U.S. National Library of Medicine
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S of DNA:

HELPING PATRONS UNRAVEL THE MYSTERY OF GENETIC INFORMATION

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Pacific Northwest Region (NNLM PNR)
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Genetics Overview

Genomic Health Literacy

Genetic Testing

Consumer Health Resources

Ethics & Privacy

All of Us Research Program

NNL…Huh?

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

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 6 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

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/

Genetics in the News

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

Human Gene Editing Receives Science Panel's Support

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

Scientists Use Genetic Engineering To Vanquish Disease-Carrying Insects

Baltimore Ravens to hand out free DNA test kits

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

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

Mail-Order CRISPR Kits Allow Absolutely Anyone to Hack DNA

Genetic Testing for Athletic Ability

Can genes predict sporting talent?

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

Genetically Modified Humans? How Genome Editing Works

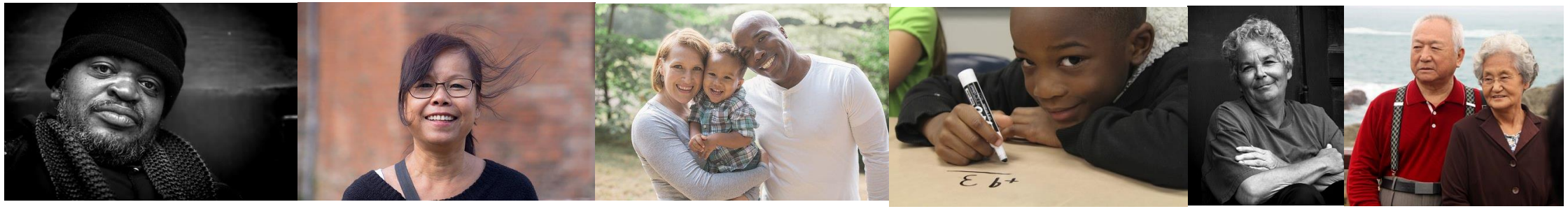
Birth of Baby With Three Parents' DNA Marks Success for Banned Technique

Genomic Health Literacy

Lack biology basics

Lack mathematical concepts

Low health literacy



Definitions

Genomic Health Literacy

- The capacity to obtain, process, understand, and use genomic information for health related decision making.

Genomic Science Literacy

- The knowledge of basic genetics and genomics concepts and processes needed to build conceptual understanding, and the necessary mathematical knowledge to support this comprehension.

Leading causes of death

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

1. Heart disease: 633,942
2. Cancer: 595,930
3. Chronic lower respiratory diseases: 155,041
4. Accidents (unintentional injuries): 146,571
5. Stroke (cerebrovascular diseases): 140,323
6. Alzheimer's disease: 110,561
7. Diabetes: 79,535
8. Influenza and pneumonia: 57,062
9. Nephritis, nephrotic syndrome, and nephrosis: 49,959
10. Intentional self-harm (suicide): 44,193

CDC FastStats

The Story of You

The Story of You

CATEGORIES OF DISEASES ATTRIBUTED TO GENES

- Chromosomal Diseases
- Monogenic Diseases
- Multifactorial Diseases

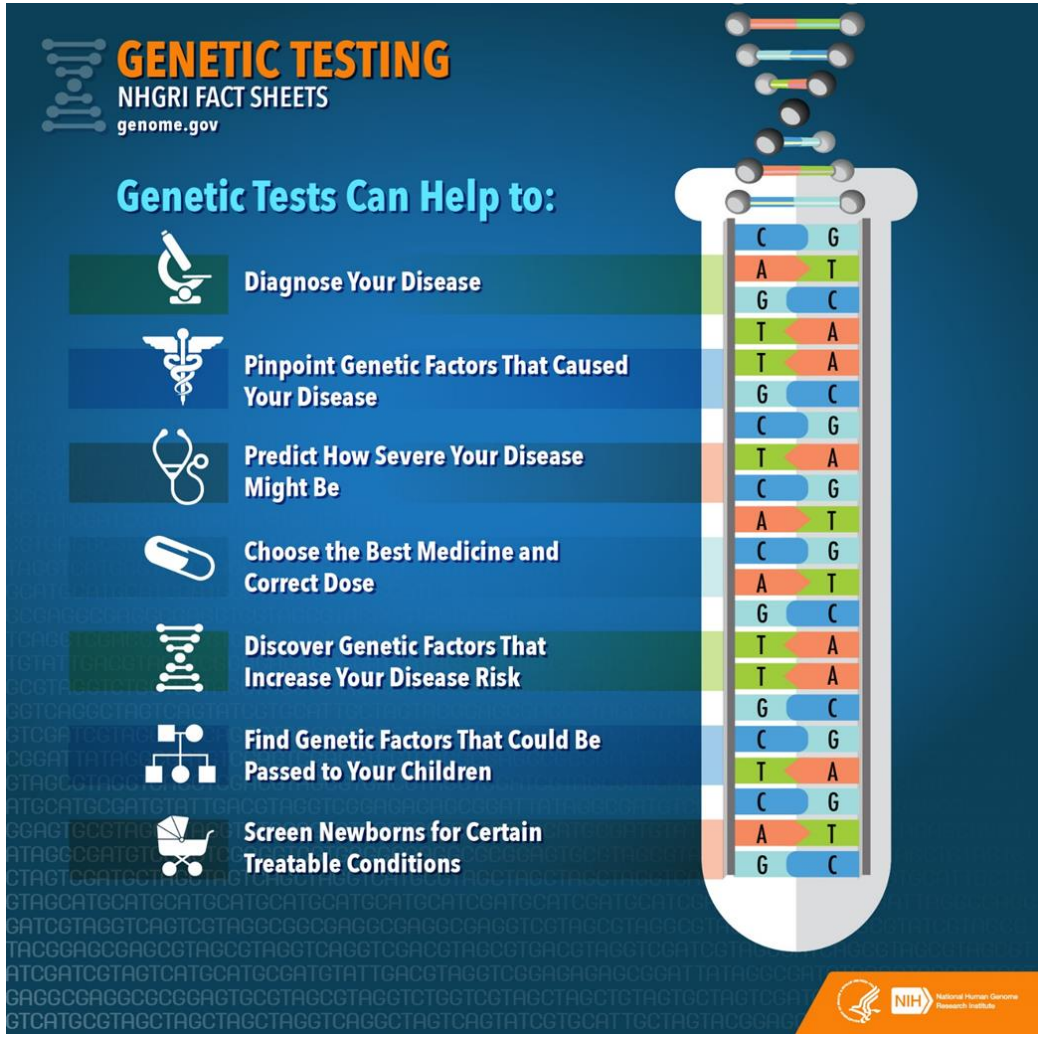
Genetic Testing

INCLUDING DIRECT-TO-CONSUMER

Types of Genetic Tests

- Diagnostic
- Predictive
- Carrier
- Prenatal
- Newborn Screening
- Research
- Pharmacogenetic

Clinical Uses of Genetic Tests



Genetic Testing

Jean's Genetic Testing Timeline

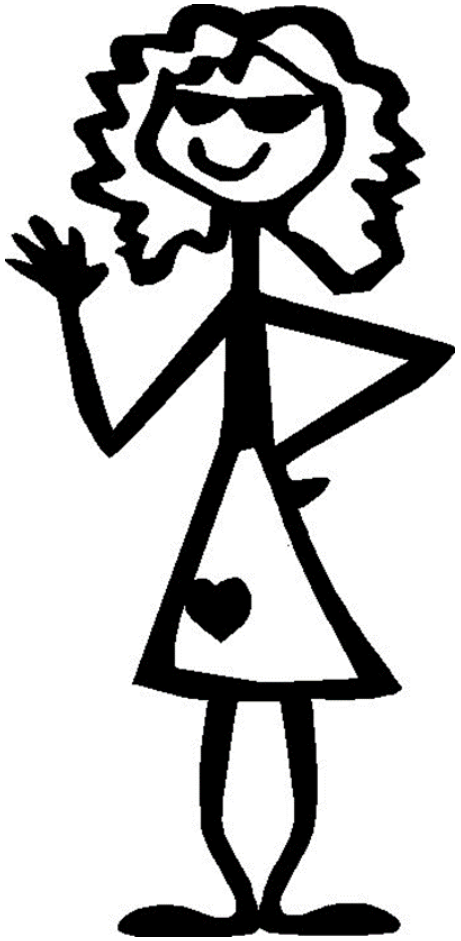
Age 1 day: **newborn** testing for a few serious childhood diseases

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 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.

