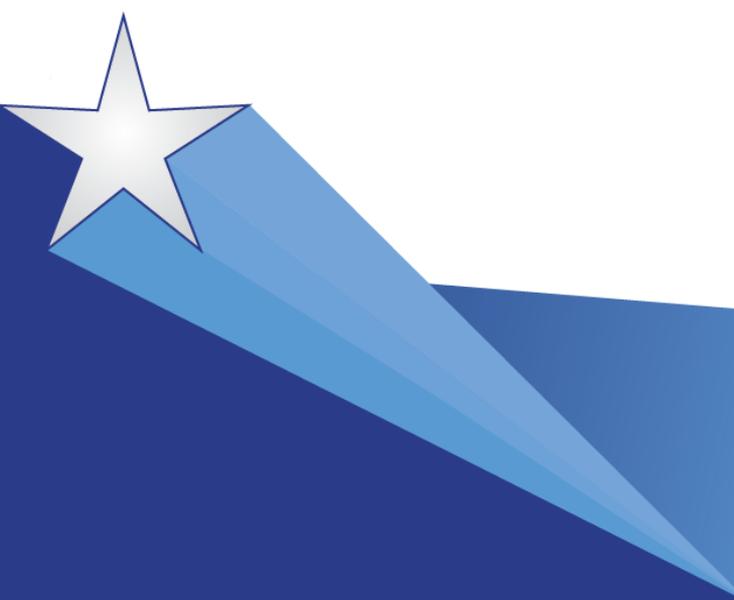


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# ABCs of DNA:

## Unraveling the Mysteries of Genetics Information for Consumers



Carolyn Martin, MLS, AHIP  
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Pacific Northwest Region (NNLM PNR)  
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# Agenda

- Basic genetics overview
- Genomic health literacy & genomic science literacy
- Genetic testing including direct-to-consumer
- Genetic consumer health resources
- Ethics and privacy
- Precision Medicine Initiative

[Presentation resources](https://nnlm.gov/pnr/guides/training-resources-you-can-use/presentations)

<https://nnlm.gov/pnr/guides/training-resources-you-can-use/presentations>

# Who We Are

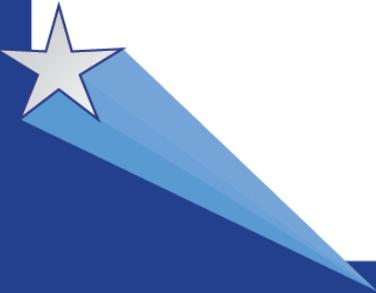


**NIH**

**NLM**

**NNLM**

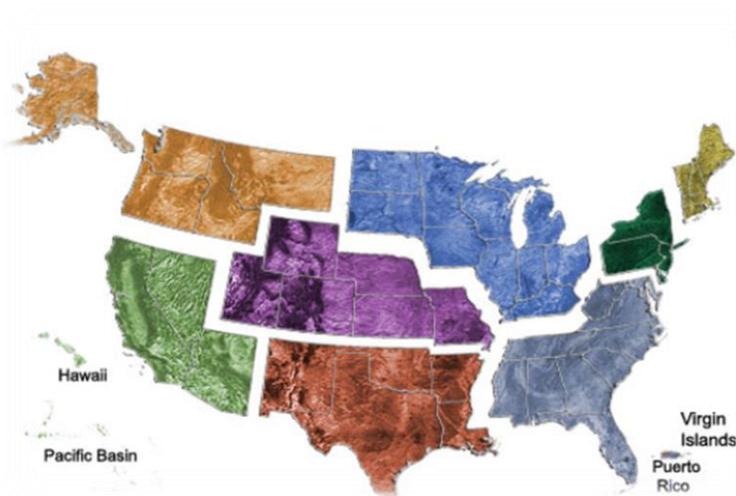
What's the difference?





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 PNR](https://nnlm.gov/pnr)

<https://nnlm.gov/pnr>

# News Headlines

## *Human Gene Editing Receives Science Panel's Support*

Scientists Say They Hope To Create A Human Genome In The Lab

Humans will be 'irrevocably altered' by genetic editing, warn scientists ahead of summit

An open letter from 177 scientists, campaigners and health experts is calling for a moratorium on gene editing ahead of a summit in Washington

Genes Plus Erratic Sleep May Raise Odds for Obesity

How much shuteye you get could tip the scale if you're predisposed to gain weight, researchers say

**The Genetic Tool That Will Modify Humanity**

Crispr allows scientists to control the human genome better than ever before.

**Clinical Genetics Has a Big Problem That's Affecting People's Lives**

Unreliable research can lead families to make health decisions they might regret.

**Genetic Privacy, as Explained by Mystery Poopers**

In Big Shift, 23andMe Will Invent Drugs Using Customer Data

Your DNA May Determine How You Handle the Time Change

**"Improving" Humans with Customized Genes Sparks Debate among Scientists**

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

**Genetically Modified Humans? How Genome Editing Works**

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

# Consumer Genomic Health Literacy

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



# Leading causes of death

1. Heart disease: 614,348
2. Cancer: 591,699
3. Chronic lower respiratory diseases: 147,101
4. Accidents (unintentional injuries): 136,053
5. Stroke (cerebrovascular diseases): 133,103
6. Alzheimer's disease: 93,541
7. Diabetes: 76,488
8. Influenza and pneumonia: 55,227
9. Nephritis, nephrotic syndrome, and nephrosis: 48,146
10. Intentional self-harm (suicide): 42,773

# The Story of You

[The Story of You](https://www.youtube.com/watch?v=TwXXgEz9o4w)

<https://www.youtube.com/watch?v=TwXXgEz9o4w>



# CATEGORIES OF DISEASES ATTRIBUTED TO GENES

- Chromosomal Diseases
- Monogenic Diseases
- Multifactorial Diseases



---

# Genetic Testing

including Direct-to-Consumer



# Clinical Uses of Genetic Tests

**GENETIC TESTING**  
NHGRI FACT SHEETS  
genome.gov

**Genetic Tests Can Help to:**

-  **Diagnose Your Disease**
-  **Pinpoint Genetic Factors That Caused Your Disease**
-  **Predict How Severe Your Disease Might Be**
-  **Choose the Best Medicine and Correct Dose**
-  **Discover Genetic Factors That Increase Your Disease Risk**
-  **Find Genetic Factors That Could Be Passed to Your Children**
-  **Screen Newborns for Certain Treatable Conditions**

**NIH** National Human Genome Research Institute

[Genetic Testing image from Genome.gov](https://www.genome.gov)

[https://www.genome.gov/images/content/genetic\\_testing.jpg](https://www.genome.gov/images/content/genetic_testing.jpg)

# 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 testing** to investigate ancestry

**Age 65: pharmacogenomics testing** when Plavix (anti-platelet drug) was not effective



# Genetic Testing Results

What genes and what variants did you test for?

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

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

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

Possibility of false positive and false negative results.



# Genetic Testing- is it necessary?

The screenshot shows the Consumer Reports Health Choices website. The main navigation bar includes links for Home, About, Partnerships, Campaigns, Catalog, In Depth, Connect, News and notes, and Patients' stories. The article page is titled 'Making Smart Decisions About Genetic Testing (ACMG)'. It includes metadata such as Campaign Series (Choosing Wisely), Medical Category (Tests), Article Type (Advice), Language (Plain English, Spanish), Affiliation (American College of Medical Genetics and Genomics), Format (HTML, PDF), and Most recent update (10/07/2015). A thumbnail image of the article is displayed. Below the article text, there is a 'Files to download' section with links for Plain English, Spanish, and Webpage file versions. A search bar is visible at the top right of the page.

