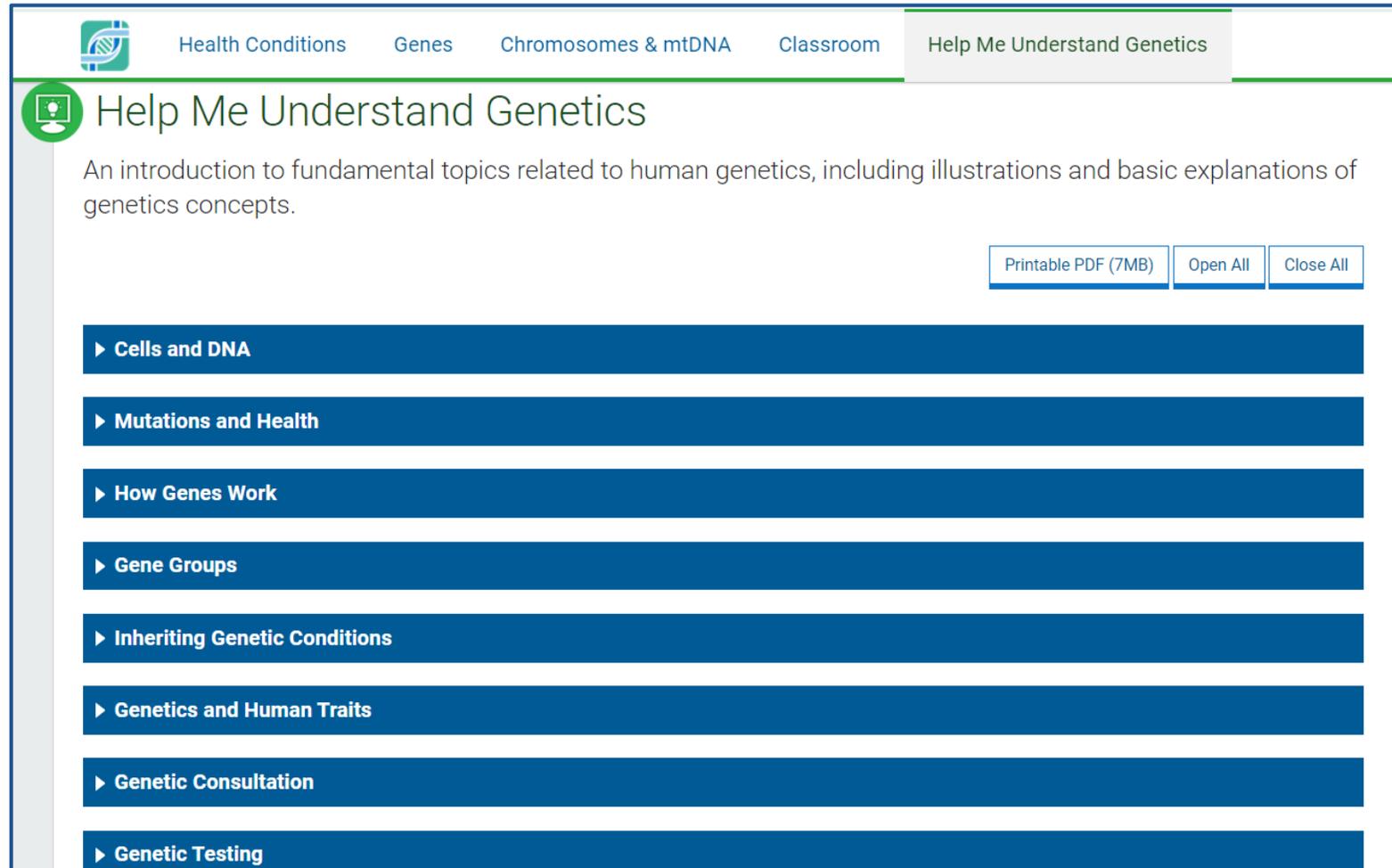
Health Conditions Genes Chromosomes & mtDNA Classroom Help Me Understand Genetics

Williams syndrome

Printable PDF Open All Close All

- ▶ Description
- ▶ Frequency
- ▶ Causes
- ▶ Inheritance Pattern
- ▶ Diagnosis & Management Links
- ▶ Other Names for This Condition
- ▶ Additional Information & Resources
- ▶ Sources for This Page

Genetics Home Reference- handbook



The screenshot shows the 'Help Me Understand Genetics' page from the Genetics Home Reference. At the top, there is a navigation bar with a DNA helix icon and links for 'Health Conditions', 'Genes', 'Chromosomes & mtDNA', 'Classroom', and 'Help Me Understand Genetics'. The main heading is 'Help Me Understand Genetics' with a person icon. Below the heading is a descriptive paragraph: 'An introduction to fundamental topics related to human genetics, including illustrations and basic explanations of genetics concepts.' To the right of this paragraph are three buttons: 'Printable PDF (7MB)', 'Open All', and 'Close All'. A list of eight topics is displayed as blue bars with white text and a right-pointing arrow:

- ▶ Cells and DNA
- ▶ Mutations and Health
- ▶ How Genes Work
- ▶ Gene Groups
- ▶ Inheriting Genetic Conditions
- ▶ Genetics and Human Traits
- ▶ Genetic Consultation
- ▶ Genetic Testing

NIH National Human Genome Research Institute- health information

The screenshot shows the NIH National Human Genome Research Institute homepage. At the top, there is a search bar with the text "Begin your search here" and a magnifying glass icon. To the right of the search bar are social media icons for Facebook, Twitter, and YouTube. Below the search bar is a navigation menu with the following items: "About Genomics", "Research Funding", "Research at NHGRI", "Health", "Careers & Training", "News & Events", and "About NHGRI". The main content area features a large banner for an "Open Session" titled "National Advisory Council for Human Genome Research". Below the banner, there is a button that says "Webcast starts Sept. 16". At the bottom of the page, there are three news release items: "Inaugural Genomic Innovator Awards", "A tea-party to launch a gene therapy trial", and "THE GENOMICS LANDSCAPE ASHG – A Vision for the Future".