BRCA 1 & 2

- Majority of breast and ovarian cancers are not linked to BRCA
- Only 0.2% carry BRCA mutations
- U.S. Preventive Services Task Force recommends that women who have family members with breast, ovarian, tubal, or peritoneal cancer be assessed
- Women who are found to have a family history that may be associated with BRCA1 or BRCA2 mutations should receive genetic counseling and subsequent BRCA testing, if indicated
- Having the mutation does not necessarily mean cancer will develop, but it does increase risk

Lab Tests Online BRCA information
NCI BRCA Fact Sheet

DTC BRCA test

U.S. Department of Health and Human Services

FDA U.S. FOOD & DRUG ADMINISTRATION

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FDA News Release

FDA authorizes, with special controls, direct-to-consumer test that reports three mutations in the BRCA breast cancer genes

Test only reports 3 out of more than 1,000 known BRCA mutations and negative result doesn't rule out increased cancer risk

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For Immediate Release March 6, 2018

Release

Español

The U.S. Food and Drug Administration today authorized the Personal Genome Service Genetic Health Risk (GHR) Report for BRCA1/BRCA2 (Selected Variants). It is the first direct-to-consumer (DTC) test to report on three specific BRCA1/BRCA2 breast cancer gene mutations that are most common in people of Ashkenazi (Eastern European) Jewish descent. These three mutations, however, are not the most common BRCA1/BRCA2 mutations in the general population.

Inquiries

Media

Deborah Kotz
301-796-5349

Consumers

888-INFO-FDA

Related Information

- FDA allows marketing of first direct-to-consumer tests that provide genetic risk information
- Commissioner statement on consumer tests that evaluate genetic health risks
- Diagnostic tests

Follow FDA

23andMeBlog

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A Major Milestone in Consumer Health Empowerment

March 6, 2018 By AnneW under Health and Traits

We wear pink ribbons. We walk or run for the cause. We have a month for awareness, and yet many women who have a higher risk variant in the BRCA1 or BRCA2 genes remain unaware of their heightened genetic risk for breast and ovarian cancer until a doctor diagnoses them with cancer.[1]

That's a failure.

This is not a radical indictment of the American healthcare system; it's just a fact. We are very good at treating illnesses, but not at preventing them.

Under the current system, there are specific guidelines for BRCA screening that limit who has access to BRCA testing. Family history of cancer and Ashkenazi Jewish ancestry are the most common criteria for screening guidelines. However we have seen at 23andMe that many people do not know their family medical history or their ancestry. So,

FDA announcement

23andMe announcement

Genetic Testing- is it necessary?

Choosing Wisely®
An initiative of the ABIM Foundation

ConsumerReportsHealth

ABIM FOUNDATION

ACMG American College of Medical Genetics and Genomics
Translating Genes Into Health®

Making smart decisions about genetic testing:

Avoid unneeded tests, and understand the tests you need

Genetic testing can help identify an inherited condition or disease risk. The test results might help you and your doctor:

- Choose ways to prevent or treat a condition.
- Decide which screening tests you need (to find a disease at an early stage when it might be more treatable).

Genetic testing may also tell you which family members are at risk.

But sometimes a genetic test is not the best way to find an inherited condition or disease risk. A routine blood test or procedure might be just as good. And it might be less costly and more easily available.

Know what to expect.
Before you have any genetic test, you should understand its possible benefits, harms, and limitations. And you should think about how it might affect others in your family.

Talk to your doctor or a medical geneticist.

- Diagnose genetic conditions.
- Select the most appropriate genetic tests.
- Explain test results to patients and their families.
- Recommend personalized treatment and prevention options.

Choosing Wisely

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?

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

ancestryDNA™

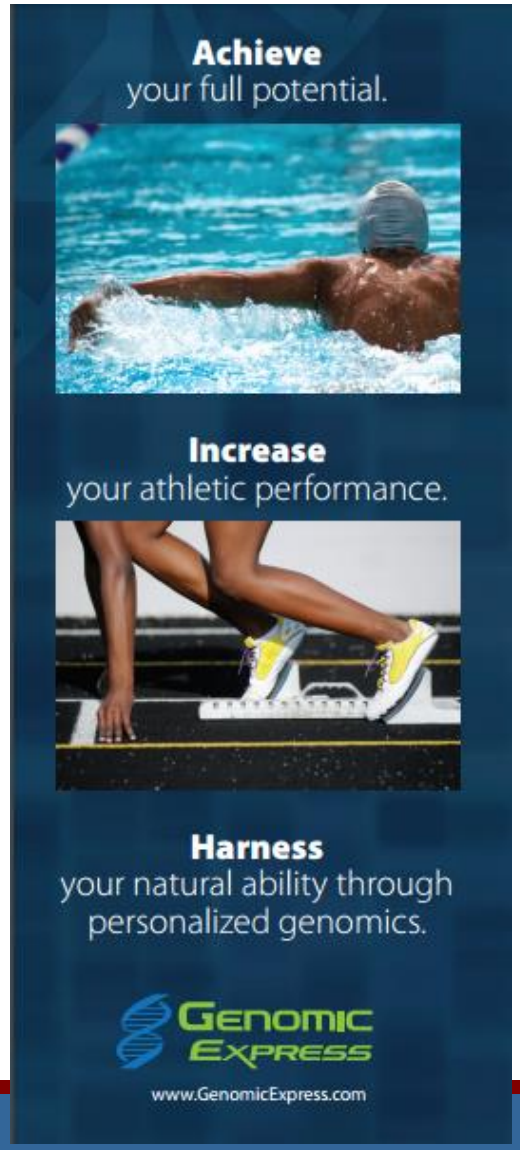
SUPERHERO
DNA Test

NUTRITION
DNA Test

FITNESS
DNA Test

Genomic Testing- Athletic Ability

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



Genomic Testing- Consensus Statement

Consensus statement

OPEN ACCESS

Direct-to-consumer genetic testing for predicting sports performance and talent identification: Consensus statement

Nick Webborn,[1] Alun Williams,[2] Mike McNamee,[3] Claude Bouchard,[4] Yannis Pitsiladis,[5] Ildus Ahmetov,[6] Euan Ashley,[7] Nuala Byrne,[8] Silvia Camporesi,[9] Malcolm Collins,[10] Paul Dijkstra,[11] Nir Eynon,[12] Noriyuki Fuku,[13] Fleur C Garton,[14] Nils Hoppe,[15] Søren Holm,[16] Jane Kaye,[17] Vassilis Klissouras,[18] Alejandro Lucia,[19] Kamiel Maase,[20] Colin Moran,[21] Kathryn N North,[14] Fabio Pigozzi,[22] Guan Wang[5]

▸ Additional material is published online only. To view please visit the journal online (http://dx.doi.org/10.1136/bjsports-2015-095343).

For numbered affiliations see end of article.

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Accepted 25 September 2015

ABSTRACT
The general consensus among sport and exercise genetics researchers is that genetic tests have no role to play in talent identification or the individualised prescription of training to maximise performance. Despite the lack of evidence, recent years have witnessed the rise of an emerging market of direct-to-consumer marketing (DTC) tests that claim to be able to identify children's athletic talents. Targeted consumers include mainly coaches and parents. There is concern among the scientific community that the current level of knowledge is being misrepresented for commercial purposes. There remains a lack of universally accepted guidelines and legislation for DTC testing in relation to all forms of genetic testing and not just for talent identification. There is concern over the lack of clarity of information over which specific genes or variants are being tested and the almost universal lack of appropriate genetic counselling for the interpretation of the genetic data to consumers. Furthermore independent studies have identified issues relating to quality control by DTC laboratories with different results being reported from of the evidence in relation to genetic testing and the limitations of current knowledge. This article reviews the issues around the currently available evidence behind the genetic testing, comments on the ethical considerations and makes recommendations about such tests.

STATEMENT ON BACKGROUND TO THE CONSENSUS PROCESS
A group of world experts in the field of genomics, exercise, sport performance, disease, injury and antidoping gathered with the International Federation of Sports Medicine (FIMS) Scientific Commission for a symposium to discuss the current state of knowledge and to share ideas. One key concern was the misuse of research evidence and the misinformation about genetic testing, particularly when marketed directly to the public, coaches or parents. This is known as DTC testing for the purpose of talent identification and to assess potential for future sports performance. There have been

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

Genetics Home Reference

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

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Genetic Testing

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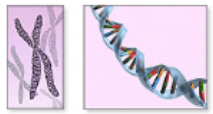

A.D.A.M.

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.