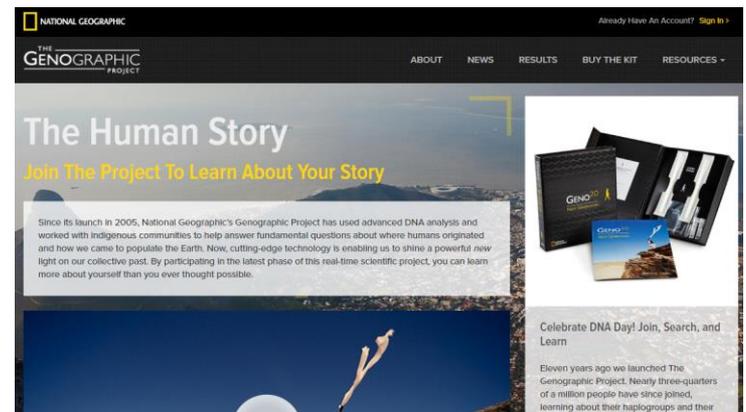
## [Choosing Wisely](http://consumerhealthchoices.org/catalog/making-smart-decisions-about-genetic-testing-acmg/)

<http://consumerhealthchoices.org/catalog/making-smart-decisions-about-genetic-testing-acmg/>

## Questions to ask:

- Am I in the group at risk and should I get tested?
- If I decide to get tested, what do the results mean?
- What are my treatment options based on results?
- How do I decide on treatment?

# Direct to Consumer Testing



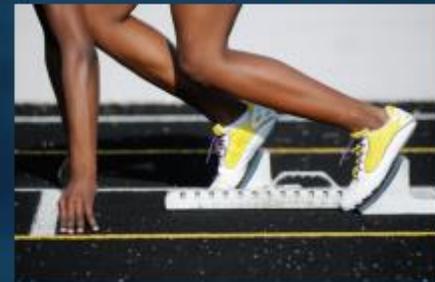
# Genomic Testing- Athletic Ability

- Over 36 companies marketing genetic tests
- Endurance and power
- Poor quality control
- Targeted to coaches and parents
- Individuals also wanting to focus training

**Achieve**  
your full potential.



**Increase**  
your athletic performance.



**Harness**  
your natural ability through  
personalized genomics.



[www.GenomicExpress.com](http://www.GenomicExpress.com)

# Concerns

- Privacy
- Legality
- Who has access?
- How useful now?
- 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
- Who can get the testing?

# Benefits

- Learn more about own health
- More effective medical treatments
- 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

# Genetics Home Reference

NIH U.S. NATIONAL LIBRARY OF MEDICINE



Your Guide to Understanding  
Genetic Conditions

Search

Health Conditions

Genes

Chromosomes & mtDNA

Resources

Help Me Understand Genetics

Home

Help Me Understand Genetics

Genetic Testing

What is direct-to-consumer genetic testing?

## What is direct-to-consumer genetic testing?

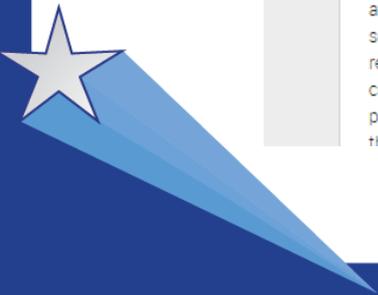
Traditionally, genetic tests have been available only through healthcare providers such as physicians, nurse practitioners, and genetic counselors. Healthcare providers order the appropriate test from a laboratory, collect and send the samples, and interpret the test results. Direct-to-consumer genetic testing refers to genetic tests that are marketed directly to consumers via television, print advertisements, or the Internet. This form of testing, which is also known as at-home genetic testing, provides access to a person's genetic information without necessarily involving a doctor or insurance company in the process.

If a consumer chooses to purchase a genetic test directly, the test kit is mailed to the consumer instead of being ordered through a doctor's office. The test typically involves collecting a DNA sample at home, often by swabbing the inside of the cheek, and mailing the sample back to the laboratory. In some cases, the person must visit a health clinic to have blood drawn. Consumers are notified of their results by mail or over the telephone, or the results are posted online. In some cases, a genetic counselor or other healthcare provider is available to explain the results and answer questions. The price for this type of at-home genetic testing ranges from several hundred dollars to more than a thousand dollars.

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

The American College of Medical Genetics, which is a national association of doctors specializing in genetics, has issued [a statement on direct-to-consumer genetic testing](#).

The American Society of Human Genetics, a professional membership organization for specialists in genetics, has also issued [a statement on direct-to-consumer genetic testing in the United](#)



# MedlinePlus

The screenshot shows the MedlinePlus website interface. At the top, it says "U.S. National Library of Medicine" and "MedlinePlus Trusted Health Information for You". There is a search bar and navigation links like "About MedlinePlus", "Site Map", "FAQs", and "Contact Us". The main navigation includes "Health Topics", "Drugs & Supplements", "Videos & Tools", and "Español". The breadcrumb trail is "Home -> Health Topics -> Genetic Testing".

## Genetic Testing

On this page

<b>Basics</b> <ul style="list-style-type: none"><li>Summary</li><li>Start Here</li><li>Latest News</li></ul>	<b>Learn More</b> <ul style="list-style-type: none"><li>Related Issues</li><li>Specifics</li></ul>	<b>See, Play and Learn</b> <ul style="list-style-type: none"><li>No links available</li></ul>
<b>Research</b> <ul style="list-style-type: none"><li>Statistics and Research</li><li>Clinical Trials</li><li>Journal Articles</li></ul>	<b>Resources</b> <ul style="list-style-type: none"><li>Reference Desk</li><li>Find an Expert</li></ul>	<b>For You</b> <ul style="list-style-type: none"><li>Patient Handouts</li></ul>

### 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

The screenshot shows the Federal Trade Commission (FTC) website's "CONSUMER INFORMATION" section. It features a search bar and a language option for "ESPAÑOL". The main heading is "Direct-to-Consumer Genetic Tests".

## 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.

### Related Items

- Dietary Supplements
- Miracle Health Claims
- Cancer Treatment Scams

Navigation menu: MONEY & CREDIT, HOMES & MORTGAGES, HEALTH & FITNESS, Healthy Living, Treatments & Cures, Weight Loss & Fitness, JOBS & MAKING MONEY, PRIVACY, IDENTITY & ONLINE SECURITY, BLOG, VIDEO & MEDIA.

Bottom navigation: Genes and Genetic Tests, Interpreting the Results, Company Claims, If You're Considering a DTC Genetic Test, For More Information.

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# Consumer Resources



# 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 Encyclopedia" (with a book icon), and "Medical Dictionary from Merriam-Webster" (with a search bar). There are also sections for "Today's Health News" (with a headline about college students), "Stay Connected" (with an email sign-up form), "NIH MedlinePlus Magazine" (with a magazine cover), and "Clinical Trials" (with a search bar). The footer contains icons for "Easy-to-Read Materials", "Organizations and Directories", "Health Information in Multiple Languages", and "MedlinePlus Connect for EHRs", along with social media links and subscription options.

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

[MedlinePlus](https://medlineplus.gov/) <https://medlineplus.gov/>

# MedlinePlus

Home → Health Topics

## Health Topics



Read about symptoms, causes, treatment and prevention for over 1000 diseases, illnesses, health conditions and wellness issues. MedlinePlus health topics are regularly reviewed, and links are updated daily.

