HELPING PATRONS UNRAVEL THE MYSTERY OF GENETIC INFORMATION

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NNL....Huh?

**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
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

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

Unrelieved 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

CDC FastStats
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
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**

**23andMe announcement**
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
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 (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 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
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. If 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.[50]

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
Genetic Testing

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 cause of a medicine that is best for a certain person

People have many different weapons 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.
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 genetic 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 exclusion 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 medical 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 assessment and a thorough understanding of the implications of results. The ACMG emphasizes the importance of genetic counseling and recommends that genetic counselors and other health-care professionals provide genetic counseling to persons before and after genetic testing. Genetic counselors should be available to help consumers 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 risk of informed consent...
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
Section: Genetics/Birth Defects

Health Topic pages:
- Genetics
- Genetic testing
- Genetic counseling
- Genetic disorders
- Genetic brain disorders
- Genes and gene therapy

text word search
# MedlinePlus – Genetics topics

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MedlinePlus – stroke topic page

Stroke
Also called: Brain attack, CVA

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-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

Health Check Tools
- Test Your Stroke Knowledge
- What's Your Stroke Risk?

Videos and Tutorials
- Know Stroke: Know the Signs, Act in Time Video

Statistics and Research
- FastStats: Cerebrovascular Disease or Stroke
- Heart Disease and Stroke Statistics
- Preventing Stroke Deaths

Clinical Trials
- ClinicalTrials.gov: Carotid Stenosis
- ClinicalTrials.gov: Cerebrovascular Disorders

Genetics
- Genetics Home Reference: Cerebral autosomal dominant arteriopathy with subcortical infarcts and leuкоencephalopathy
- Genetics Home Reference: Grange syndrome
- Genetics Home Reference: mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes
- Genetics Home Reference: moyamoya disease

Images
- Carotidotomy - slideshow (Medical Encyclopedia)
Textword search ‘genetics’
Genetics Home Reference

- Health conditions
- Genes
- Chromosomes and DNA
- Resources
- Genetic handbook (Help Me Understand Genetics)
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.
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?
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.
H. R. 1313

Preserving Employee Wellness Programs Act

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

PMID: 28537794
NIH National Human Genome Research Institute

Issues in Genetics
Policy, legal and ethical issues in genetic research

- Coverage and Reimbursement of Genetic Tests: Information about insurance coverage for genetic testing
- Regulation of Genetic Tests: How the federal government regulates genetic tests
- Intellectual Property and Genomics: Can a gene be patented?
- Human Subjects Research: Human subject participation for biomedical, clinical and social behavioral research
- Privacy in Genomics: How best to ensure that genomic information remains private
- Genetic Discrimination: How Americans are protected from discrimination based on their genetics
- Informed Consent: The rights of participants when consenting to research projects
- 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 2001-2017 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 shape 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, Nelia Meadow, 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...
Informing the Public

CRG - Council for Responsible Genetics

Genetic Alliance

Center for Genetics and Society
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.

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 - 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.

Sign up to be notified of announcements, events, funding news and more.

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!

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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