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BRCA1 and BRCA2 gene testing
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Direct-to-Consumer Genetic Tests

Vea esta página en español

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

Anatomy of a Cancer Treatment Scam

- Dietary Supplements
- Miracle Health Claims
- Cancer Treatment Scams

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

American College of Medical Genetics and Genomics

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ACMG STATEMENT | Genetics inMedicine

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

ACMG Board of Directors[1]

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

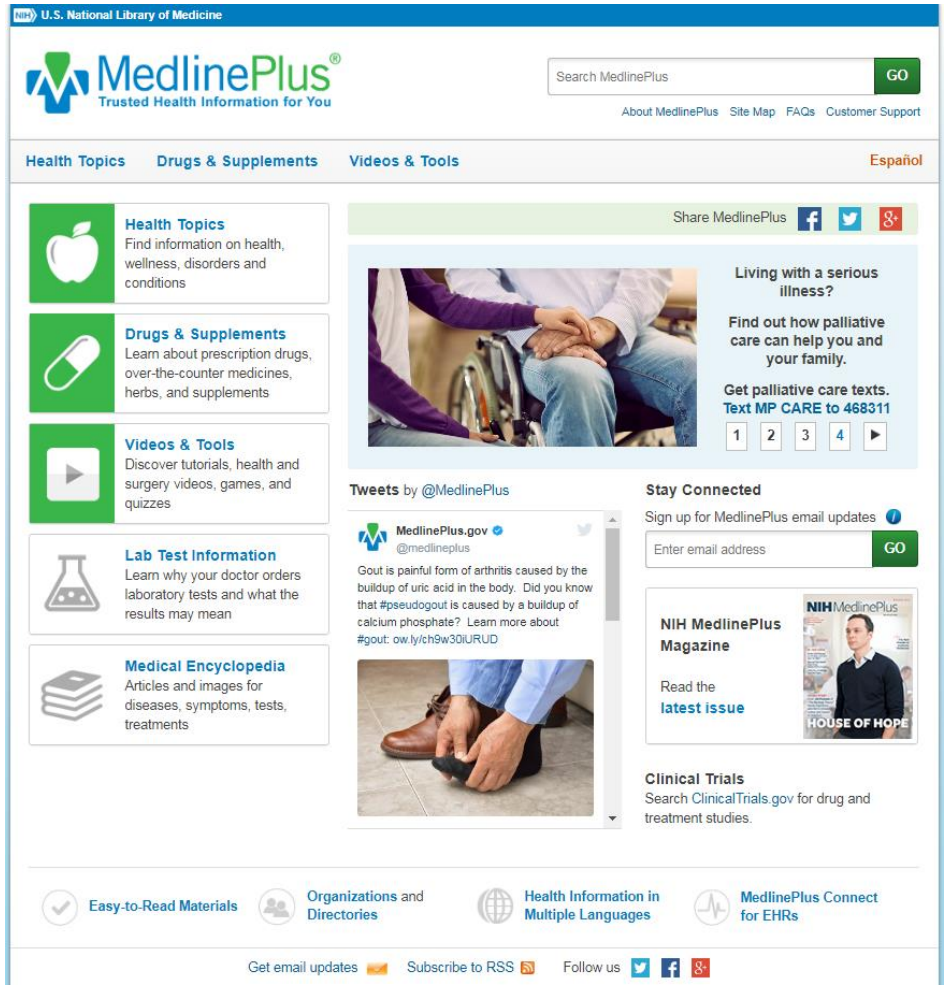
Questions to ask before using a Direct to Consumer Genetic Test

- 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?

Consumer Resources

PATIENT AND K-12 EDUCATION

MedlinePlus



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  - Genetic testing
  - Genetic counseling
  - Genetic disorders
  - Genetic brain disorders
  - Genes and gene therapy
- text word search

MedlinePlus – Genetics topics

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

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MedlinePlus – stroke topic page

U.S. National Library of Medicine

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Stroke

Also called: Brain attack, CVA

On this page

Basics
- Summary
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- Symptoms
- Diagnosis and Tests
- Prevention and Risk Factors
- Treatments and Therapies

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MEDICAL ENCYCLOPEDIA

- BAER - brainstem auditory evoked response
- Brain herniation
- Brain surgery
- Brain surgery - discharge
- Craniotomy - slideshow
- EEG

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

Genetics

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

Images

- Craniotomy - slideshow (Medical Encyclopedia)
  Also in Spanish

Health Check Tools

- Test Your Stroke Knowledge (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
  (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 (National Institutes of Health)
- ClinicalTrials.gov: Cerebrovascular Disorders (National Institutes of Health)

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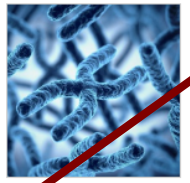
Genetic Disorders

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.

(Read more)

Results **1** - **10** of **5,973** for **genetics**

1. **Genetic** Disorders (National Library of Medicine)
   ... This can cause a medical condition called a **genetic** disorder. You can inherit a gene mutation from ... during your lifetime. There are three types of **genetic** disorders: Single-gene disorders, where a mutation affects ...
   https://medlineplus.gov/geneticdisorders.html - Health Topics
2. **Genetic** Brain Disorders (National Library of Medicine)
   A **genetic** brain disorder is caused by a variation or a mutation in a gene. A variation is a ... A mutation is a change in a gene. **Genetic** brain disorders affect the development and function of ...
   https://medlineplus.gov/geneticbraindisorders.html - Health Topics
3. **Genetic** Counseling (National Library of Medicine)
   Genetic counseling provides information and support to people who have, or may be at risk for, **genetic** disorders. A **genetic** counselor meets with you to discuss **genetic** risks. ...
   https://medlineplus.gov/geneticcounseling.html - Health Topics

Textword search ‘genetics’

Genetics Home Reference

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

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Your Guide to Understanding Genetic Conditions

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Health Conditions

Explore the signs and symptoms, genetic cause, and inheritance pattern of various health conditions.

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Q R S T U V W X Y Z

A-alphalipoprotein Neuropathy, *see* Tangier 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
AATD, *see* Alpha-1 antitrypsin deficiency
AB variant, *see* GM2-gangliosidosis, AB variant
ABCB11-related intrahepatic cholestasis, *see* Progressive familial intrahepatic cholestasis

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?

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

Genetics Home Reference

Genetics Home Reference | Your Guide to Understanding Genetic Conditions

Search

Health Conditions | Genes | Chromosomes & mtDNA | Resources | 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 people with Williams syndrome. Supravalvular aortic stenosis is a narrowing of the large blood vessel that carries blood from the heart to the rest of the body (the aorta). If this condition is not treated, the aortic narrowing can lead to shortness of breath, chest pain, and heart failure. Other problems with the heart and blood vessels, including high blood pressure (hypertension), have also been reported in people with Williams syndrome.

Additional signs and symptoms of Williams syndrome include abnormalities of connective tissue (tissue that supports the body's joints and organs) such as joint problems and soft, loose skin. Affected people may also have increased calcium levels in the blood (hypercalcemia) in infancy, developmental delays, problems with coordination, and short stature. Medical problems involving the eyes and vision, the digestive tract, and the urinary system are also possible.

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

Genetics Home Reference | Your Guide to Understanding Genetic Conditions

Search

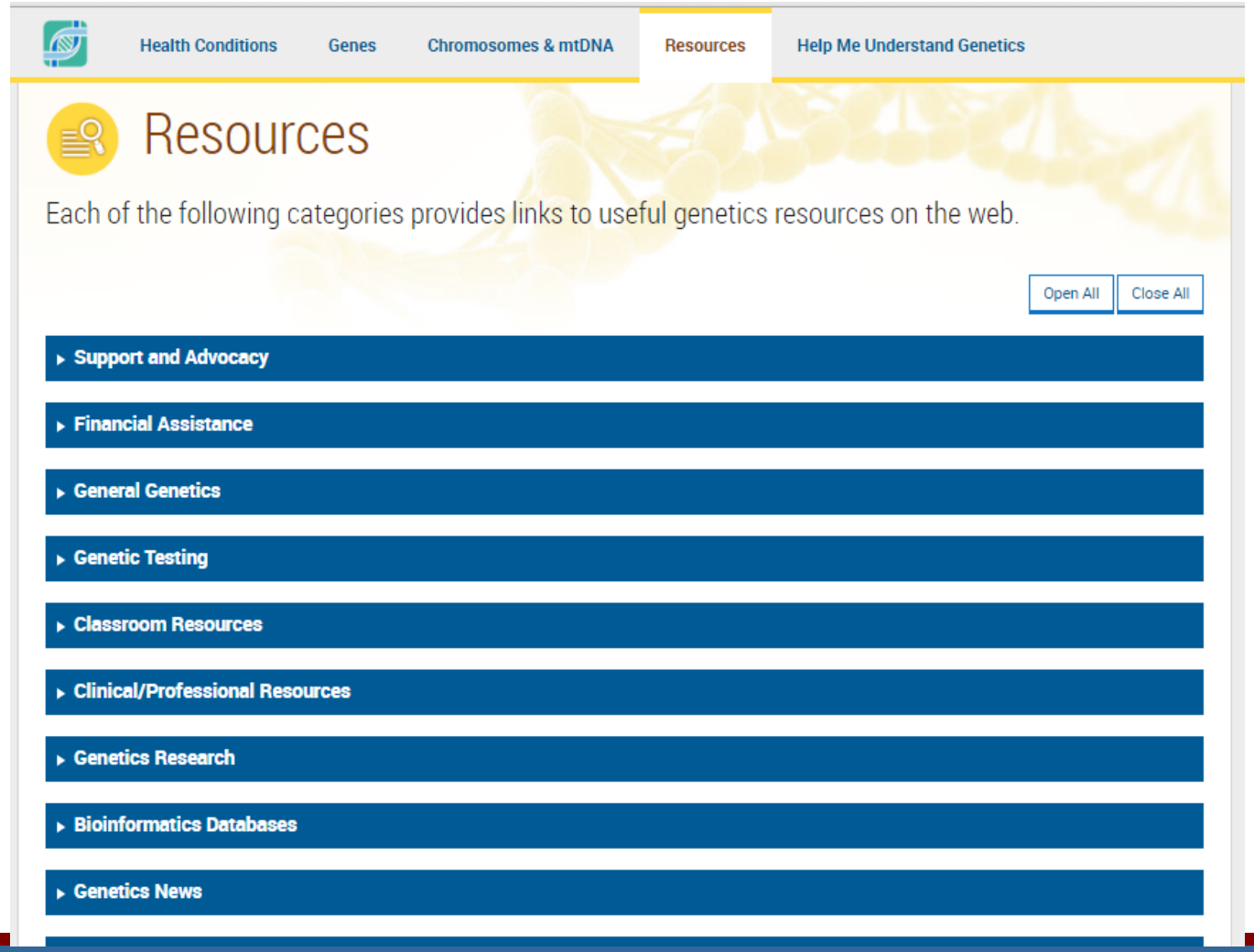
Health Conditions | Genes | Chromosomes & mtDNA | Resources | Help Me Understand Genetics