### Find topics A-Z

[A](#) [B](#) [C](#) [D](#) [E](#) [F](#) [G](#) [H](#) [I](#) [J](#) [K](#) [L](#) [M](#) [N](#) [O](#) [P](#) [Q](#) [R](#) [S](#) [T](#) [U](#) [V](#) [W](#) [XYZ](#) [List of All Topics](#)

#### Body Location/Systems

- [Blood, Heart and Circulation](#)
- [Bones, Joints and Muscles](#)
- [Brain and Nerves](#)
- [Digestive System](#)
- [Ear, Nose and Throat](#)
- [Endocrine System](#)
- [Eyes and Vision](#)
- [Immune System](#)
- [Kidneys and Urinary System](#)
- [Lungs and Breathing](#)
- [Mouth and Teeth](#)
- [Skin, Hair and Nails](#)
- [Female Reproductive System](#)
- [Male Reproductive System](#)

#### Disorders and Conditions

- [Cancers](#)
- [Diabetes Mellitus](#)
- [Genetics/Birth Defects](#)
- [Infections](#)
- [Injuries and Wounds](#)
- [Mental Health and Behavior](#)
- [Metabolic Problems](#)
- [Poisoning, Toxicology, Environmental Health](#)
- [Pregnancy and Reproduction](#)
- [Substance Abuse Problems](#)

#### Diagnosis and Therapy

- [Complementary and Alternative Therapies](#)
- [Diagnostic Tests](#)
- [Drug Therapy](#)
- [Surgery and Rehabilitation](#)
- [Symptoms](#)
- [Transplantation and Donation](#)

#### Demographic Groups

- [Children and Teenagers](#)
- [Men](#)
- [Population Groups](#)
- [Seniors](#)
- [Women](#)

#### Health and Wellness

- [Disasters](#)
- [Fitness and Exercise](#)
- [Food and Nutrition](#)
- [Health System](#)
- [Personal Health Issues](#)
- [Safety Issues](#)
- [Sexual Health Issues](#)
- [Social/Family Issues](#)
- [Wellness and Lifestyle](#)

# MedlinePlus

U.S. National Library of Medicine

**MedlinePlus**  
Trusted Health Information for You

Search MedlinePlus

About MedlinePlus Site Map FAQs Contact Us

Health Topics Drugs & Supplements Videos & Tools Español

Home → Health Topics → Genetic Disorders

## Genetic Disorders

On this page

Basics	Learn More	See, Play and Learn
<ul style="list-style-type: none"> <li>Summary</li> <li>Start Here</li> <li>Diagnosis and Tests</li> <li>Treatments and Therapies</li> </ul>	<ul style="list-style-type: none"> <li>Living With</li> <li>Related Issues</li> <li>Specifics</li> <li>Genetics</li> </ul>	<ul style="list-style-type: none"> <li>No links available</li> </ul>

Research	Resources	For You
<ul style="list-style-type: none"> <li>Statistics and Research</li> <li>Clinical Trials</li> <li>Journal Articles</li> </ul>	<ul style="list-style-type: none"> <li>Reference Desk</li> <li>Find an Expert</li> </ul>	<ul style="list-style-type: none"> <li>Children</li> <li>Teenagers</li> <li>Women</li> <li>Patient Handouts</li> </ul>

**Summary**

Genes are the building blocks of heredity. They are passed from parent to child. They hold DNA, the instructions for making proteins. Proteins do most of the work in cells. They move molecules from one place to another, build structures, break down toxins, and do many other maintenance jobs.

Sometimes there is a mutation, a change in a gene or genes. The mutation changes the gene's instructions for making a protein, so the protein does not work properly or is missing entirely. This can cause a medical condition called a genetic disorder.

You can inherit a gene mutation from one or both parents. A mutation can also happen during your lifetime.

There are three types of genetic disorders:

Single gene disorders, where a mutation affects one gene. Cystic fibrosis is an example.





#ADAM

**Get Genetic Disorders updates by email**

Enter email address

**MEDICAL ENCYCLOPEDIA**

- Alström syndrome
- Basal cell nevus syndrome
- Beriberi
- Cystinuria
- Genetics
- Hemochromatosis

### Diagnosis and Tests

- How Are Genetic Conditions Diagnosed? [NH](#) (National Library of Medicine)
- MedlinePlus: Newborn Screening [NH](#) (National Library of Medicine)  
Available in Spanish
- Progeria Research Foundation Diagnostic Testing Program (Progeria Research Foundation, Inc.)

### Treatments and Therapies

- How Are Genetic Conditions Treated or Managed? [NH](#) (National Library of Medicine)

### Living With

- Physical Therapy and Occupational Therapy in Progeria (Progeria Research Foundation, Inc.) - PDF

### Related Issues

- Learning about an Undiagnosed Condition in an Adult [NH](#) (National Human Genome Research Institute)

### Specifics

- Bloom's Syndrome (Chicago Center for Jewish Genetic Disorders)
- Chromosomal Conditions (March of Dimes Birth Defects Foundation)  
Available in Spanish
- Cryopyrin-Associated Autoinflammatory Syndromes (CAPS) - Juvenile (American College of Rheumatology)
- Genetics Home Reference [NH](#) (National Library of Medicine)
- Learning about Poland Anomaly [NH](#) (National Human Genome Research Institute)
- Learning about Progeria [NH](#) (National Human Genome Research Institute)
- Noonan Syndrome (Mayo Foundation for Medical Education and Research)
- Progeria 101/FAQ (Progeria Research Foundation, Inc.)
- Specific Genetic Disorders [NH](#) (National Human Genome Research Institute)
- Triple X Syndrome (Mayo Foundation for Medical Education and Research)
- Williams Syndrome [NH](#) (National Institute of Neurological Disorders and Stroke)

### Genetics

- Genetics Home Reference: Genetic Conditions [NH](#) (National Library of Medicine)

### Statistics and Research

### Cystic Fibrosis

- Down Syndrome
- Dwarfism
- Fragile X Syndrome
- Genetic Brain Disorders
- Genetic Counseling
- Genetic Testing
- Hemochromatosis
- Leukodystrophies
- Osteogenesis Imperfecta
- Prader-Willi Syndrome
- Rare Diseases
- Sickle Cell Anemia

### National Institutes of Health

The primary NIH organization for research on Genetic Disorders is the National Institute of Child Health and Human Development

### NIH MedlinePlus Magazine

- Medical Mysteries: "Thankful They Found a Cause"
- Medical Mysteries: "We Feel Deep Compassion for Patients..."
- Medical Mysteries: NIH Clinical Center: There's No Other Hospital Like It
- Medical Mysteries: NIH Undiagnosed Diseases Program
- Promise and Payoff of Rare Diseases Research

# Genetics Home Reference

NIH U.S. NATIONAL LIBRARY OF MEDICINE

Genetics Home Reference  
Your Guide to Understanding Genetic Conditions

Search

Health Conditions Genes Chromosomes & mtDNA Resources Help Me Understand Genetics

Genetics Home Reference provides consumer-friendly information about the effects of genetic variation on human health.

**Health Conditions**  
More than 1,100 health conditions, diseases, and syndromes  
Browse A-Z

**New & Updated**

**New**

- Hartnup disease
- multiple myeloma
- SYNGAP1-related intellectual disability
- Past 12 Months