The screenshot shows the NIH National Human Genome Research Institute website page for "Health". The navigation menu is the same as in the previous screenshot, but the "Health" link is circled in red. Below the navigation menu, there is a breadcrumb trail: "Home / Health / For Patients and Families". The main content area features a large banner with a photograph of a healthcare professional examining a baby. Below the banner, there is a section titled "For Patients and Families" with the text: "Genetic diseases impact not only patients but their whole families. These resources will help you understand genetic diseases and the importance of knowing your family health history."

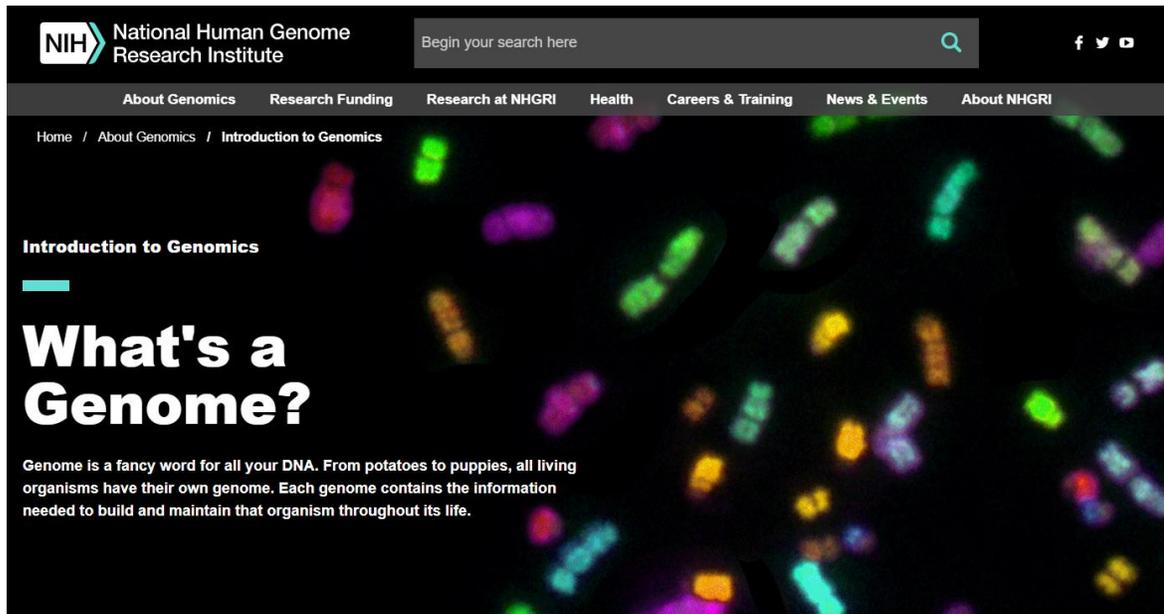
NHGRI

National Organization for Rare Disorders

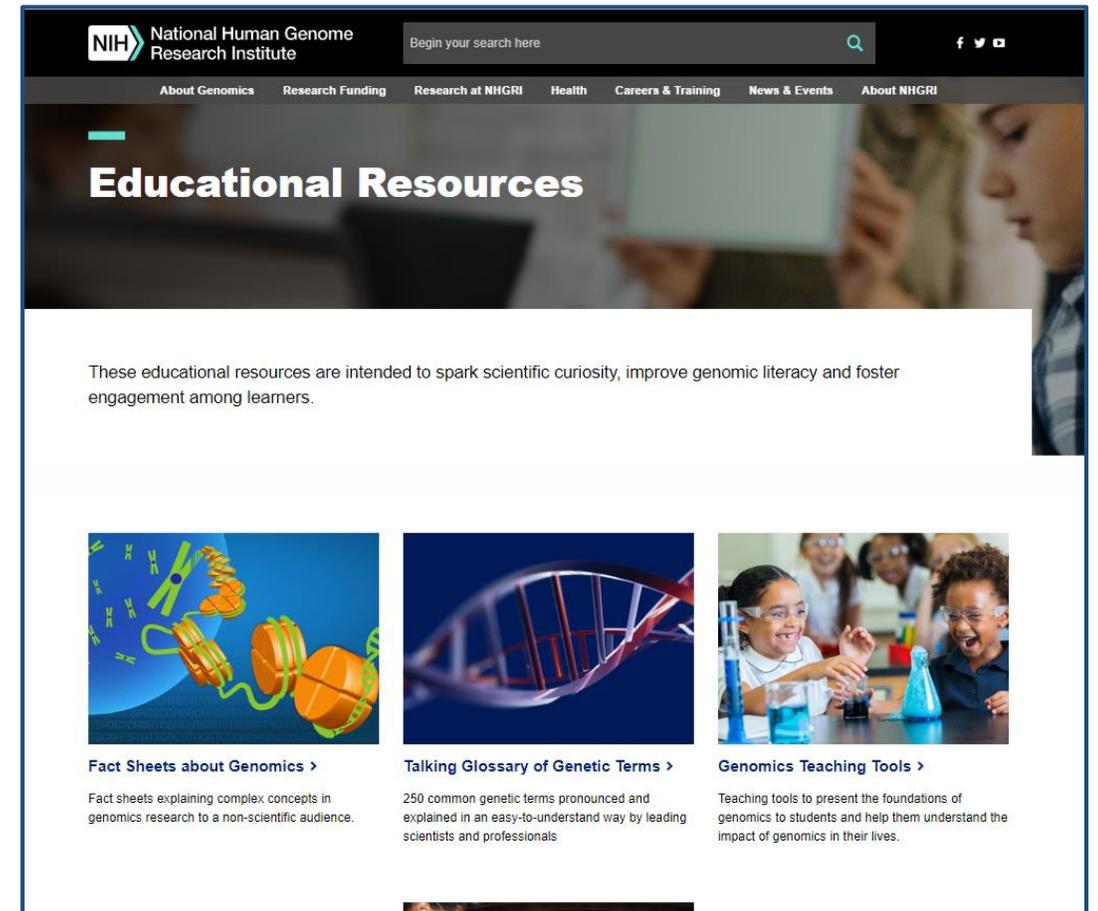
The screenshot shows the NORD website homepage. At the top, there is a navigation bar with links for ABOUT, NEWS, EVENTS, CONTACT, and a prominent DONATE button. The NORD logo and a 35th Anniversary banner are on the left. A search bar and social media icons (Facebook, LinkedIn, Twitter, YouTube, Blogger) are on the right. Below the navigation bar, there are six tabs: for PATIENTS AND FAMILIES, for PATIENT ORGANIZATIONS, for INDUSTRY, for CLINICIANS AND RESEARCHERS, ADVOCATE, and GET INVOLVED. The main banner features a collage of diverse people's faces with the text: "7,000 RARE DISEASES AFFECT 30 MILLION AMERICANS. HOW CAN YOU HELP? #DOYOURSHARE" and a button for "DoYourShare.com". Below the banner, a section titled "What's happening at NORD" contains four promotional cards: "RARE DISEASE DAY" with a colorful hand logo, "NORD 35th Anniversary presenting the RARE IMPACT AWARDS" for May 17, 2018 in Washington, D.C., with "Registration Open!", "Rare Summit" with a building icon, and "RUNNING FOR RARE" with a silhouette of a runner and the text "Accepting Applications". At the bottom of the page, there are links for "Tools and Resources" and "News".