Williams syndrome

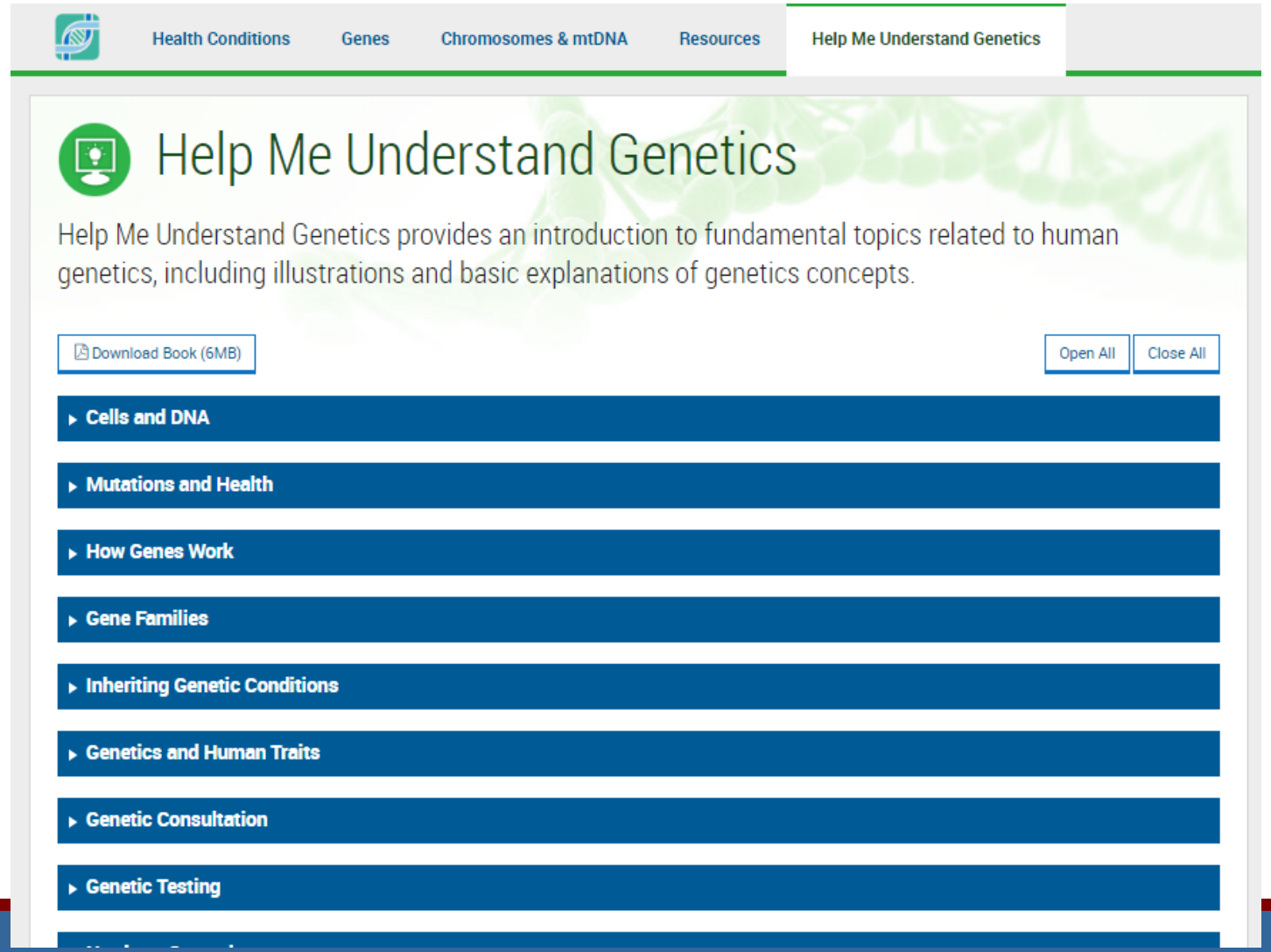
Printable PDF | Open All | Close All

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

Genetics Home Reference



Genetics Home Reference



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NIH National Human Genome Research Institute- health information

National Human Genome Research Institute

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Español

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

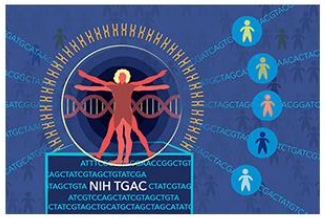
Highlights

The 2018 Jeffrey M. Trent Lecture in Cancer Research

NHGRI's Division of Intramural Research will present the 14th Jeffrey M. Trent Lecture in Cancer Research on March 15, 2018, 12:00 - 1:00 p.m., at the Lipsett Amphitheater, Building 10 (Clinical Center), on the National Institutes of Health Bethesda campus. Joan Brugge, Ph.D., co-director, Ludwig Center at Harvard Medical School, will deliver the lecture *Role of the TRPA1 Ca2+-permeable Channel in Oxidative Stress Defenses in Cancer*. Watch it at Genome TV Live on March 15 starting at 12:00 p.m. Eastern.

NIH researchers highlight virtual reality research on Reddit

Virtual Reality (VR) is a rapidly expanding area in tech and gaming. Now it's also playing an important role in medicine and health, expanding opportunities for researchers, clinicians and patients. Simulated experience can reduce stress, help doctors practice surgical techniques or allow medical students a way to practice their bedside manner in virtual scenarios. On February 23, NHGRI hosted a Reddit "Ask Me Anything" (AMA) with NIH researchers who use VR to study a host of research questions. Read our recap here.



NIH pilot project will match researchers to genes, gene variants of interest

The National Institutes of Health and Inova have launched a new match-making service between genes and gene variants and the researchers who study them. The Genomic Ascertainment Cohort (TGAC) project will be based in the Washington, D.C., area so that researchers can recall genotyped participants to examine the genes and gene variants that influence their phenotype.

Email Updates

Enter your email | Subscribe

Genomics News

From the African Academy of Sciences: **African Academy of Sciences awards grants to better understand the diseases that affect Africa most**
March 1, 2018

From The National Institute of Environmental and Health Sciences: **Building Capacity in Africa for Genomics and Environmental Health Research**
February 20, 2018

From The Ohio State University Comprehensive Cancer Center: **New Report Labs Differ Widely in BRCA Testing Protocols**
February 16, 2018

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Quick Links

Director Eric Green

Strategic Planning

National Human Genome Research Institute

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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

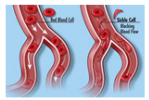
For Health Professionals

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

Researchers pinpoint origin of sickle cell mutation



NHGRI researchers used whole genome sequence data to pinpoint the single origin of the sickle cell mutation to the "wet" period of the Sahara 7,300 years ago. The mutation causes blood hemoglobin to be crescent shaped, reducing its ability to carry oxygen. Charles N. Rotimi, Ph.D., study co-author and NHGRI senior investigator, said the finding overturns previous theories that the mutation arose in multiple locations. This will help clinicians redefine sickle cell subgroups and treat patients more effectively, said lead author Daniel Shriner, Ph.D. Read more in the March 8 *American Journal of Human Genetics*.