**Updated**

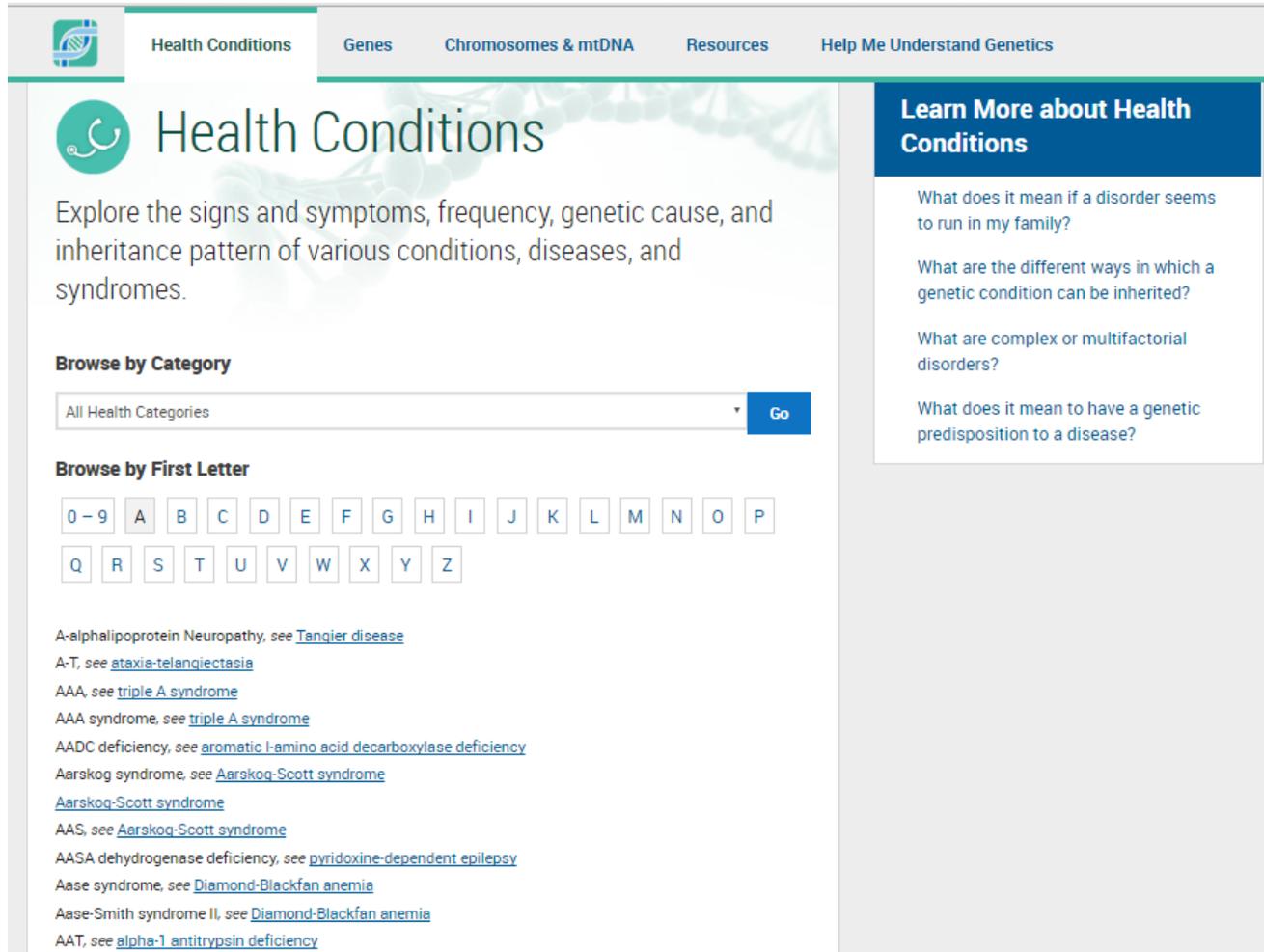
- Hutchinson-Gilford progeria syndrome
- otospondylomeqasepiphyseal dysplasia

**Genes**

- Health conditions
- Genes
- Chromosomes and DNA
- Resources
- Genetic handbook

[Genetics Home Reference](https://ghr.nlm.nih.gov/) <https://ghr.nlm.nih.gov/>

# Genetics Home Reference



The screenshot shows the Genetics Home Reference website. At the top, there is a navigation bar with a logo on the left and several menu items: "Health Conditions", "Genes", "Chromosomes & mtDNA", "Resources", and "Help Me Understand Genetics". Below the navigation bar, the "Health Conditions" section is highlighted. It features a circular icon with a DNA helix and the text "Health Conditions". Below this, there is a paragraph explaining the purpose of the section: "Explore the signs and symptoms, frequency, genetic cause, and inheritance pattern of various conditions, diseases, and syndromes." To the right of this section, there is a blue box titled "Learn More about Health Conditions" containing four 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?", "What are complex or multifactorial disorders?", and "What does it mean to have a genetic predisposition to a disease?". Below the introductory text, there are two browsing options: "Browse by Category" with a dropdown menu set to "All Health Categories" and a "Go" button, and "Browse by First Letter" with a grid of buttons for letters A through Z and numbers 0-9. At the bottom, there is a list of links for various conditions, such as "A-alpha1ipoprotein Neuropathy, see [Tanquer disease](#)" and "A-T, see [ataxia-telangiectasia](#)".

**Health Conditions**

Explore the signs and symptoms, frequency, genetic cause, and inheritance pattern of various conditions, diseases, and syndromes.

**Browse by Category**

All Health Categories

**Browse by First Letter**

0-9 A B C D E F G H I J K L M N O P  
Q R S T U V W X Y Z

A-alpha1ipoprotein Neuropathy, see [Tanquer disease](#)  
A-T, see [ataxia-telangiectasia](#)  
AAA, see [triple A syndrome](#)  
AAA syndrome, see [triple A syndrome](#)  
AADC deficiency, see [aromatic l-amino acid decarboxylase deficiency](#)  
Aarskog syndrome, see [Aarskog-Scott syndrome](#)  
[Aarskog-Scott syndrome](#)  
AAS, see [Aarskog-Scott syndrome](#)  
AASA dehydrogenase deficiency, see [pyridoxine-dependent epilepsy](#)  
Aase syndrome, see [Diamond-Blackfan anemia](#)  
Aase-Smith syndrome II, see [Diamond-Blackfan anemia](#)  
AAT, see [alpha-1 antitrypsin deficiency](#)

**Learn More about Health Conditions**

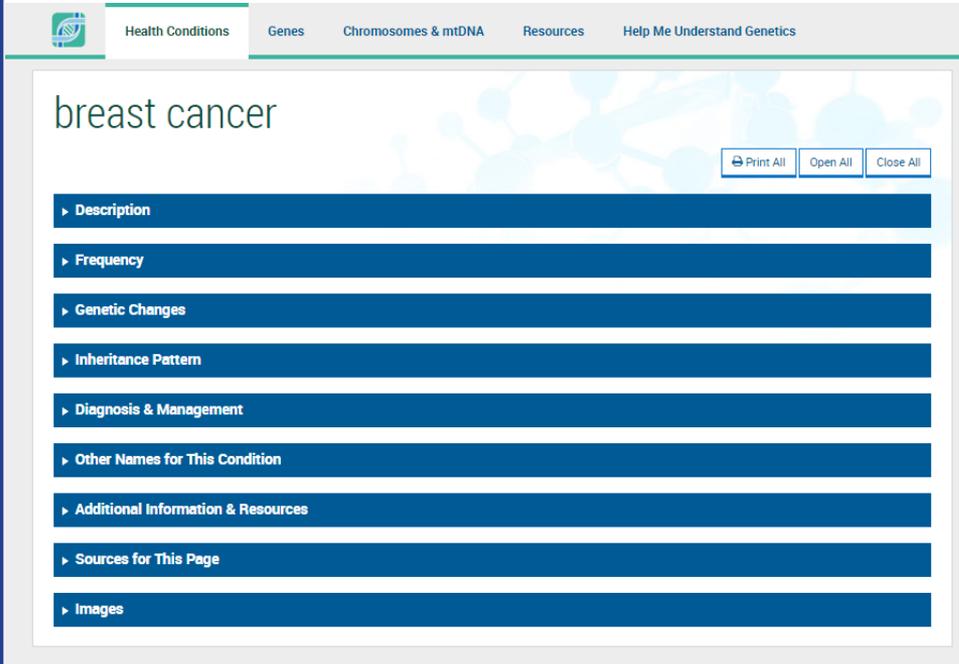
What does it mean if a disorder seems to run in my family?

What are the different ways in which a genetic condition can be inherited?

What are complex or multifactorial disorders?

What does it mean to have a genetic predisposition to a disease?