NORD

NHGRI Education Resources



[Introduction to Genomics](#)



[Educational Resources](#)

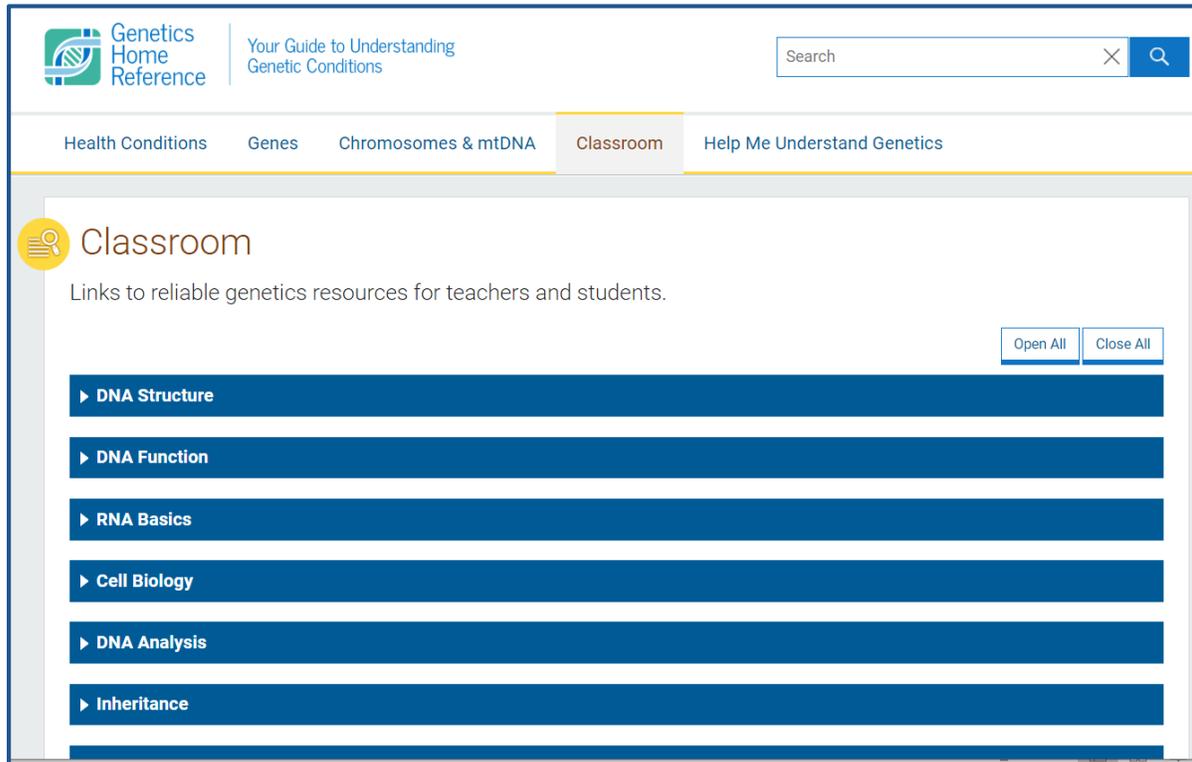
National DNA Day – April 25

The screenshot shows the top portion of the NIH website. At the top left is the NIH logo and the text "National Human Genome Research Institute". To the right is a search bar with the placeholder text "Begin your search here" and a magnifying glass icon. Further right are social media icons for Facebook, Twitter, and YouTube. Below this is a navigation menu with links: "About Genomics", "Research Funding", "Research at NHGRI", "Health", "Careers & Training", "News & Events", and "About NHGRI". Below the navigation is a breadcrumb trail: "Home / About Genomics / National DNA Day". The main hero image features a young girl with red-rimmed glasses smiling while holding a colorful DNA model. On the left side of the hero image is a "National DNA Day 25" logo. At the bottom of the hero image, the text "National DNA Day" is displayed in large white font.