Reddit "Ask Me Anything" Recap: The importance of knowing your family health history

See Also

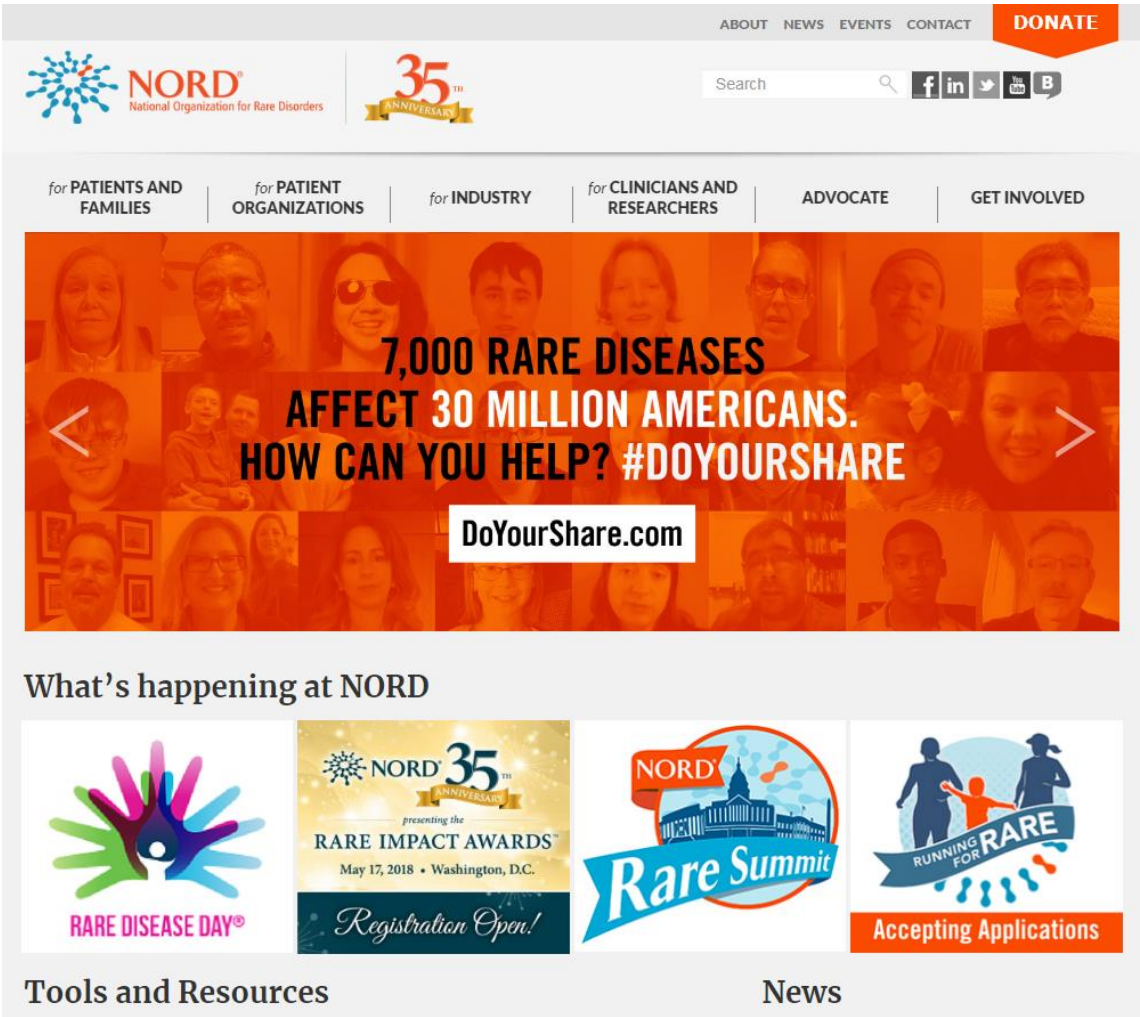
- GenomeTV
- Genomic Healthcare Branch
- Fact Sheets
- Genetic Education Resources for Teachers
- All About the Human Genome Project
- Health Archive

On Other Sites:

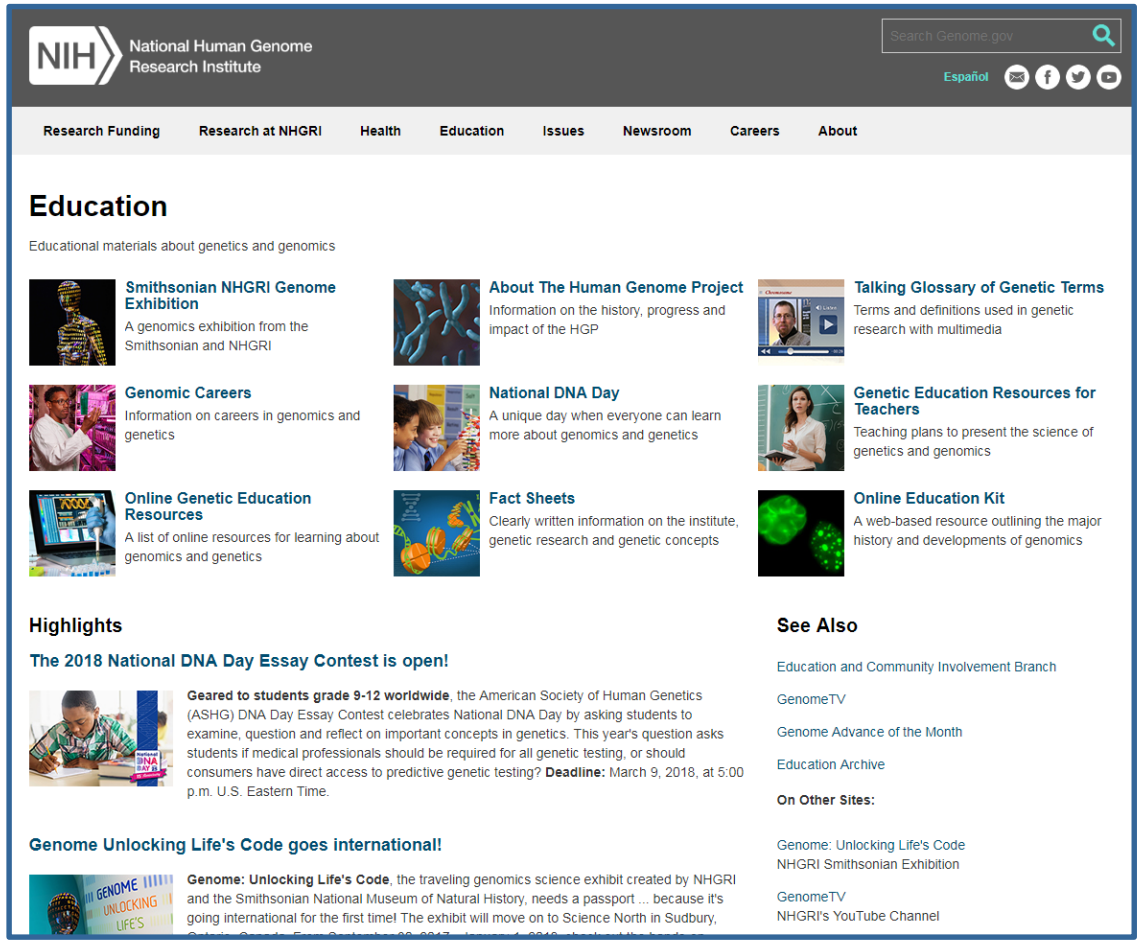
NHGRI's YouTube channel: GenomeTV

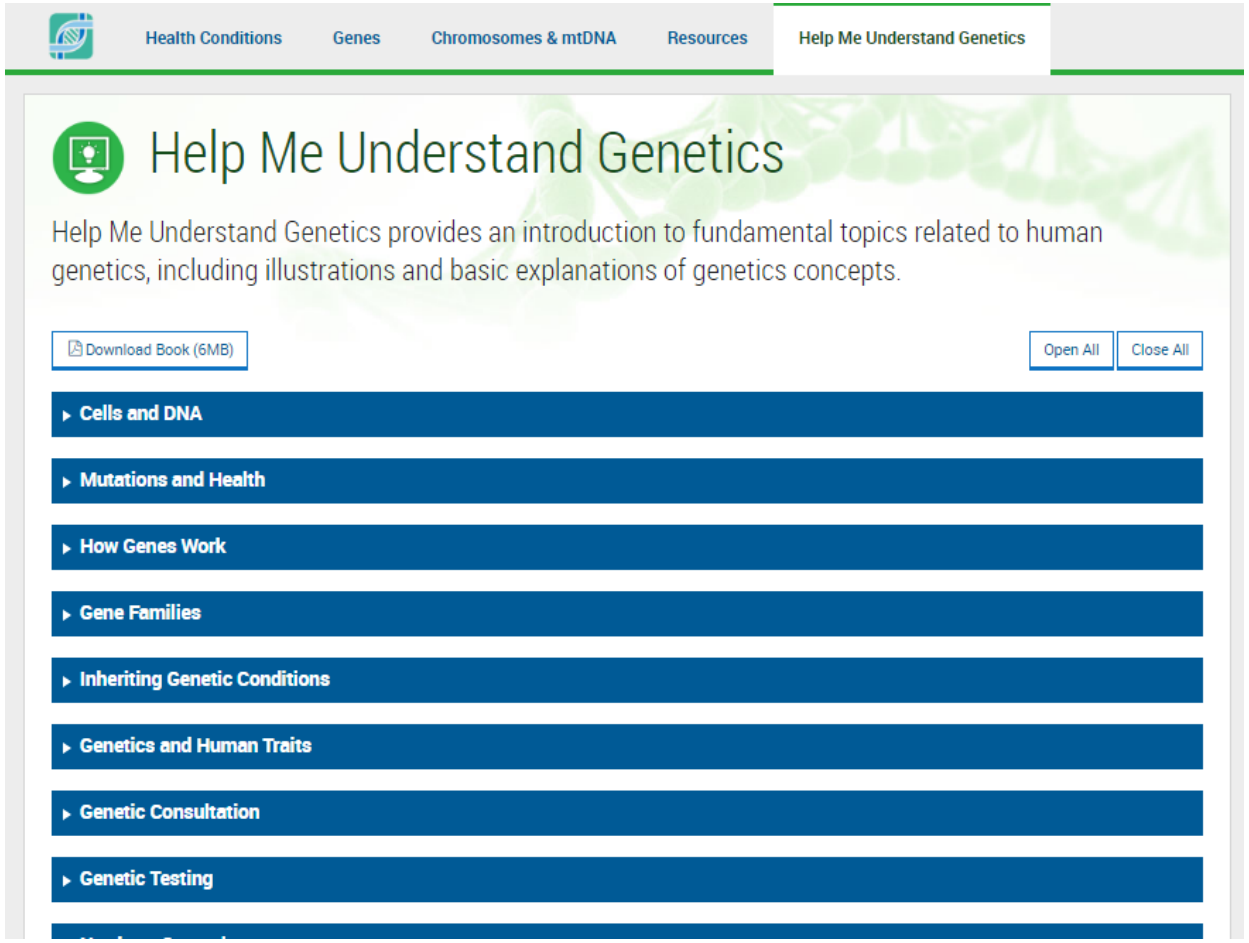
NHGRI

National Organization for Rare Disorders



Education Resources





National DNA Day- April 25

National DNA DAY APRIL 25 15th Anniversary

Celebrating Genomics Through Awareness

About DNA Day | Find Events | Celebrate With NHGRI | Get Activity Ideas | Get Starter Kit | Register An Event

Learn about the Celebration
15 for 15

National DNA Day
- 15 for 15 Celebration
  - April 05: DNA Sequencing
  - April 06: Human Genomic Variation
  - April 09: Cancer Genomics
  - April 10: Human Origins and Ancestry
- About DNA Day
- Celebrate with NHGRI
- Find Events
- Get Activity Ideas
- Get Starter Kit
- Register an Event

'15 for 15' Celebration

Fifteen ways genomics is now influencing our world

Whether you realize it or not, many parts of our daily lives are influenced by genomic infor
now provides a powerful lens for use in various areas - from medical decisions, to food saf

April 2018 will mark the 15th anniversary of the completion of the Human Genome Project.
genomic advances that have been made since 2003, the National Human Genome Resea
15' Celebration - unveiling 15 ways that genomics has and will continue to transform our w

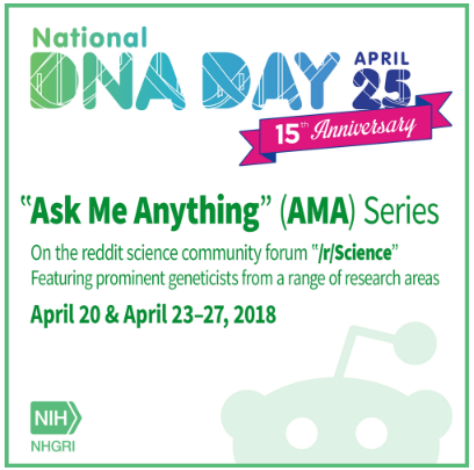
We hope that you will join us on this journey and learn how genomics is influencing the wo
you discover today?

Celebrate with NHGRI

- Bench to Bedside to Business
- Smithsonian Hot Topic - Lita Proctor, Ph.D.

The National DNA Day Reddit "Ask Me Anything" Series

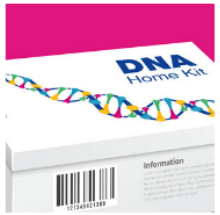
April 20 and April 23-27, 2018



The National Human Genome Research Institute (NHGRI) will launch the National DNA Day Reddit "Ask Me Anything" (AMA) Series on Friday, April 20, continuing each week day until Friday, April 27, 2018, from 1:00 - 3:00 p.m. Eastern. Genomics experts will answer questions at the Reddit Science community forum, "**/r/Science**". A Reddit AMA is an opportunity to ask interesting individuals questions about anything and everything.

Series Events

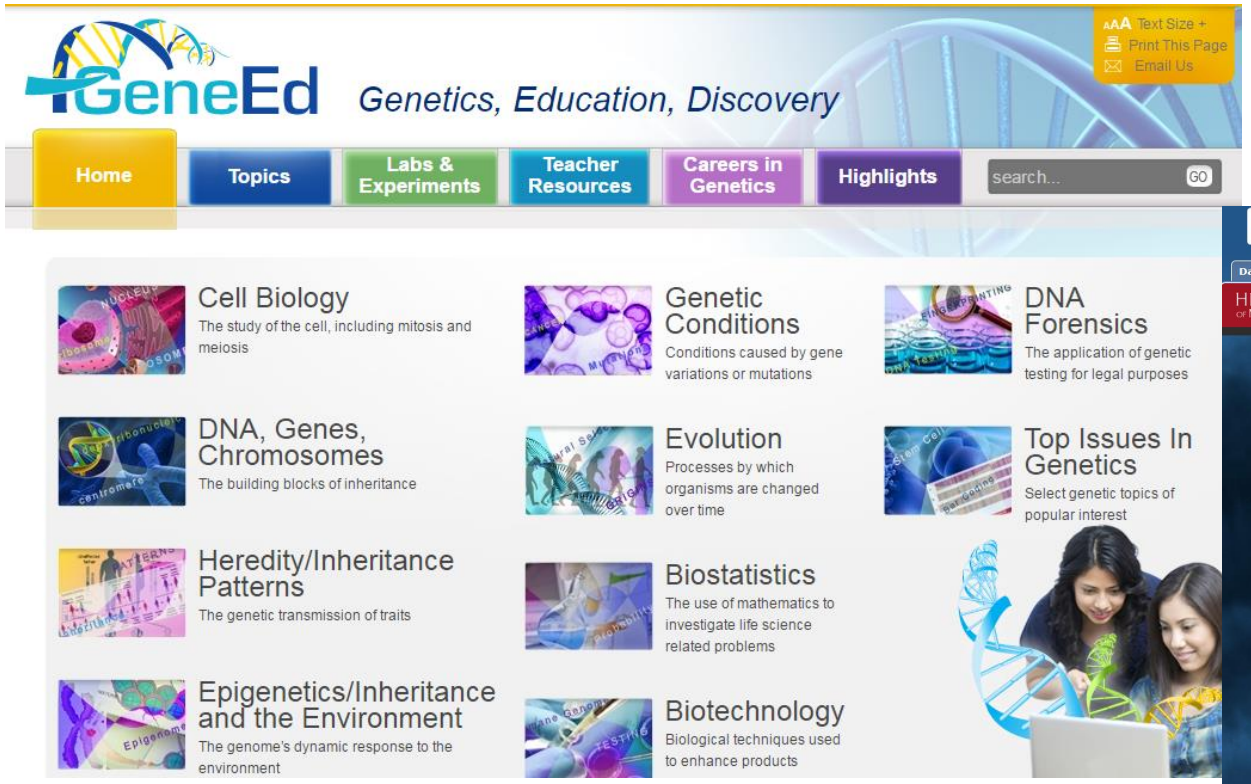
Friday, April 20, 2018
National Institutes of Health Director Francis Collins, M.D., Ph.D.
"The future of precision medicine"
The former director of the NHGRI, Dr. Collins earned a reputation as a gene hunter at the **University of Michigan** and subsequently lead the successful completion of the Human Genome Project 15 years ago. Now, in his current role as the director of the National Institutes of Health (NIH), Dr. Collins manages the NIH's efforts in building innovative enterprises, such as the **All of Us Research Program**. This AMA will focus on Dr. Collins' experiences during the Human Genome Project and how he envisions the future of precision medicine.