# Genetics Home Reference

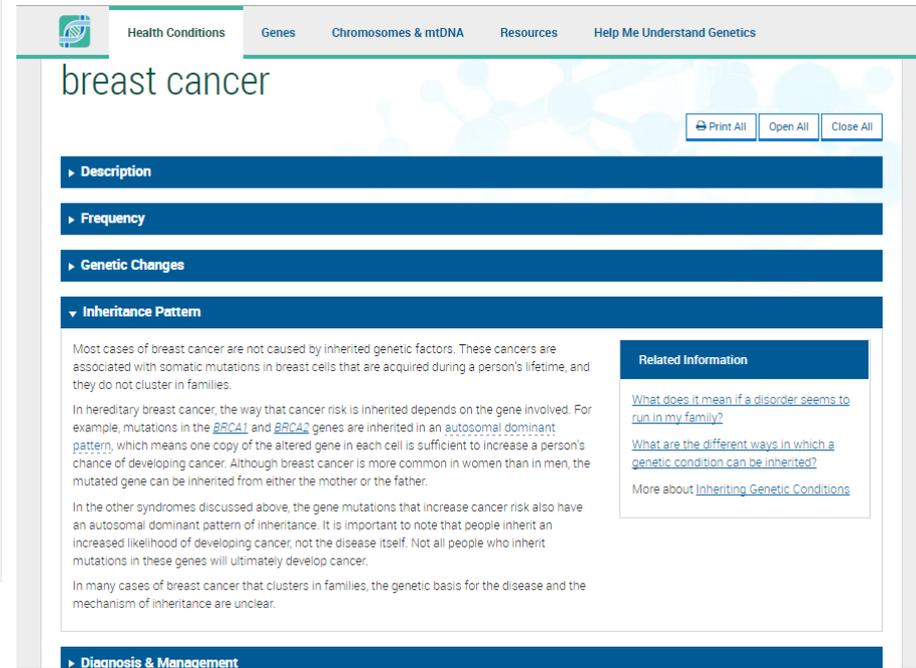


Health Conditions Genes Chromosomes & mtDNA Resources Help Me Understand Genetics

## breast cancer

Print All Open All Close All

- ▶ Description
- ▶ Frequency
- ▶ Genetic Changes
- ▶ Inheritance Pattern
- ▶ Diagnosis & Management
- ▶ Other Names for This Condition
- ▶ Additional Information & Resources
- ▶ Sources for This Page
- ▶ Images



Health Conditions Genes Chromosomes & mtDNA Resources Help Me Understand Genetics

## breast cancer

Print All Open All Close All

- ▶ Description
- ▶ Frequency
- ▶ Genetic Changes
- ▼ Inheritance Pattern

Most cases of breast cancer are not caused by inherited genetic factors. These cancers are associated with somatic mutations in breast cells that are acquired during a person's lifetime, and they do not cluster in families.

In hereditary breast cancer, the way that cancer risk is inherited depends on the gene involved. For example, mutations in the [BRCA1](#) and [BRCA2](#) genes are inherited in an [autosomal dominant pattern](#), which means one copy of the altered gene in each cell is sufficient to increase a person's chance of developing cancer. Although breast cancer is more common in women than in men, the mutated gene can be inherited from either the mother or the father.

In the other syndromes discussed above, the gene mutations that increase cancer risk also have an autosomal dominant pattern of inheritance. It is important to note that people inherit an increased likelihood of developing cancer, not the disease itself. Not all people who inherit mutations in these genes will ultimately develop cancer.

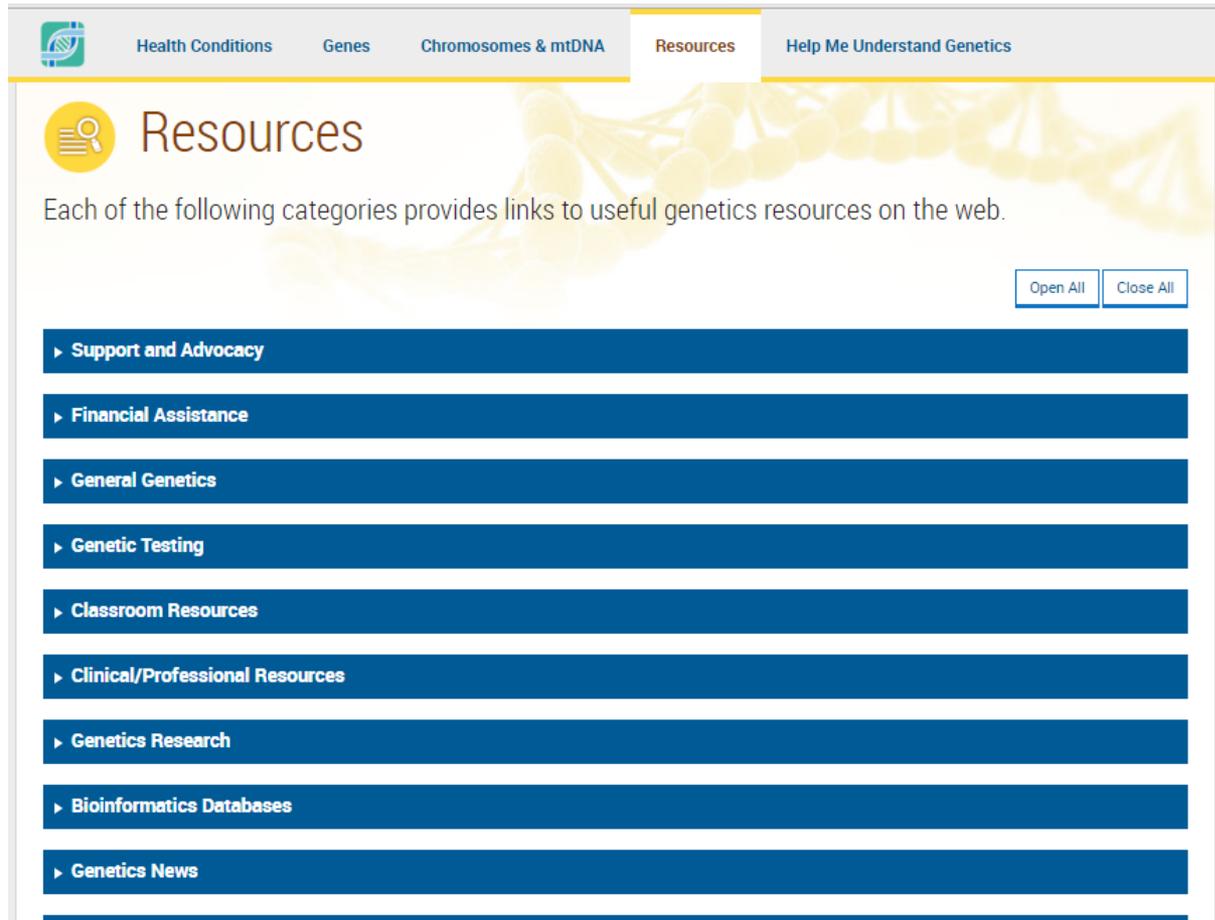
In many cases of breast cancer that clusters in families, the genetic basis for the disease and the mechanism of inheritance are unclear.

▶ Diagnosis & Management

### Related Information

- [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?](#)
- More about [Inheriting Genetic Conditions](#)

# Genetics Home Reference



The screenshot shows the 'Resources' page of the Genetics Home Reference website. The navigation bar at the top includes 'Health Conditions', 'Genes', 'Chromosomes & mtDNA', 'Resources' (which is highlighted), and 'Help Me Understand Genetics'. The main heading is 'Resources' with a magnifying glass icon. Below the heading, a text block states: 'Each of the following categories provides links to useful genetics resources on the web.' To the right of this text are two buttons: 'Open All' and 'Close All'. A list of ten resource categories is displayed as blue horizontal bars, each with a right-pointing arrow and the category name: 'Support and Advocacy', 'Financial Assistance', 'General Genetics', 'Genetic Testing', 'Classroom Resources', 'Clinical/Professional Resources', 'Genetics Research', 'Bioinformatics Databases', and 'Genetics News'.