[DNA Day](#)

This section of the website provides navigation and information about the DNA Day Starter Kit. It features a grid of four navigation links: "Explore DNA Day", "DNA Day Events >", "DNA Day Starter Kit >", and "DNA Day Activity Ideas >". Below these are two sub-links for "DNA Day Event Registration >" and "Celebrate DNA Day >". The "About DNA Day >" link is also present. Below the navigation is a "Starter Kit" section with a teal underline. The text reads: "The DNA Day Starter Kit contains a style guide, press release templates and other materials you need to plan the perfect classroom or community event. Download the entire kit or just the things you need to make your event a success." A blue "Get Started" button is located at the bottom of this section. To the right is a pink graphic showing various DNA Day 25 materials, including a "National DNA Day 25 Network Partner" badge, a "National DNA Day 25 Celebrating Genomics Through Awareness" banner, a "National DNA Day 25 Style Guide" document, and a name tag that says "Your Name" and "National DNA Day 25".

Genetics Home Reference- Classroom



Genetics Home Reference | Your Guide to Understanding Genetic Conditions

Search

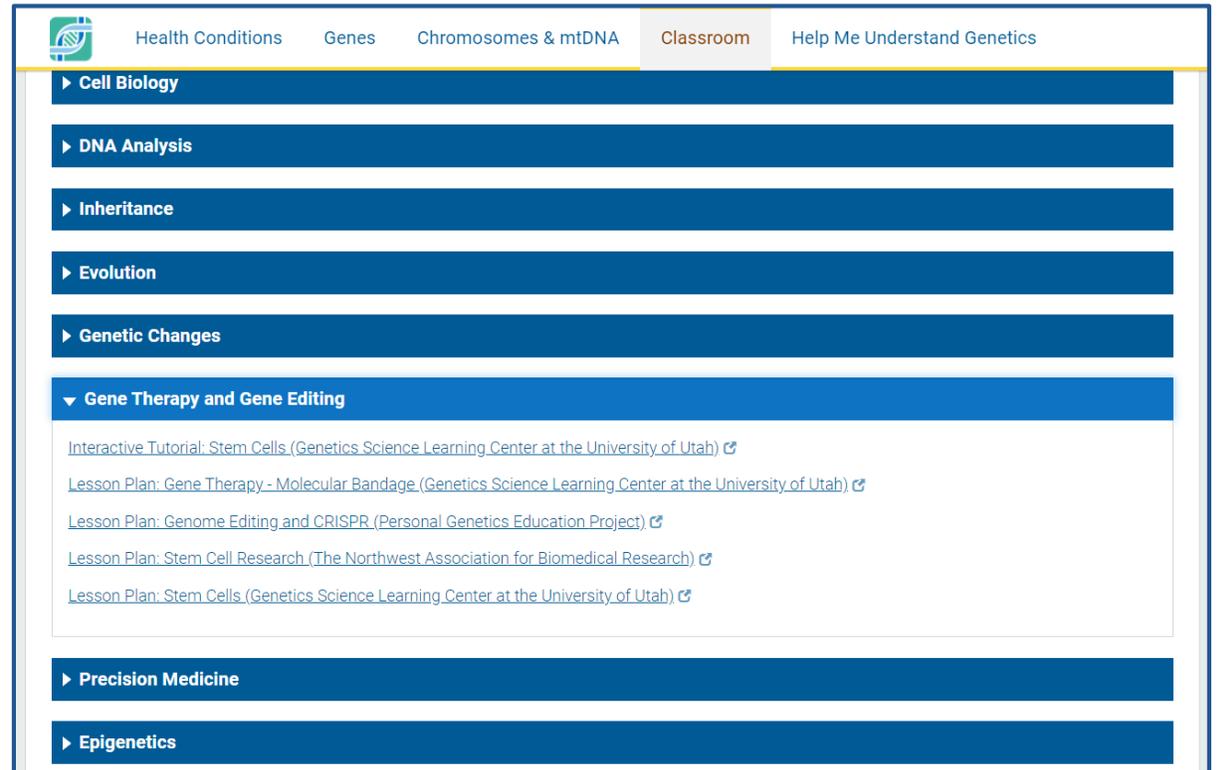
Health Conditions Genes Chromosomes & mtDNA **Classroom** Help Me Understand Genetics

Classroom

Links to reliable genetics resources for teachers and students.

Open All Close All

- ▶ DNA Structure
- ▶ DNA Function
- ▶ RNA Basics
- ▶ Cell Biology
- ▶ DNA Analysis
- ▶ Inheritance



Health Conditions Genes Chromosomes & mtDNA **Classroom** Help Me Understand Genetics

- ▶ Cell Biology
- ▶ DNA Analysis
- ▶ Inheritance
- ▶ Evolution
- ▶ Genetic Changes
- ▼ Gene Therapy and Gene Editing
 - [Interactive Tutorial: Stem Cells \(Genetics Science Learning Center at the University of Utah\)](#) ↗
 - [Lesson Plan: Gene Therapy - Molecular Bandage \(Genetics Science Learning Center at the University of Utah\)](#) ↗
 - [Lesson Plan: Genome Editing and CRISPR \(Personal Genetics Education Project\)](#) ↗
 - [Lesson Plan: Stem Cell Research \(The Northwest Association for Biomedical Research\)](#) ↗
 - [Lesson Plan: Stem Cells \(Genetics Science Learning Center at the University of Utah\)](#) ↗
- ▶ Precision Medicine
- ▶ Epigenetics

K-12 Resources

ASHG | Education

Overview

K-12

Trainee

Faculty

Genetics Professionals

General Public

Education

ASHG's primary educational objectives are to promote awareness of human genetics, encourage young people to enter genetics-related careers, and foster trust and support for genetics research. ASHG accomplishes those goals by educating students, health care professionals, health policy makers, the media, and the public about human genetics and its translation to health care practice. Learn more about ASHG's efforts in academic/professional training and public/consumer education.