Monday, April 23, 2018
Representatives from Personal Genetics Companies
"Personal genetics and you"
Fifteen years after the completion of the Human Genome Project, we're now at a time when taking a detailed look at our genome can be sequenced as easily as ordering a kit online, spitting into a tube or swabbing the inside of the cheek and sending it off through the mail to a lab. Personal genetics companies are using these at-home, genetic-testing kits to help people access and understand their genome. The market for at-home genetic testing is ever-growing and can offer different aspects about what makes you, you! This AMA will answer your questions on what you can learn from your genome at home.

Tuesday, April 24, 2018
The Smithsonian Conservation Biology Institute's Center for Conservation Genomics researchers
Jesus Maldonado, Ph.D. and **Nancy Rotzel McInerney, B.S.**

K-12 Resources



[GeneEd]

[Harry Potter's World]

My Family Health Portrait
U.S. Surgeon General

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

HHS.gov
U.S. Department of Health & Human Services

About HHS | Programs & Services | Grants & Contracts | Laws & Regulations

Home > Programs > Prevention & Wellness > The Surgeon General's Family Health History Initiative

Family Health History

- About the Family Health History Initiative
- Before You Start Your Family Health History
- My Family Health Portrait Tool
  - English Web Tool
  - Printable Versions
  - Source Code

Text Resize A A A | Print | Share

The Surgeon General's Family Health History Initiative

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.

About the Family Health History Initiative

Learn about the Surgeon General's Family Health History Initiative

Before You Start

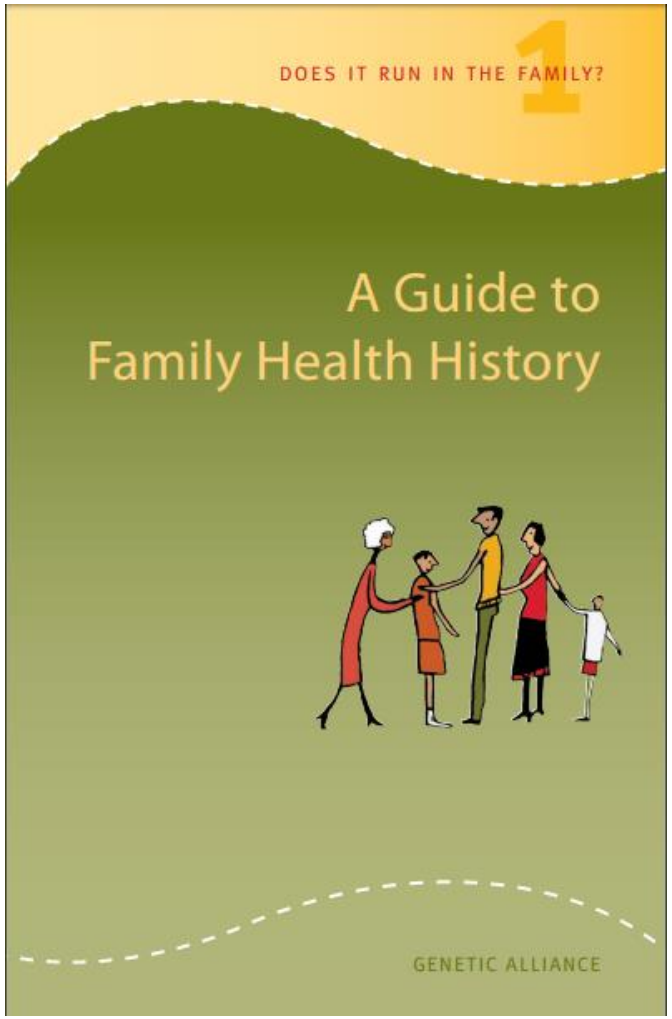
Find tips to help you begin gathering information about your family's health history.

My Family Health History Portrait Tool

Find out about this web-based tool to help you organize and print your family history

Surgeon General's Family Health History Initiative

Does It Run In the Family? Toolkit



Does it Run In the Family? toolkit

Literacy/Education Resources

pgEd Personal Genetics Education Project

ACMG Translating Genes Into Health®

ASHG THE AMERICAN SOCIETY OF HUMAN GENETICS discover. educate. advocate.

Genes in Life

GENOME UNLOCKING LIFE'S CODE

GENETIC LITERACY PROJECT SCIENCE TRUMPS IDEOLOGY

Learn.Genetics GENETIC SCIENCE LEARNING CENTER

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?
- 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

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?

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

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!

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GINA Help

H. R. 1313

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All Legislation | Examples: hr5, sres9, "health care"

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

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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

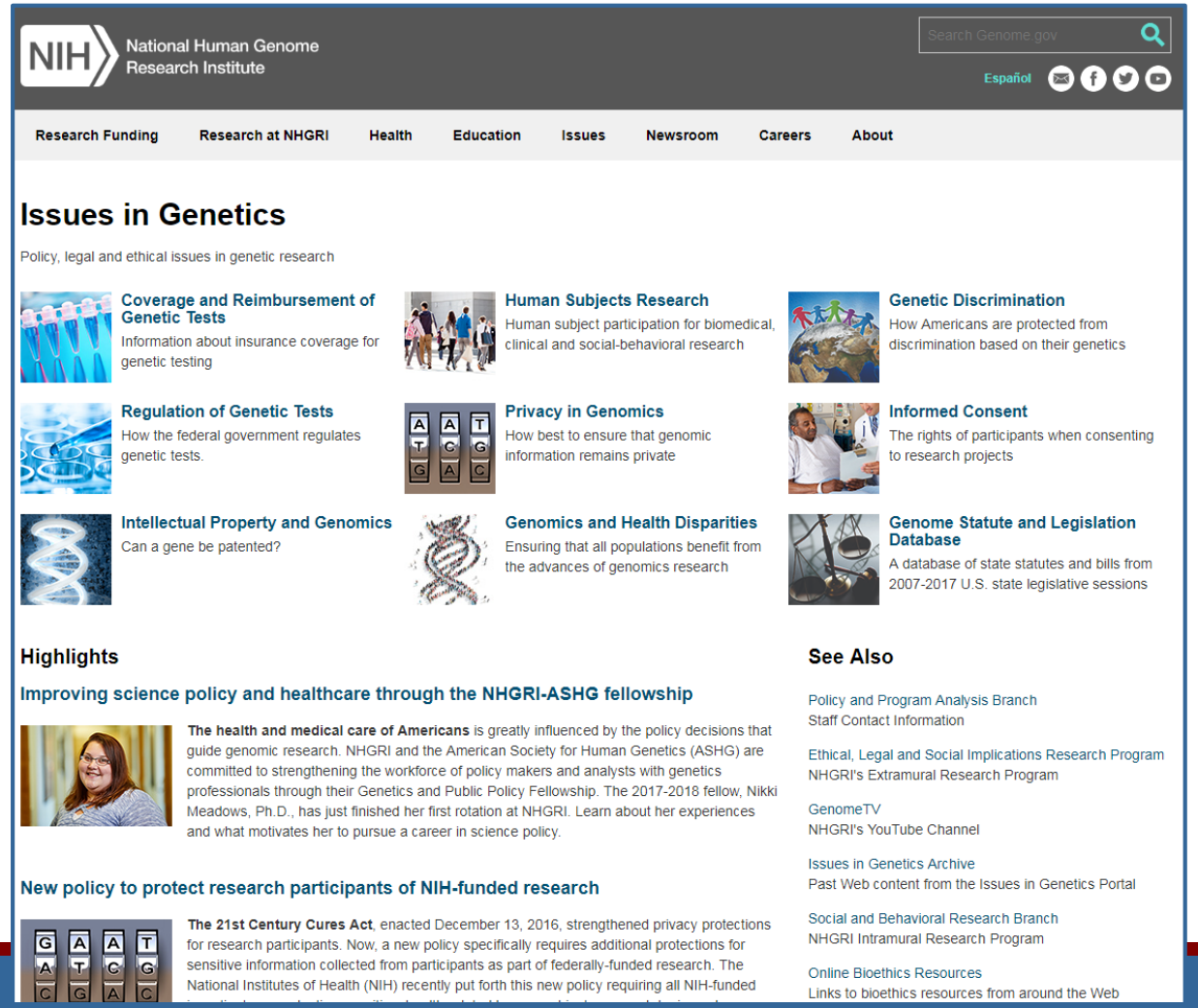
Kathy L. Hudson, Ph.D., and Karen Pollitz, M.P.P.

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

NIH National Human Genome Research Institute



NIH National Human Genome Research Institute

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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?

Genomics and Health Disparities
Ensuring that all populations benefit from the advances of genomics research

Genome Statute and Legislation Database
A database of state statutes and bills from 2007-2017 U.S. state legislative sessions

Highlights

Improving science policy and healthcare through the NHGRI-ASHG fellowship

The health and medical care of Americans is greatly influenced by the policy decisions that guide genomic research. NHGRI and the American Society for Human Genetics (ASHG) are committed to strengthening the workforce of policy makers and analysts with genetics professionals through their Genetics and Public Policy Fellowship. The 2017-2018 fellow, Nikki Meadows, Ph.D., has just finished her first rotation at NHGRI. Learn about her experiences and what motivates her to pursue a career in science policy.

