ABCs of DNA:
Unraveling the Mysteries of Genetics
Information for Consumers

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Agenda

- Basic genetics overview
- Genomic health literacy & genomic science literacy
- Genetic testing including direct-to-consumer
- Genetic consumer health resources
- Ethics and privacy
- Precision Medicine Initiative

Presentation resources
https://nnlm.gov/pnr/guides/training-resources-you-can-use/presentations
Who We Are

NIH
NLM
NNLM

What’s the difference?
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 PNR
https://nnlm.gov/pnr
News Headlines

*Human Gene Editing Receives Science Panel’s Support*

Scientists say they hope to create a human genome in the lab. Humans will be 'irrevocably altered' by genetic editing, warn scientists ahead of summit.

An open letter from campaigners and health experts is calling ahead of a summit in Washington.

*Genes Plus Erratic Sleep May Raise Odds for Obesity*

How much study you get could tip the scale if you're predisposed to gain weight, researchers say.

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

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

*The Genetic Tool That Will Modify Humanity*

Crispr allows scientists to control the DNA worse.

*In Big Shift, 23andMe Will Invent Drugs Using Customer Data*

Your DNA may determine how you handle the time change.

*How Genome Editing Works*

"Improving" humans with customized genes sparks debate among scientists.

*Genetically Modified Humans?*

"Improving" humans with customized genes sparks debate among scientists.

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

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

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

1. Heart disease: 614,348
2. Cancer: 591,699
3. Chronic lower respiratory diseases: 147,101
4. Accidents (unintentional injuries): 136,053
5. Stroke (cerebrovascular diseases): 133,103
6. Alzheimer's disease: 93,541
7. Diabetes: 76,488
8. Influenza and pneumonia: 55,227
9. Nephritis, nephrotic syndrome, and nephrosis: 48,146
10. Intentional self-harm (suicide): 42,773
The Story of You
https://www.youtube.com/watch?v=TwXXgEz9o4w
CATEGORIES OF DISEASES ATTRIBUTED TO GENES

- Chromosomal Diseases
- Monogenic Diseases
- Multifactorial Diseases
Genetic Testing

including Direct-to-Consumer
Clinical Uses of Genetic Tests

[Image: Genetic Testing image from Genome.gov
https://www.genome.gov/images/content/genetic_testing.jpg]
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 testing** to investigate ancestry

Age 65: **pharmacogenomics testing** when Plavix (anti-platelet drug) was not 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?

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?

Choosing Wisely
http://consumerhealthchoices.org/catalog/making-smart-decisions-about-genetic-testing-acmg/
Direct to Consumer Testing

welcome to you®

23andMe

GENE By GENE

ancestry.com®

The Human Story
Join The Project To Learn About Your Story

Since its launch in 2005, National Geographic's Genographic Project has used advanced DNA analysis and evolved with Indigenous communities to help answer fundamental questions about where humans originated and how we came to populate the Earth. Now, cutting-edge technology is enabling us to shine a powerful new light on our collective past. By participating in the latest phase of this real-time scientific project, you can learn more about yourself than you ever thought possible.
Genomic Testing- Athletic Ability

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

- Privacy
- Legality
- Who has access?
- How useful now?
- 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
- Who can get the testing?
Benefits

- Learn more about own health
- More effective medical treatments
- 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.

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.
MedlinePlus

Genetic Testing

Summary
Genetic tests are tests on blood and other tissue to find genetic disorders. Over 2000 tests are available. Doctors use panels tests for several reasons. Some include:

- Finding genetic diseases in unborn babies
- Detecting 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 it 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 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
Consumer Resources
MedlinePlus

- Section: Genetics/Birth Defects (and others)
- Health Topic pages:
  - Genetics
  - Genetic testing
  - Genetic counseling
  - Genetic disorders
  - Genes and gene therapy
- text word search

MedlinePlus https://medlineplus.gov/
Genetics Home Reference

- Health conditions
- Genes
- Chromosomes and DNA
- Resources
- Genetic handbook

Genetics Home Reference
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Genetics Home Reference

Each of the following categories provides links to useful genetics resources on the web.

- Support and Advocacy
- Financial Assistance
- General Genetics
- Genetic Testing
- Classroom Resources
- Clinical/Professional Resources
- Genetics Research
- Bioinformatics Databases
- Genetics News
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
NIH National Human Genome Research Institute

[Website Screenshot]

NHGRI https://www.genome.gov/
My Family Health Portrait
U.S. Surgeon General

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 Health History Initiative, to encourage all American families to learn more about their family health history.

Surgeon General’s Family Health History Initiative:
Does it Run In the Family? toolkit

Education Resources

GeneEd
https://geneed.nlm.nih.gov/

National Human Genome Research Institute
https://www.genome.gov/education/
Harry Potter’s World

More 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?
- What are the larger societal issues raised by new reproductive technologies?
- How will genetic tests be evaluated and regulated for accuracy, reliability and utility?
- How do we prepare healthcare professionals and the public?
- What is considered acceptable diversity?
- Where is the line between medical treatment and enhancement?
- Should testing be performed when no treatment is available?
- Who can afford genetic testing?
GINA

[GINA Help](http://ginahelp.org/)
Informing the Public
Precision Medicine

“...a bold new research effort to revolutionize how we improve health and treat disease.”
Mission statement:

To enable a new era of medicine through research, technology, and policies that empower patients, researchers, and providers to work together toward development of individualized care.
Precision Medicine is...

- 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 this patient.
Precision Medicine Initiative

Near Term goals:
- Clinical trials focusing on pediatric cancers and drug therapies for adults
- Use of combination therapies
- Overcoming drug resistance

Long Term Goals:
- Create research cohort of 1 million volunteers
- New model of medicine
  - engage participants
  - responsible data sharing
  - privacy protection
- Advance pharmacogenomics
- Identify new targets for treatment and prevention
- Test if mobile devices encourages healthy behaviors
- Lay scientific foundation for many diseases

All of Us infographic:
All of Us Research Program

Two ways to participate:

1. Through the participant website
2. With participating Healthcare Provider Organizations (HPOs)
NIH and Precision Medicine Initiative

All of Us Research Program:
https://www.nih.gov/research-training/allofus-research-program
Health Care Tailored to You
Adventures in Precision Medicine: A Major Public Research Initiative and its Implications for Healthcare Consumers and Institutions
September 21, 2016

Presenter: Malia Fullerton, Associate Professor of Bioethics and Humanities at the University of Washington School of Medicine
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”
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4115323/
Show What You Know!

• What initiative refers to strategies for determining what treatment is right for an INDIVIDUAL rather than what treatment is recommended for a DISEASE?

• What is the name of the volunteer research program that is looking to collect data on 1 million volunteers to assist with the Precision Medicine Initiative?

• True or False? GINA (Genetic Information Nondiscrimination Act) protects you from life insurance discrimination.

• What resource would you recommend to consumers who wanted to learn more about a genetic testing?
Questions?

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**Presentation resources**
https://nnlmgov/pnr/guides/training-resources-you-can-use/presentations