K-12 Trainee Faculty Genetics Professionals General Public

Highlights

- 2019 DNA Day Activities and Essay Contest
- Human Genetics Scholars Initiative
- Genetics Engagement & Education Network
- Lesson Plan Database
- Undergraduate Workshop
- Trainee Paper Spotlight
- Six Things Everyone Should Know About Genetics

A Statement on the Importance of Participation of Scientists in K-12 Science Education

American Society of Human Genetics

Harry Potter's World

NIH U.S. National Library of Medicine

HISTORY of MEDICINE

HARRY POTTER'S WORLD
RENAISSANCE, SCIENCE, MAGIC, AND MEDICINE

In 1997, British author J. K. Rowling introduced the world to Harry Potter and a literary phenomenon was born. Although a fantasy story, the *Harry Potter* book series features magic that is based partially on Renaissance...

EXPLORE

LEARN more

SEE the digital gallery

BOOK the traveling exhibition

FIND OUT

My Family Health Portrait

The screenshot shows the homepage of the My Family Health Portrait tool. At the top, the title "My Family Health Portrait" is displayed in a blue, cursive font, with the subtitle "A tool from the Surgeon General" in a smaller, orange font below it. A blue "Get Help" button is located in the top right corner. The main content area features a list of bullet points under the heading "Using My Family Health Portrait you can:", detailing the tool's capabilities. Below this is a paragraph encouraging users to talk with their health care provider. A blue link "Learn more about My Family Health Portrait" is provided. Two orange buttons, "Create a Family Health History" and "Use a Saved History", are positioned at the bottom of the main content area. The background of the main content area is a large photograph of a diverse family of four. At the bottom of the screenshot, there is a horizontal strip of four smaller photographs showing various groups of people, including a family of three, a group of men, a man with a child, and a large group of people.

My Family Health Portrait
A tool from the Surgeon General

Get Help

Using My Family Health Portrait you can:

- Enter your family health history.
- Learn about your risk for conditions that can run in families.
- Print your family health history to share with family or your health care provider
- Save your family health history so you can update it over time.

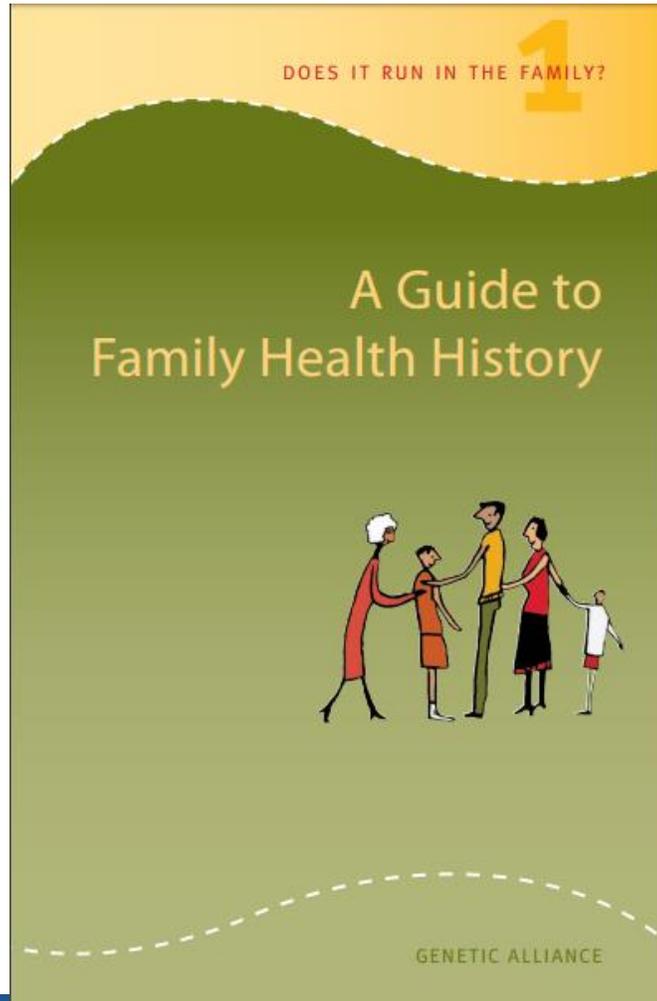
Talking with your health care provider about your family health history can help you stay healthy!

[Learn more about My Family Health Portrait](#)

Create a Family Health History Use a Saved History

[CDC Family Health Portrait](#)

Does It Run In the Family? Toolkit



[Does it Run In the Family? toolkit](#)



Literacy/Education Resources



Societal Issues

Privacy, Security, Ethical, and Societal Issues

Societal Concerns

- Who should have access to personal genetic information, and how will it be used?
- Who owns and controls genetic information?
- How does personal genetic information affect an individual and society's perceptions of that individual?
- How will genetic tests be evaluated and regulated for accuracy, reliability and utility?
- Where is the line between medical treatment and enhancement?
- Should testing be performed when no treatment is available?