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

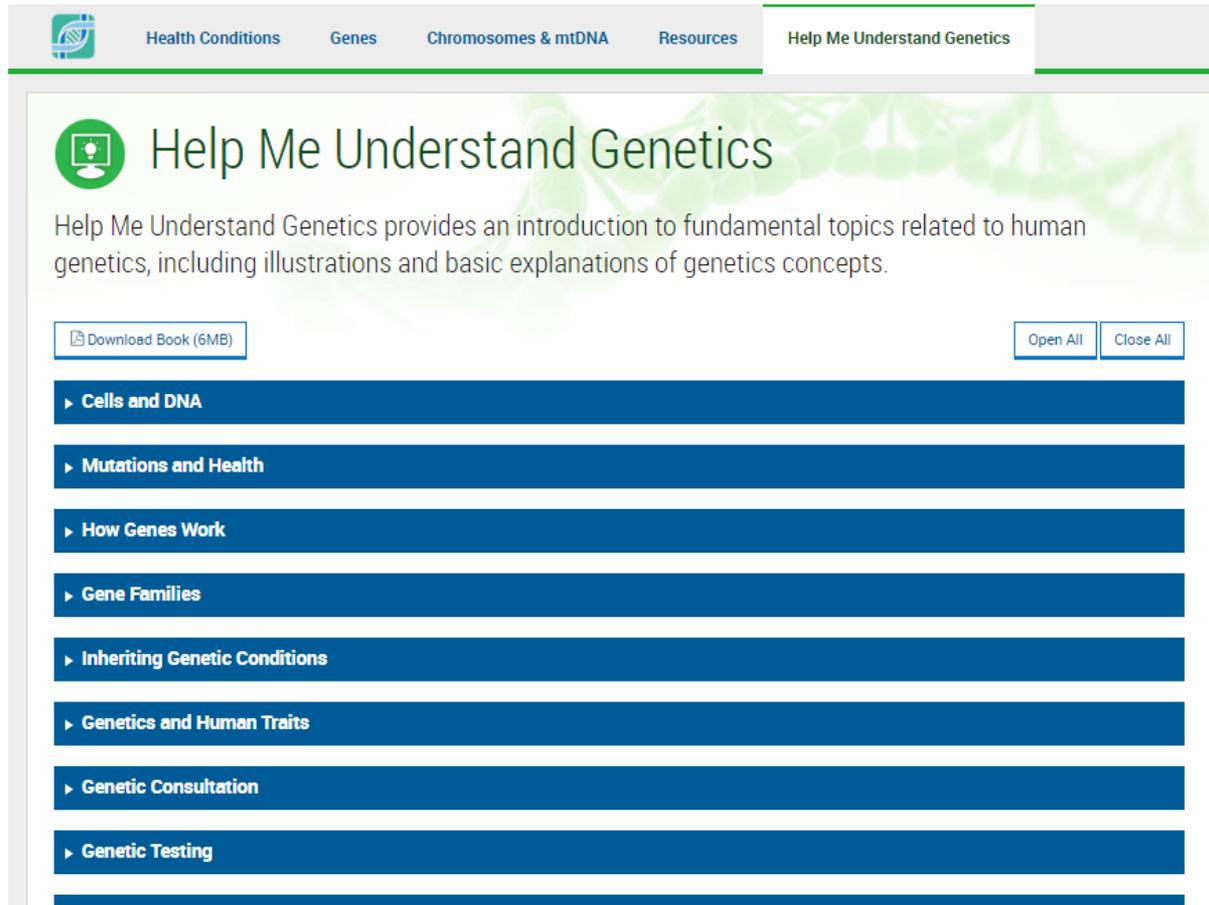
## Resources

Each of the following categories provides links to useful genetics resources on the web.

[Open All](#) [Close All](#)

- ▶ Support and Advocacy
- ▶ Financial Assistance
- ▶ General Genetics
- ▶ Genetic Testing
- ▶ Classroom Resources
- ▶ Clinical/Professional Resources
- ▶ Genetics Research
- ▶ Bioinformatics Databases
- ▶ Genetics News

# Genetics Home Reference



The screenshot displays the 'Help Me Understand Genetics' page. At the top, a navigation bar includes links for 'Health Conditions', 'Genes', 'Chromosomes & mtDNA', 'Resources', and 'Help Me Understand Genetics'. The main heading is 'Help Me Understand Genetics' with a lightbulb icon. Below the heading, a paragraph states: 'Help Me Understand Genetics provides an introduction to fundamental topics related to human genetics, including illustrations and basic explanations of genetics concepts.' There are two buttons: 'Download Book (6MB)' and 'Open All'. A list of topics is shown as blue bars with white text and a right-pointing arrow:

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

# NIH National Human Genome Research Institute

NIH National Human Genome Research Institute

Search Genome.gov

Español

Research Funding Research at NHGRI **Health** Education Issues Newsroom Careers About

## Health

Information about genetics and genomics, rare diseases, patient care and more

### For Patients and the Public

Detailed information about genetic disorders, background on genetic and genomic science, pharmacogenomics, family health history tool and online health resources



- › Community Engagement and Community Health
- › Family History
- › Genetics & Genomics Science & Research
- › Genetic & Rare Diseases Information Center
- › Genomic Medicine and Health Care
- › Online Health and Support Resources
- › Specific Genetic Disorders

### For Health Professionals

Genetics and genomics information related to patient management, education, NIH and NHGRI research and ethical, legal and social issues



- › Competency & Curricular Resources
- › Genetics 101
- › Genomic Medicine and Health Care
- › Inter-Society Coordinating Committee (ISCC)
- › New Horizons and Research
- › Patient Management
- › Policy and Ethics Issues

### Highlights

NIH awards \$55 million to build million-person precision medicine study



Bethesda, Md., Thurs., July 7, 2016 - The U.S. Food and Drug Administration (FDA) has announced two draft guidances to support President Obama's Precision Medicine Initiative. The guidances will help provide oversight for tests based on next generation sequencing, a technology that examines a person's DNA to detect medically important differences in genomic make-up that could increase the risk for disease.

### See Also

- GenomeTV
- Genomic Healthcare Branch
- Fact Sheets
- Genetic Education Resources for Teachers
- All About the Human Genome Project

# My Family Health Portrait U.S. Surgeon General

The screenshot shows the HHS.gov website with a blue header containing the HHS.gov logo and the text "U.S. Department of Health & Human Services". Below the header is a navigation bar with four tabs: "About HHS" (blue), "Programs & Services" (white), "Grants & Contracts" (purple), and "Laws & Regulations" (green). The "About HHS" tab is selected, and a sidebar on the left lists: "About the Family Health History Initiative", "Before You Start Your Family Health History", "My Family Health Portrait Tool", "English Web Tool", "Printable Versions", and "Source Code". The main content area features the title "The Surgeon General's Family Health History Initiative" and a paragraph: "To help focus attention on the importance of family history, the Surgeon General, in cooperation with other agencies with the U.S. Department of Health and Human Services, has launched a national public health campaign, called the Surgeon General's Family History Initiative, to encourage all American families to learn more about their family health history." Below this are three light blue boxes: "About the Family Health History Initiative" (with a target icon) containing the text "Learn about the Surgeon General's Family Health History Initiative"; "Before You Start" (with a magnifying glass icon) containing "Find tips to help you begin gathering information about your family's health history."; and "My Family Health History Portrait Tool" containing "Find out about this web-based tool to help you organize and print your family history".

