HELPING PATRONS UNRAVEL THE MYSTERY OF GENETIC INFORMATION

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Genetics Overview

Genomic Health Literacy

Genetic Testing

Consumer Health Resources

Societal Issues

All of Us Research Program
NNLM...?

NIH
- National Institutes of Health
- Nation’s research agency

NLM
- National Library of Medicine
- World’s largest biomedical library

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

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

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

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

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

Afraid of Needles? You May Want to Blame Your Genes

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

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

CRISPR Gene Editing Is Being Tested in Human Patients, and the Results Could Revolutionize Health Care

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

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

• Lack biology basics
• Lack mathematical concepts
• Low health literacy
Leading causes of death

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

CDC FastStats
The Story of You
Genetic Testing

including Direct-to-Consumer
Clinical Uses of Genetic Tests

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

- Diagnostic
- Predictive
- Carrier
- Prenatal
- Newborn Screening
- Research
- Pharmacogenetic
Jean’s Genetic Testing Timeline

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

What genes and what variants did you test for?
- Different tests offered for the same conditions
- Knowledge is always changing

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

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

Possibility of false positives and false negative results.
Genetic Testing- is it necessary?

Before testing:

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

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

National Society of Genetic Counselors
Direct to Consumer Testing
Testing for talent

Map My Gene
Create the Champion in Your Child through DNA Mapping

ORIG3N
SUPERHERO
DNA Test

KarmaGenes
meet yourself

Genomic Express
your natural ability through personalized genomics.
DNA dating
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 testing for the evaluation of patients and individuals. They are based on the best information available at the time of publication. Any individual should apply their own professional judgment to the specific clinical circumstances presented by the individual patient or family. 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, revised, 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 complete process that includes genetic health professionals and patients. Clinicians should apply their own professional judgment to the specific clinical circumstances presented by the individual patient or family. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from the recommendations.

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 underestimating risk of inherited cancer.
MedlinePlus

FEDERAL TRADE COMMISSION

CONSUMER INFORMATION

Direct-to-Consumer Genetic Tests

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

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

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

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

Direct-to-Consumer Genetic Testing

What is direct-to-consumer genetic testing?
What kinds of direct-to-consumer genetic tests are available?
What is genetic ancestry testing?
What are the benefits and risks of direct-to-consumer genetic testing?
How do I choose a direct-to-consumer genetic testing company?
How is direct-to-consumer genetic testing done?
How much does direct-to-consumer genetic testing cost, and is it covered by health insurance?
What do the results of direct-to-consumer genetic testing mean?
What can raw data from a direct-to-consumer genetic test tell me?
Can a direct-to-consumer genetic test tell me whether I will develop cancer?
Can a direct-to-consumer genetic test tell me whether I will develop Alzheimer disease?
What does it mean to have Ashkenazi or Sephardic DNA?
How do direct-to-consumer genetic testing companies protect their customers' privacy?
Can the results of direct-to-consumer genetic testing affect my ability to get insurance?
Where can I read more about the diseases and traits covered in my direct-to-consumer genetic testing report?

What is direct-to-consumer genetic testing?

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

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

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

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

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

▪ Learn more about own health
▪ Learn more about ethnicity and family history
▪ Bring awareness to family health issues for future generations
▪ Motivation to work on health habits
▪ Encourages patient engagement
▪ Contributing to advancement of healthcare and science
▪ Moral obligation
Questions to ask before using a Direct to Consumer Genetic Test

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

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

Consumer 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
# Genetics/Birth Defects

- Abnormalities see Birth Defects
- Achondroplasia see Dwarfism
- Adrenoleukodystrophy see Leukodystrophies
- Alpha-1 Antitrypsin Deficiency
- Amniocentesis see Prenatal Testing
- Anencephaly see Neural Tube Defects
- Arnold-Chiari Malformation see Chiari Malformation
- Ataxia see Friedreich's Ataxia
- Ataxia Telangiectasia
- Birth Defects
- Blood Coagulation Disorders see Hemophilia
- Brain Disorders, Inborn Genetic see Genetic Brain Disorders
- Brain Malformations
- Canavan Disease see Leukodystrophies
- Cephalic Disorders see Brain Malformations
- Cerebral Palsy
- Charcot-Marie-Tooth Disease
- Chiari Malformation
- Chorionic Villi Sampling see Prenatal Testing
- Cleft Lip and Palate
- Cleft Palate see Cleft Lip and Palate
- Cleft Spine see Spina Bifida
- Cloning
- Color Blindness
- Congenital Heart Defects
MedlinePlus – text search

Text word search ‘genetics’
Genetics Home Reference

- Health conditions
- Genes
- Chromosomes and DNA
- Classroom Resources
- Help Me Understand Genetics (a handbook)
Genetics Home Reference - health conditions
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, 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.
Help Me Understand Genetics

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 Groups
- Inheriting Genetic Conditions
- Genetics and Human Traits
- Genetic Consultation
- Genetic Testing
NIH National Human Genome Research Institute - health information
National Organization for Rare Disorders

7,000 RARE DISEASES AFFECT 30 MILLION AMERICANS. HOW CAN YOU HELP? #DOYOURSHARE

DoYourShare.com

What’s happening at NORD

Tools and Resources

NORD

NIH U.S. National Library of Medicine
National Network of Libraries of Medicine
Pacific Northwest Region
NHGRI Education Resources

Introduction to Genomics

Educational Resources

What's a Genome?

Genome is a fancy word for all your DNA. From potatoes to puppies, all living organisms have their own genome. Each one contains the information needed to build and maintain that organism throughout its life.

These educational resources are intended to spark scientific curiosity, improve genomic literacy and foster engagement among learners.

Fact Sheets about Genomics

Talking Glossary of Genetic Terms

Genomics Teaching Tools
National DNA Day – April 25
Genetics Home Reference - Classroom
K-12 Resources

American Society of Human Genetics

Harry Potter’s World
My Family Health Portrait

Using My Family Health Portrait you can:
• Enter your family health history.
• Learn about your risk for conditions that can run in families.
• Print your family health history to share with family or your health care provider.
• Save your family health history so you can update it over time.

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

Learn more about My Family Health Portrait.

Create a Family Health History  Use a Saved History

CDC Family Health Portrait
Does It Run In the Family? Toolkit

A Guide to Family Health History

Does it Run In the Family? toolkit
Literacy/Education Resources
Societal Issues
Privacy, Security, Ethical, and Societal Issues
Societal Concerns

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

PMID: 28537794
Usage by law enforcement

AMERICA

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

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

Office of Inspector General, Fraud Alert

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

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

Scammers are offering Medicare beneficiaries “free” screenings or cheek swabs for genetic testing to obtain their Medicare information for identity theft or fraudulent billing purposes. Fraudsters are targeting beneficiaries through telemarketing calls.
NIH National Human Genome Research Institute

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

NHGRI Policy Issues
Informing the Public
Precision Medicine

All of Us Research Program
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
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
Library role

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

“What Does it Mean to be Genomically Literate? National Human Genome Research Institute Meeting Report”
Libraries and genetic health information
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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