GINA

The screenshot shows the GINA Help website. At the top, there is a dark blue header with the GINA logo (a stylized DNA helix) and the text "GENETIC INFORMATION NONDISCRIMINATION ACT". To the right of the header are links for "About" and "Contact". Below the header, there are four main content boxes. The first three boxes are on the left, each with a colored header and a question: "Genetic Information" (green header, "What is genetic information and why is it important?"), "GINA & Health Insurance" (orange header, "What are GINA's health insurance protections?"), and "GINA & Employment" (blue header, "What are GINA's employment protections?"). The fourth box is on the right, titled "What is GINA?", and contains a paragraph of text. Below these boxes is a light blue box with four links: "Have questions, comments or suggestions? Send us a note.", "Click here for a printer friendly version.", "For healthcare provider resources click here.", and "Click here for the GINA & You Information Sheet". At the bottom of the page, there is a permission statement and a design credit.

GINA GENETIC INFORMATION
NONDISCRIMINATION ACT [About](#) | [Contact](#)

Genetic Information
What is genetic information and why is it important?

GINA & Health Insurance
What are GINA's health insurance protections?

GINA & Employment
What are GINA's employment protections?

What is GINA?
The Genetic Information Nondiscrimination Act of 2008 (GINA) is a federal law that protects individuals from genetic discrimination in health insurance and employment. Genetic discrimination is the misuse of genetic information. This resource provides an introduction to GINA and its protections in health insurance and employment. It includes answers to common questions and examples to help you learn. Choose from one of the boxes to the left to begin!

✉ Have questions, comments or suggestions? [Send us a note.](#)
🖨 [Click here](#) for a printer friendly version.
✉ For healthcare provider resources [click here.](#)
🖨 [Click here](#) for the GINA & You Information Sheet

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:: DESIGN & DEVELOPMENT BY - WWW.PROJECTMISO.NET ::

[**GINA Help**](#)

H. R. 1313

CONGRESS.GOV

Legislation Congressional Record Committees Members Sign In

Quick Search Advanced Search Browse Search Tools Glossary Resources Help Contact

All Legislation [Examples: hr5, sres9, "health care"]

Home > Legislation > 115th Congress > H.R.1313

H.R.1313 - Preserving Employee Wellness Programs Act
115th Congress (2017-2018) | Get alerts

BILL Hide Overview

Sponsor: [Rep. Foxx, Virginia \[R-NC-5\]](#) (Introduced 03/02/2017)

Committees: House - Education and the Workforce; Energy and Commerce; Ways and Means

Committee Reports: [H. Rept. 115-459](#)

Latest Action: House - 12/11/2017 Placed on the Union Calendar, Calendar No. 341. ([All Actions](#))

Tracker:

Introduced > Passed House > Passed Senate > To President > Became Law

More on This Bill
[Constitutional Authority Statement](#)
[CBO Cost Estimates \[1\]](#)

Subject — Policy Area:
Health
[View subjects >](#)

Summary (1) Text (2) Actions (13) Titles (3) Amendments (0) Cosponsors (5) Committees (3) Related Bills (0)

Summary: H.R.1313 — 115th Congress (2017-2018) [All Information](#) (Except Text)

Listen to this page

There is one summary for H.R.1313. [Bill summaries](#) are authored by [CRS](#).

Shown Here:
Introduced in House (03/02/2017)

Preserving Employee Wellness Programs Act

This bill exempts workplace wellness programs from: (1) limitations under the Americans with Disabilities Act of 1990 on medical examinations and inquiries of employees, (2) the prohibition on collecting genetic information in connection with issuing health insurance, and (3) limitations under the Genetic Information Nondiscrimination Act of 2008 on collecting the genetic information of employees or family members of employees. This exemption applies to workplace wellness programs that comply with limits on rewards for employees participating in the program.

Workplace wellness programs may provide for more favorable treatment of individuals with adverse health factors, such as a disability.

Collection of information about a disease or disorder of a family member as part of a workplace wellness program is not an unlawful acquisition of genetic information about another family member.



The NEW ENGLAND JOURNAL of MEDICINE

Perspective
JULY 6, 2017

Undermining Genetic Privacy? Employee Wellness Programs and the Law

Kathy L. Hudson, Ph.D., and Karen Pollitz, M.P.P.

Genetic information is becoming ubiquitous in research and medicine. The cost of genetic analysis continues to fall, and its medical and personal value continues to grow.

The Genetic Information Nondiscrimination Act of 2008 (GINA) prohibits both employment and health insurance discrimination based on genetic information, and

PMID: 28537794

Usage by law enforcement

AMERICA

In Hunt For Golden State Killer, Investigators Uploaded His DNA To Genealogy Site

by LAUREL WAMSLEY

April 27, 2018 • After failing to find a match within criminal databases, law enforcement uploaded the killer's DNA profile to a no-frills website used to trace ancestry. The tactic has spurred privacy concerns.

Genetic Testing Scams

An official website of the United States government [Here's how you know](#)

U.S. Department of Health and Human Services
Office of Inspector General

Search Submit a Complaint

About OIG Reports Fraud Compliance Exclusions Newsroom Careers

Fraud
Child Support Enforcement
Consumer Alerts
Contract Fraud
Enforcement Actions
Fraud Risk Indicator
Grant Fraud
Medicaid Fraud Control Units
Medicare Fraud Strike Force
OIG Most Wanted Fugitives
Report Fraud
State False Claims Act Reviews
Whistleblower Ombudsman

Fraud Alert: Genetic Testing Scam



The U.S. Department of Health and Human Services Office of Inspector General is alerting the public about a fraud scheme involving genetic testing.

