HELPING PATRONS UNRAVEL THE MYSTERY OF GENETIC INFORMATION

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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
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

https://nnlm.gov/
Genetics in the News

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

Mail-Order CRISPR Kits Allow Absolutely Anyone to Hack DNA

Baltimore Ravens to hand out free DNA test kits

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

Unrelievable research can lead families to make health decisions they might regret

Genetic Testing for Athletic Ability

Can genes predict sporting talent?

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

Genetically Modified Humans?

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

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
8. Influenza and pneumonia: 57,062
9. Nephritis, nephrotic syndrome, and nephrosis: 49,959
10. Intentional self-harm (suicide): 44,193
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 Tests Can Help to:

1. Diagnose Your Disease
2. Pinpoint Genetic Factors That Caused Your Disease
3. Predict How Severe Your Disease Might Be
4. Choose the Best Medicine and Correct Dose
5. Discover Genetic Factors That Increase Your Disease Risk
6. Find Genetic Factors That Could Be Passed to Your Children
7. Screen Newborns for Certain Treatable Conditions
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
DTC BRCA test

FDA announcement

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.[10]

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,
Genetic Testing - is it necessary?

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

Various genetic testing services listed

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

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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 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 testing.

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
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.20

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 States.
American College of Medical Genetics and Genomics

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

ACMG Board of Directors

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 acmginacmg.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 counseling.

- 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 adequate risk 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

- Section: Genetics/Birth Defects
- Health Topic pages:
  - Genetics
  - Genetic testing
  - Genetic counseling
  - Genetic disorders
  - Genetic brain disorders
  - Genes and gene therapy
  - text word search
# 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 Triangularis
- Birth Defects
- Blood Coagulation Disorders see Hemophilia
- Brain Disorders, Inborn Genetic see Genetic Brain Disorders
- Brain Malformations
- Caravan Disease see Leukodystrophies
- Cephalic Disorders see Brain Malformations
- Cerebral Palsy
- Charcot-Marie-Tooth Disease
- Chiari Malformation
- Chronic IlliSampling see Prenatal Testing
- Cleft Lip and Palate
- Cleft Palate see Cleft Lip and Palate
- Cleft Spine see Spina Bifida
- Cloning
- Color Blindness
- Congenital Heart Defects
MedlinePlus – stroke topic page

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 floods into the brain. "Mini-stroke" 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

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)
Textword search ‘genetics’
Genetics Home Reference

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

Help Me Understand Genetics provides an introduction to fundamental topics related to human genetics, including illustrations and basic explanations of genetics concepts.

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

Help Me Understand Genetics

Help Me Understand Genetics provides an introduction to fundamental topics related to human genetics, including illustrations and basic explanations of genetics concepts.

See Also
- Education and Community Involvement Branch
- Genome TV
- Genome Alliance of the Americas
- Education Kit

- Cells and DNA
- Mutations and Health
- How Genes Work
- Gene Families
- Inheriting Genetic Conditions
- Genetics and Human Traits
- Genetic Consultation
- Genetic Testing
National DNA Day - April 25
K-12 Resources

GeneEd

Harry Potter’s World
My Family Health Portrait
U.S. Surgeon General

Surgeon General’s Family Health History Initiative
Does It Run In the Family? Toolkit
Literacy/Education Resources
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?
H. R. 1313

Preserving Employee Wellness Programs Act

Summary: H.R.1313 — 116th Congress (2017-2018)

This bill amends the tax treatment of wellness programs. Specifically, it:

(1) Clarifies that provisions of the Patient Protection and Affordable Care Act (PPACA) are applicable to wellness programs.

(2) Requires the Internal Revenue Service to provide guidance on the tax treatment of wellness programs.

There is one summary for H.R.1313. Bill summaries are authored by CBO.

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
Informing the Public
All of Us

1 MILLION + VOLUNTEERS
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.
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 - video

What is All of Us? video
The future of health begins with All of Us

The All of Us Research Program is a historic effort to gather data from one million or more people living in the United States to accelerate research and improve health. By taking into account individual differences in lifestyle, environment, and biology, researchers will uncover paths toward delivering precision medicine.

WATCH VIDEO

We're beta testing. Director Eric Dishman introduces the program.

All of Us Research Program
“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.”
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!

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