HELPING PATRONS UNRAVEL THE MYSTERY OF GENETIC INFORMATION

Carolyn Martin, MLS, AHIP
Consumer Health Coordinator
National Network of Libraries of Medicine
Pacific Northwest Region (NNLM PNR)
martinc4@uw.edu
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 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
Genetics in the News

Human Gene Editing Receives Science Panel’s Support

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

Mail-Order CRISPR Kits Allow Absolutely Anyone to Hack DNA

Baltimore Ravens to hand out free DNA test kits

Scientists Use Genetic Engineering To Vanquish Disease-Carrying Insects

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

Genetically Modified Humans?

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

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

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

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

1. Heart disease: 633,842
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

The Story of You: ENCODE and the human genome
Genetic Testing

INCLUDING DIRECT-TO-CONSUMER
Clinical Uses of Genetic Tests

Genetic Tests Can Help to:

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

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

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

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

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
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 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 revisited its 2008 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, family history, and potential health outcomes. It is critical for consumers to be informed about the potential risks, benefits, and limitations of genetic testing.

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

Most of the time, genetic testing is done through healthcare providers such as physicians, nurses, practitioners, and genetic counselors. Healthcare providers determine which tests are needed, order the test from a laboratory, collect and 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 little regulation of 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 genetic testing, direct-access genetic testing, at-home genetic testing, and home DNA testing. Ancestry testing (also called genealogy testing) is also considered a form of direct-to-consumer genetic testing.

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

- National Society of Genetic Counselors: What is At-Home Genetic Testing?
- American Medical Association: Consumer Genetic Testing
- The Federal Trade Commission: Consumer Genetic Tests
- Genetics in Life: Direct-to-Consumer Genetic Testing
- Johns Hopkins Medicine: Five Things to Know about 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?
- Can I ask what 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’s disease?
- What does it mean to have Neanderthal or Denisovan 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?
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

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

Health 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
- Cerebral Palsy
- Choroid Plateau-Plate Disease
- Chiari Malformation
- Chorionic Villus 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

Drugs & Supplements

Videos & Tools
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 I.Q.? (American Heart Association)

Videos and Tutorials
- Know Stroke: Know the Signs, Act in Time Video
- Get Stroke updates by email

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

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
MedlinePlus – text search

Text word search ‘genetics’
Genetics Home Reference

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

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 visuospatial 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 subaortic stenosis (SVAS) occurs frequently in people with Williams syndrome. Subaortic 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 - resources
Genetics Home Reference- handbook
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

News
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.

- 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

Fifteen ways genomics is now influencing our world

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

April 2010 marks the 15th anniversary of the completion of the Human Genome Project. The Human Genome Project's 15th anniversary has the potential to transform our understanding of human health and disease.

We hope that you will join us on this journey and learn how genomics is influencing the world as you discover today.

Learn about the Celebration

Celebrate 15 for 15

The National DNA Day Reddit "Ask Me Anything" Series

April 20 and April 23-27, 2016

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 weekday until Tuesday, April 27, 2016, from 1:00 - 3:00 p.m. Eastern. Genomics experts will answer questions at the Reddit Science community forum.

"Ask Me Anything" (AMA) Series

On the Reddit science community forum "r/Science"

Featuring prominent genomics from a range of research areas

April 20 & April 23-27, 2016

"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 led 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 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 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 Waldron, Ph.D. and Nancy Rottet Moneymaker, B.S.
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
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

GINA

GENETIC INFORMATION NONDISCRIMINATION ACT

What is genetic information and why is it important?

What are GINA’s health insurance protections?

What are GINA’s employment protections?

What is GINA?
The Genetic Information Nondiscrimination Act of 2008 (GINA) is a federal law that protects individuals from genetic discrimination in health insurance and employment. Genetic discrimination is the misuse of genetic information. This resource provides an introduction to GINA and its protections in health insurance and employment. It includes answers to common questions and examples to help you learn. Choose from one of the boxes to the left to begin!

Have questions, comments or suggestions? Send us a note.

Click here for a printer friendly version.

For healthcare provider resources click here.

Click here for the GINA & You Information Sheet.

Permission is granted under the copyright for educational, non-commercial use of these materials. Other uses require permission of the copyright holders.

© DESIGN & DEVELOPMENT BY INNO PROJECTS, INC. 2011

GINA Help
H. R. 1313

Preserving Employee Wellness Programs Act

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

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

Introduced: Mar 20, 2017

Preserving Employee Wellness Programs Act

This bill enforces workplace wellness programs that: (1) is not motivated by a disease or health risk factor, and (2) does not provide financial or other incentives for health improvement. The bill would also prohibit workplace wellness programs that discriminate based on genetic information.

The NEW ENGLAND JOURNAL OF MEDICINE

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
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.
NIH National Human Genome Research Institute
Informing the Public
All of Us

1 MILLION + VOLUNTEERS
Precision Medicine Initiative announcement

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
All of Us – more information

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.

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!

Carolyn Martin, MLS, AHIP
NNLM PNR
martinc4@uw.edu