Genetic testing fraud occurs when Medicare is billed for a test or screening that was not medically necessary and/or was not ordered by a Medicare beneficiary's treating physician.

Scammers are offering Medicare beneficiaries "free" screenings or cheek swabs for genetic testing to obtain their Medicare information for identity theft or fraudulent billing purposes. Fraudsters are targeting beneficiaries through telemarketing calls,

Report the Scam

Online:
oig.hhs.gov/fraud/hotline

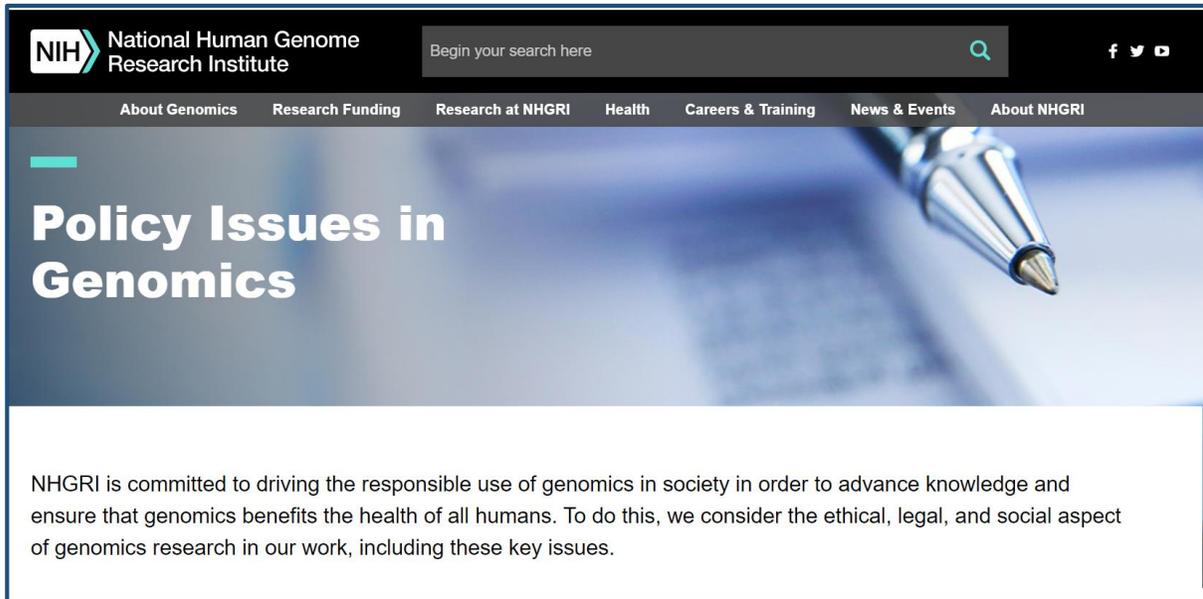
Phone: 1-800-447-8477
(1-800-HHS-TIPS)

TTY: 1-800-377-4950

f t e in + 3.9K

[Office of Inspector General, Fraud Alert](#)

NIH National Human Genome Research Institute



- Genetic discrimination
- Health disparities
- What is genome editing
- Intellectual property
- Privacy
- Regulation of genetic tests

[NHGRI Policy Issues](#)

Informing the Public



CENTER FOR
GENETICS AND
SOCIETY



U.S. National Library of Medicine

National Network of Libraries of Medicine
Pacific Northwest Region

Precision Medicine

All of Us Research Program

Precision Medicine



THE PRECISION MEDICINE INITIATIVE



PRECISION MEDICINE INITIATIVE PRINCIPLES STORIES

f t GO TO TOP

- Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person.
- Instead of what treatment is right for this disease it is what treatment is right for the patient.

[Precision Medicine Initiative announcement](#)