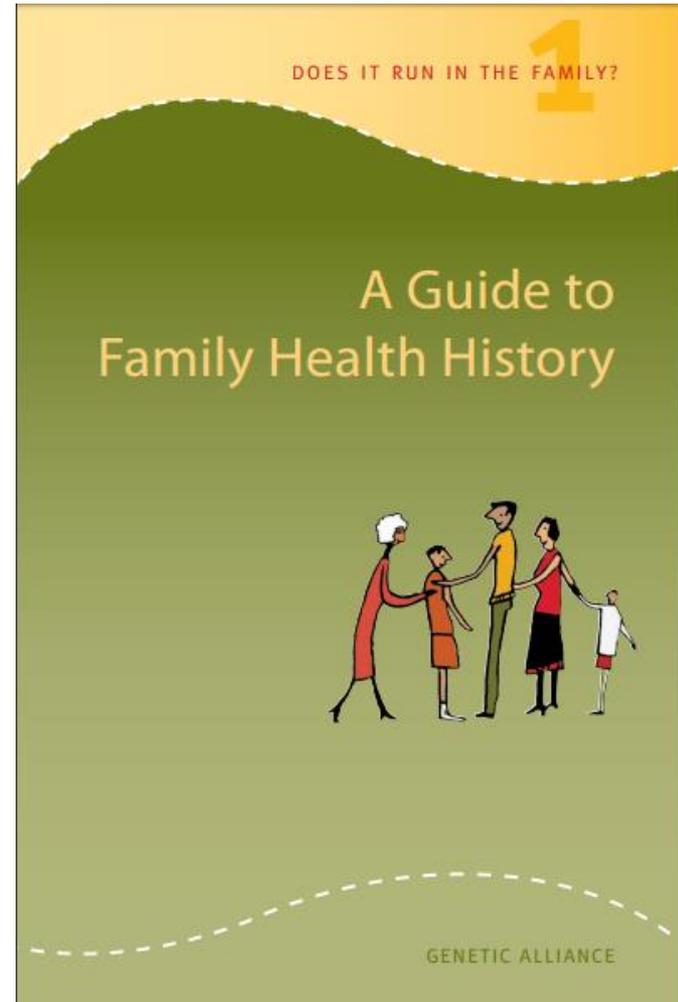
**[Surgeon General's Family Health History Initiative:](http://www.hhs.gov/programs/prevention-and-wellness/family-health-history/index.html)**

<http://www.hhs.gov/programs/prevention-and-wellness/family-health-history/index.html>

# Does It Run In the Family? toolkit

## [Does it Run In the Family? toolkit](http://www.geneticalliance.org/sites/default/files/GuideToFHH/GuidetoFHH.pdf)

<http://www.geneticalliance.org/sites/default/files/GuideToFHH/GuidetoFHH.pdf>



# Education Resources

**GeneEd** Genetics, Education, Discovery

Home Topics Labs & Experiments Teacher Resources Careers in Genetics Highlights search...

**Cell Biology**  
The study of the cell, including mitosis and meiosis

**Genetic Conditions**  
Conditions caused by gene variations or mutations

**DNA Forensics**  
The application of genetic testing for legal purposes

**DNA, Genes, Chromosomes**  
The building blocks of inheritance

**Evolution**  
Processes by which organisms are changed over time

**Top Issues In Genetics**  
Select genetic topics of popular interest

**Heredity/Inheritance Patterns**  
The genetic transmission of traits

**Biostatistics**  
The use of mathematics to investigate life science related problems

**Epigenetics/Inheritance and the Environment**  
The genome's dynamic response to the environment

**Biotechnology**  
Biological techniques used to enhance products

[GeneEd](https://geneed.nlm.nih.gov/)

<https://geneed.nlm.nih.gov/>

**NHGRI** National Human Genome Research Institute

Search Genome.gov

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Research Funding Research at NHGRI Health Education Issues Newsroom Careers About

**Education**  
Educational materials about genetics and genomics

**Smithsonian NHGRI Genome Exhibition**  
A genomics exhibition from the Smithsonian and NHGRI

**About The Human Genome Project**  
Information on the history, progress and impact of the HGP

**National DNA Day**  
A unique day when everyone can learn more about genomics and genetics

**Fact Sheets**  
Clearly written information on the institute, genetic research and genetic concepts

**Talking Glossary of Genetic Terms**  
Terms and definitions used in genetic research with multimedia

**Genetic Education Resources for Teachers**  
Teaching plans to present the science of genetics and genomics

**Online Education Kit**  
A web-based resource outlining the major history and developments of genomics

**Highlights**  
**Harry Potter and the Genetics of Wizarding**  
For NHGRI's National DNA Day inaugural speaker series, Eric P. Spana, Ph.D. from Duke University presented *Harry Potter and the Genetics of Wizarding*. The event took place on the NIH campus in Building 35A (Porter Building), Room 640. It is now available on NHGRI's YouTube channel, GenomeTV. Dr. Spana is an award-winning instructor in biology who helps students place new information in context with ideas they already find familiar, like Harry Potter, Star Wars and The Avenger.

**Student and Teacher Educational Resources from *Genome: Unlocking Life's Code***  
The NHGRI/Smithsonian traveling exhibit, *Genome Unlocking Life's Code*, has developed a wealth of educational digital assets for both students and teachers. Visit the *Genome: Unlocking Life's Code* site to discover all the ways you can enhance your learning and...

**See Also**  
Education and Community Involvement Branch  
GenomeTV  
Genome Advance of the Month  
Education Archive  
On Other Sites:  
Genome: Unlocking Life's Code NHGRI Smithsonian Exhibition  
GenomeTV  
NHGRI's YouTube Channel

[National Human Genome Research Institute](https://www.genome.gov/education/)

<https://www.genome.gov/education/>

# Harry Potter's World

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

Home Exhibition Education Resources Traveling Information Exhibition Program

“There was a lot more to magic,  
as Harry quickly found out,  
than waving your wand  
and saying a few funny words.”  
—*Harry Potter and the Philosopher's Stone*,  
J. K. Rowling



Illustration of an owl  
Konrad Gesner,  
*Historiae Animalium*, 1551

**EXHIBITION**



**EDUCATION RESOURCES**



**TRAVELING INFORMATION**



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

Home Exhibition Education Resources Traveling Information Exhibition Program

MIDDLE & HIGH SCHOOL HIGHER EDUCATION ONLINE ACTIVITIES BIBLIOGRAPHY

[English Lesson Plan](#) [Science Lesson Plan](#)

## GENETIC TRAITS IN HARRY POTTER

[Learning Outcomes](#) [Background Information](#) [Vocabulary](#) [Materials](#)

[Pre-Lesson](#) [Lesson 1](#) [Lesson 2](#) [Evaluations](#) [Extension](#) [Standards](#)

**Grade Level:** 7th-11th grades

**Time Needed:**

- Four 40-minute class periods for younger students with little knowledge of genetics (two periods for Pre-lesson Act
- Two 40-minute class periods for older students with strong knowledge of genetics (one period for Pre-lesson Activ

**Description:** The purpose of this lesson is to give students an introductory understanding of genetic inheritance. Studen heterozygous, recessive and dominant genes, genotype, phenotype, complex traits, Medelian inheritance, and Punnett Sq *Harry Potter*. Students also examine inheritance patterns of magical ability in Harry Potter, and use the concepts they ha

**Note:** The purpose of this lesson is to give students an introductory understanding of genetic inheritance. Therefore, the r

[Harry Potter's World](#)

<https://www.nlm.nih.gov/exhibition/harrypottersworld/index.html>

# More Resources



Public Health Genomics



*discover. educate. advocate.*



**GENETIC LITERACY PROJECT**  
SCIENCE TRUMPS IDEOLOGY

The text is set against a black background with a pattern of white DNA base pairs (A, T, C, G). A blue star graphic is positioned to the left of the text.

---

# Ethics and Privacy



# 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?
- What are the larger societal issues raised by new reproductive technologies?
- How will genetic tests be evaluated and regulated for accuracy, reliability and utility?
- How do we prepare healthcare professionals and the public?
- What is considered acceptable diversity?
- Where is the line between medical treatment and enhancement?
- Should testing be performed when no treatment is available?
- Who can afford genetic testing?