New policy to protect research participants of NIH-funded research

The 21st Century Cures Act, enacted December 13, 2016, strengthened privacy protections for research participants. Now, a new policy specifically requires additional protections for sensitive information collected from participants as part of federally-funded research. The National Institutes of Health (NIH) recently put forth this new policy requiring all NIH-funded

See Also

Policy and Program Analysis Branch
Staff Contact Information

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

GenomeTV
NHGRI's YouTube Channel

Issues in Genetics Archive
Past Web content from the Issues in Genetics Portal

Social and Behavioral Research Branch
NHGRI Intramural Research Program

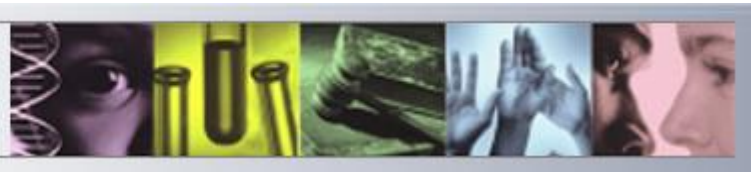
Online Bioethics Resources
Links to bioethics resources from around the Web

Informing the Public

CRG Council for Responsible Genetics

Genetic Alliance

Center for Genetics and Society



All of Us

1 MILLION + VOLUNTEERS

Precision Medicine

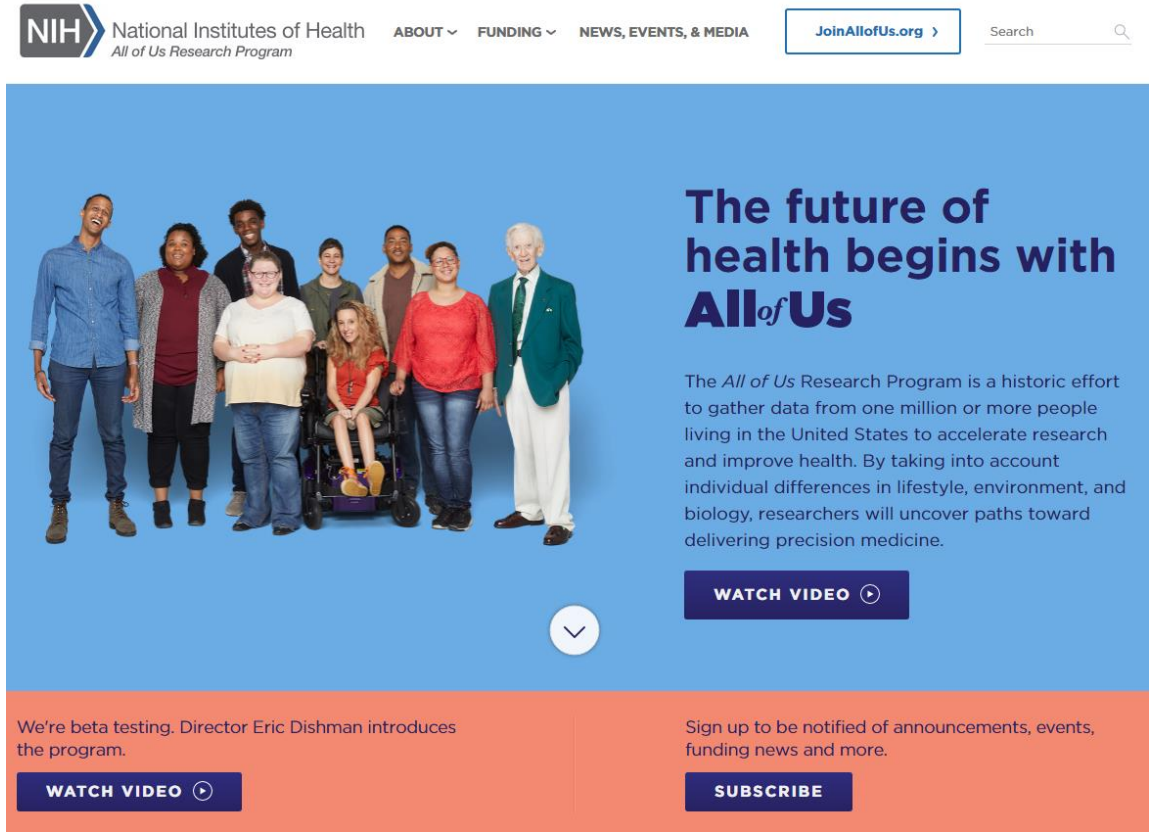
Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person.

Instead of what treatment is right for this disease it is what treatment is right for the patient.

Precision Medicine Initiative announcement

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.

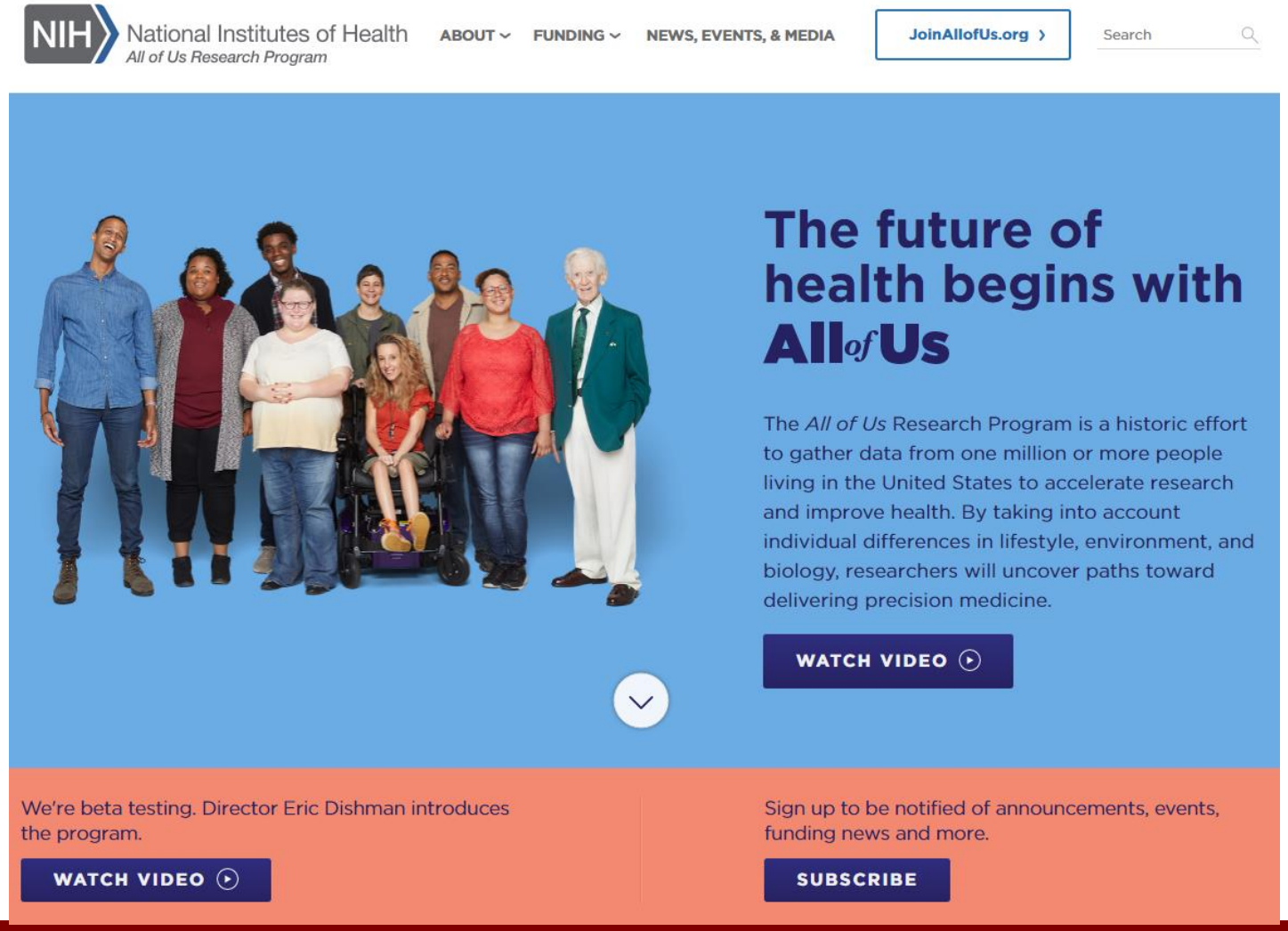


***All of Us* Research Program**

All of Us Research Program- video

[What is *All of Us*? video]

All of Us – more information



***All of Us* Research Program**

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"

Show What You Know!

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

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