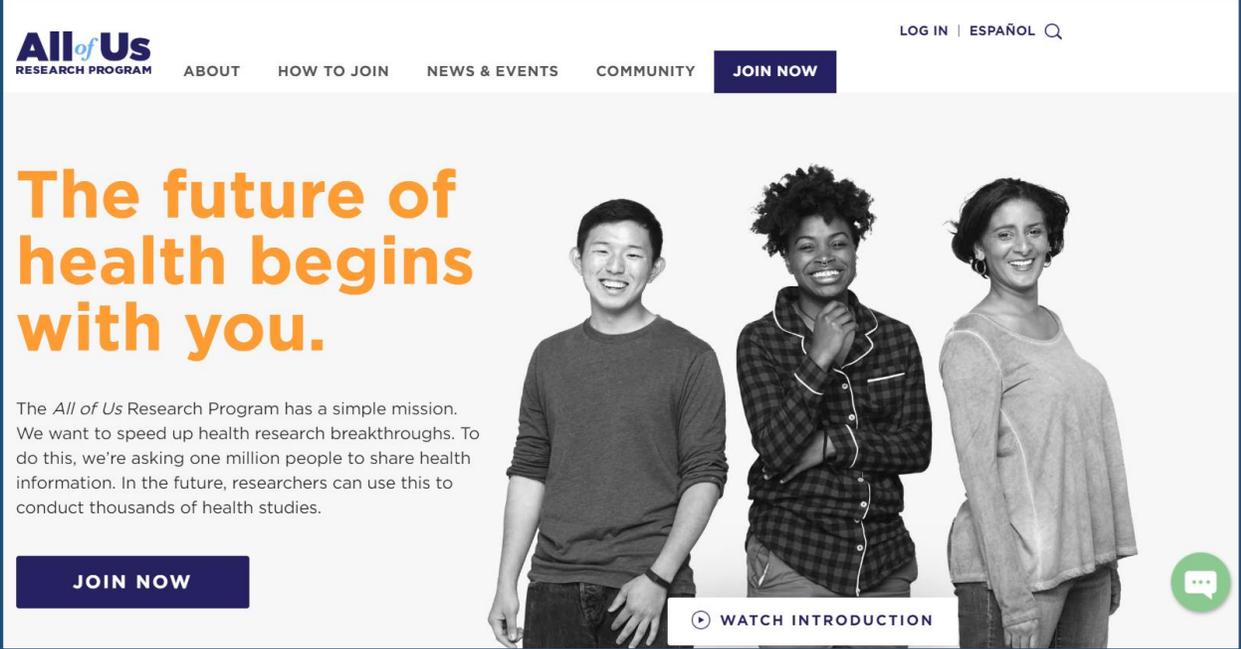


U.S. National Library of Medicine

National Network of Libraries of Medicine
Pacific Northwest Region

All of Us Research Program

The mission of the *All of Us* Research Program is to accelerate health research and medical breakthroughs, enabling individualized prevention, treatment, and care for all of us.



The screenshot shows the homepage of the All of Us Research Program. At the top left is the logo "All of Us RESEARCH PROGRAM". To its right are navigation links: "ABOUT", "HOW TO JOIN", "NEWS & EVENTS", "COMMUNITY", and a prominent "JOIN NOW" button. In the top right corner, there are links for "LOG IN" and "ESPAÑOL" with a search icon. The main content area features the headline "The future of health begins with you." in orange and black text. Below the headline is a paragraph: "The All of Us Research Program has a simple mission. We want to speed up health research breakthroughs. To do this, we're asking one million people to share health information. In the future, researchers can use this to conduct thousands of health studies." To the right of the text is a photograph of three diverse people (a young man, a woman, and another woman) smiling. Below the text is a "JOIN NOW" button and a "WATCH INTRODUCTION" button with a play icon. A green chat bubble icon is in the bottom right corner.

[All of Us Research Program](#)

All of Us Research Program- video



[What is All of Us? video](#)

Library role

“Preparing the public to make educated personal and family health decisions in a time of rapidly evolving genetic and genomic knowledge will require new partnerships between the education system, health care systems, the government, community advocacy organizations, consumers and the media.”

[“What Does it Mean to be Genomically Literate? National Human Genome Research Institute Meeting Report”](#)

Libraries and genetic health information

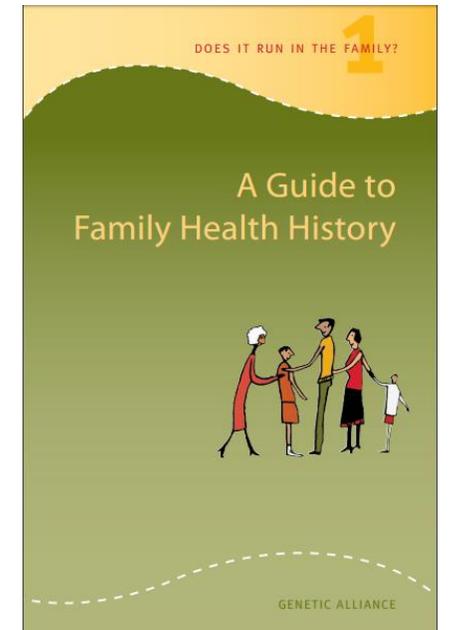
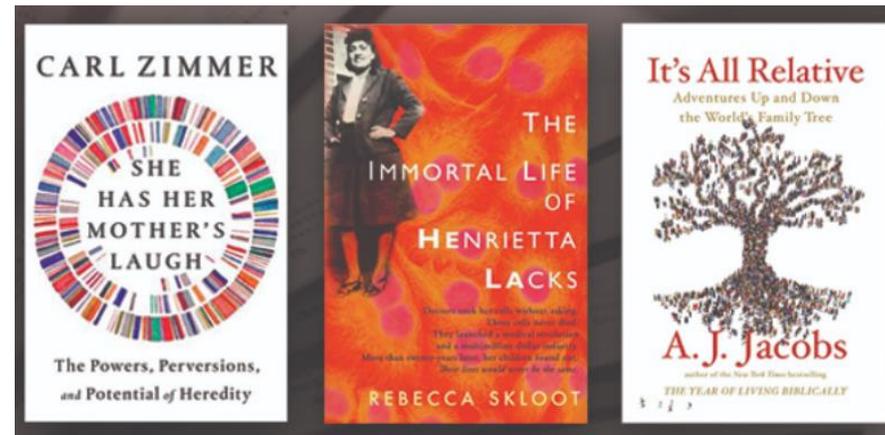
BECAUSE YOUR DNA DOESN'T HAVE TO BE YOUR DESTINY.

LIBRARIES TRANSFORM
At American Library Association

NIH U.S. National Library of Medicine
National Network of Libraries of Medicine



Your Guide to Understanding Genetic Conditions



Show What You Know!

1. The CDC's top 10 causes of death all have a genetic component.
True or False?
2. The American College of Medical Genetics and Genomics (ACMG) recommends everyone should use a direct to consumer genetic test.
True or False?
3. What is the name of the research program that is looking to collect data on 1 million volunteers in order to provide more precise health care through prevention and treatment?
4. GINA (Genetic Information Nondiscrimination Act) protects you from life insurance discrimination.
True or False?
5. What resource would you recommend to patrons who wanted to learn more about genetic testing?

Thank You!

Carolyn Martin, MLS, AHIP
NNLM PNR
martinc4@uw.edu