# GINA

**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.PROJECTHISO.NET ::

# NIH National Human Genome Research Institute

NIH National Human Genome Research Institute

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Research Funding Research at NHGRI Health Education **Issues** Newsroom Careers About

## Issues in Genetics

Policy, legal and ethical issues in genetic research



**Coverage and Reimbursement of Genetic Tests**  
Information about insurance coverage for genetic testing



**Human Subjects Research**  
Human subject participation for biomedical, clinical and social-behavioral research



**Genetic Discrimination**  
How Americans are protected from discrimination based on their genetics



**Regulation of Genetic Tests**  
How the federal government regulates genetic tests.



**Privacy in Genomics**  
How best to ensure that genomic information remains private



**Informed Consent**  
The rights of participants when consenting to research projects



**Intellectual Property and Genomics**  
Can a gene be patented?



**Genetics and Public Policy Fellowship**  
A fellowship for genetics professionals interested in public policy



**Genome Statute and Legislation Database**  
A database of state statutes and bills from 2007-2016 U.S. state legislative sessions

## Highlights

FDA requests comments on draft guidance for Precision Medicine Initiative



The U.S. Food and Drug Administration (FDA) has announced two draft guidances to support President Obama's Precision Medicine Initiative. The guidances will help provide oversight for tests based on next generation sequencing.

## See Also

Policy and Program Analysis Branch  
Staff Contact Information

Ethical, Legal and Social Implications Research Program  
NHGRI's Extramural Research Program

GenomeTV

# Informing the Public



CENTER FOR  
GENETICS AND  
SOCIETY



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# Precision Medicine

“...a bold new research effort to revolutionize how we improve health and treat disease.”



# Precision Medicine Initiative

## Mission statement:

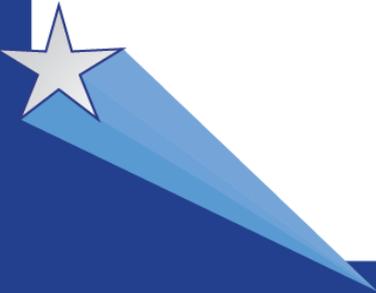
*To enable a new era of medicine through research, technology, and policies that empower patients, researchers, and providers to work together toward development of individualized care.*



[PMI announcement](https://www.whitehouse.gov/precision-medicine) <https://www.whitehouse.gov/precision-medicine>

# Precision Medicine is...

- 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 this patient.



# Precision Medicine Initiative

## Near Term goals:

- Clinical trials focusing on pediatric cancers and drug therapies for adults
- Use of combination therapies
- Overcoming drug resistance

## Long Term Goals:

- Create research cohort of 1 million volunteers
- New model of medicine
  - engage participants
  - responsible data sharing
  - privacy protection
- Advance pharmacogenomics
- Identify new targets for treatment and prevention
- Test if mobile devices encourages healthy behaviors
- Lay scientific foundation for many diseases

[All of Us infographic:](#)

<https://www.nih.gov/sites/default/files/research-training/initiatives/pmi/allofus-inforgraphic-20161117.pdf>

# All of Us Research Program

## Two ways to participate:

1. Through the participant website
2. With participating Healthcare Provider Organizations (HPOs)



**All of Us<sup>SM</sup> Research Program**



**WHAT IS IT?**

**Precision medicine** is a groundbreaking approach to disease prevention and treatment based on people's individual differences in environment, genes and lifestyle.

The *All of Us* Research Program will lay the foundation for using this approach in **clinical practice**.

**WHAT ARE THE GOALS?**

Engage a group of **1 million or more U.S. research participants** who will share biological samples, genetic data and diet/lifestyle information, all linked to their electronic health records. This data will allow researchers to develop more precise treatments for **many diseases and conditions**.

Pioneer a new model of research that emphasizes **engaged research participants, responsible data sharing and privacy protection**.



Research based on the cohort data will:

- Lay **scientific foundation** for precision medicine
- Help identify new ways to **treat and prevent disease**
- Test whether **mobile devices**, such as phones and tablets, can encourage healthy behaviors
- Help develop the **right drug** for the **right person** at the **right dose**

**WHY NOW?**

The **time is right** because:

We have a greater understanding of human genes	People are more engaged in healthcare and research
	
We have the tools to track health information and use large databases	Research technologies have improved
	

Follow the Program's progress and be one of the first to join this landmark effort.

[www.nih.gov/AllOfUs-Research-Program](http://www.nih.gov/AllOfUs-Research-Program)

# NIH and Precision Medicine Initiative

U.S. Department of Health & Human Services

**NIH** National Institutes of Health  
Turning Discovery Into Health

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## ALL OF US RESEARCH PROGRAM

**All of Us Research Program**

- Scale and Scope
- Participation
- Program Components
- Funding
- FAQ
- Advisory Groups
- Events
- Announcements
- In the News
- Multimedia

**All of Us Infographic**

**All of Us Research Program**

**WHAT IS IT?**

Precision medicine is a groundbreaking approach to disease prevention and treatment based on people's individual differences in environment, genes and lifestyle. The All of Us Research Program will lay the foundation for using this approach in clinical practice.

**WHAT ARE THE GOALS?**

Dara Richardson-Heron, M.D. Named Chief Engagement Officer of the All of Us Research Program

All of Us Research Program announces funding opportunity for community partners.

### About the All of Us Research Program

Far too many diseases do not have proven preventions or treatments. To make a difference for the millions of Americans who suffer from them, we must gain better insights into the biological, environmental, and behavioral factors that drive these diseases. Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in environment, lifestyle and genes for each person.

As part of the federal Precision Medicine Initiative, the NIH is leading the effort to build a national, large-scale research enterprise with one million or more volunteers to extend precision medicine to all diseases. The All of Us Research Program, formerly known as the PMI Cohort Program, will be a participant-engaged, data-driven enterprise supporting research at the intersection of lifestyle, environment, and genetics to produce new knowledge with the goal of developing more effective ways to prolong health and treat

### Email Updates

Sign up to receive email updates about the Precision Medicine Initiative.

[Sign up for updates](#)

### Related Links

- [PMI Working Group Final Report](#)
- [NEJM Perspective: A New Initiative on Precision Medicine](#)
- [Precision Medicine Initiative and Cancer Research](#)
- [Precision Medicine Initiative YouTube Channel](#)

All of Us, the All of Us logo, Precision Medicine Initiative, PMI and The Future of Health Begins with You are service marks of the U.S. Department of Health and Human Services (HHS).

[All of Us Research Program:](https://www.nih.gov/research-training/allofus-research-program)

<https://www.nih.gov/research-training/allofus-research-program>

# MedlinePlus Magazine- Fall 2015



**Health Care Tailored to You**

# PNR Rendezvous

**Adventures in Precision  
Medicine: A Major Public  
Research Initiative and its  
Implications for Healthcare  
Consumers and Institutions**  
September 21, 2016



**Presenter: Malia Fullerton,**  
Associate Professor of Bioethics and  
Humanities at the University of  
Washington School of Medicine



# 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”](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4115323/)

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4115323/>

# Show What You Know!

- What initiative refers to strategies for determining what treatment is right for an INDIVIDUAL rather than what treatment is recommended for a DISEASE?
- What is the name of the volunteer research program that is looking to collect data on 1 million volunteers to assist with the Precision Medicine Initiative?
- True or False? GINA (Genetic Information Nondiscrimination Act) protects you from life insurance discrimination.
- What resource would you recommend to consumers who wanted to learn more about a genetic testing?

# Questions?

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Consumer Health Coordinator  
NNLM PNR  
martinc4@uw.edu

## [Presentation resources](https://nnlm.gov/pnr/guides/training-resources-you-can-use/presentations)

[https://nnlm.gov/pnr/guides/  
training-resources-you-can-use/presentations](https://nnlm.gov/pnr/guides/training-resources-you-can-use/presentations)

