GENOMICS: WHY SHOULD WE CARE?

The importance of being: genomic literate
    genomic informed
Health Literacy
Orphan Black

Orphan Black video
News Headlines

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

The Genetic Tool That Will Modify Humanity

Humans will be 'irrevocably altered' by genetic editing, warn scientists ahead of summit
An open letter from 150 scientists, campaigners and health experts is calling for a worldwide ban on genetic editing ahead of a summit in Washington

Clinical Genetics Has a Big Problem That's Affecting People's Lives
Unreliable research can lead families to make health decisions they might regret.

British Scientists Seek Permission To Edit DNA In Human Embryos

Genetically Modified Humans? How Genome Editing Works

Having a baby? Best to run a gene screening test
Most physicians have no formal training in genetics

Little research has focused on their understanding of the recent developments in genomics

Patients ill-informed, inflated expectations, little scientific evidence regarding clinical utility of genomic interventions

Health professionals have not always kept up to date with the genetic advances

Need to improve medical education and beyond initial training

PMID 19341496 http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2664958/
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
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.
Genomic literacy

- Genomic health literacy
- Genomic science literacy
- Role of media in genomic literacy
Genomic Health Literacy

- Lack biology basics
- Lack mathematical concepts
- Low health literacy
Genomic Science Literacy

- K-12 education unable to keep up with scientific advancements
- Low emphasis on genomics
- Some teachers have misconceptions about genetics/genomics and little understanding
- Teachers need updated skills and have little access to genetic/genomic quality science curriculum
- Encourage partnerships with scientists
Media role in genomic literacy

- Scientists lack training when communicating with media and general public
- Popular and mass media lack knowledge and often relay incorrect information
- Educators and researchers need to adapt and learn to inform through newer media platforms such as social media and podcasts
Genomic discoveries will increasingly advance the science of medicine. Limited genomic literacy may adversely impact the public’s understanding and use of the power of genetics and genomics in health care and public health. In November 2011, a meeting was held by the National Human Genome Research Institute to examine the challenge of achieving genomic literacy for the general public, from K-12 to adult education. The role of the media in disseminating scientific messages and in perpetuating, or reducing, misconceptions was also discussed. Workshop participants agreed that genomic literacy will only be achieved through active engagement between genomics experts and the varied constituencies that comprise the public.
National Coalition for Health Professional Education in Genetics (NCHPEG)
NLM GeneEd

NCBI Bookshelf

Genes and Diseases

- Organized by the parts of the body that genetic disorders affect
- Over 80 genetic disorder summaries
- Images and interesting facts
- PDF downloads of chapters
- Links to related research literature and pertinent websites

Genes and Diseases
http://www.ncbi.nlm.nih.gov/books/NBK22183/
Literacy/Education Resources

- CDC Centers for Disease Control and Prevention
- ACMG Translating Genes Into Health®
- Public Health Genomics
- ASHG American Society of Human Genetics
- PGEd Personal Genetics Education Project
- Genetics in Life
- Genomic Literacy Project: Science Trumps Ideology
Tips for communicating to patients

- Listen, pay attention, respond
- Use plain language
- Use patient’s words
- Slow down
- Limit and repeat content
- Show examples
- Invited patient participating
- Use teach back

Health Literacy and Precision Medicine
Tips for communicating to healthcare professionals

- Bring a family member or a friend
- Write down or record the information
- Speak your mind
- Repeat the information as you understand it
- Ask questions (who, what, where, why, how)
- Ask for more information (website, printed handout, a library)
My Family Health Portrait, A tool from the Surgeon General

My Family Health Portrait is an Internet-based tool that makes it easy to create your family’s health history. It is simple to fill out. It is private. It is valuable health information that you can share with family members, for their benefit, and with your healthcare practitioner, for your better health.

My Family Health History is available at:
https://familyhistory.bhs.gov/fhh-web/home.action

Using My Family Health Portrait, you can:
• Record your family’s health history
• Print out and share the history with your family and your healthcare provider
• Save and regularly update your family health history for future use

Why is it important to know my family medical history?

Your family medical history is a record of health information about you and three generations of close relatives. Family history can be an important risk factor for problems like heart disease, stroke, diabetes, and cancer. A risk factor is anything that increases your chance of getting a disease. The reason a family history can help predict risk is that families share their genes, as well as other factors that affect health, like environment, lifestyles, and habits. A family medical history allows you to take steps to reduce your risk.

To Find Out More

MedlinePlus: Family History
www.nlm.nih.gov/medlineplus/familyhistory.html

Family Health History
www.cdc.gov/genomics/famhistory/index.htm
My Family Health Portrait

Find out how learning your family's health history can help you discover your genetic heritage and risks, and guide you in making healthy environment and lifestyle choices. Learn about how to obtain and create a family health history.

- My Family Health Portrait
- Family History: Resources and Tools [cdc.gov]
- Family Health History Tools [geneticalliance.org]
- NHGRI:SeniortHealth: Creating a Family Health History [nihseniorhealth.gov]

NHGRI My Family Health Portrait https://www.genome.gov/27527640/family-history-my-family-health-portrait/
My Family Health History

Update My Family History

On this screen you can:

- Use the tools to view your family tree diagram, find out your risks for certain diseases, or export your tree to help your close family members get started.
- Change your Family Health History by adding, removing or changing your relatives.
  - Add information for a family member by pressing the Add History button next to the name in the list.
  - Change your information or a family member’s information by pressing the Update History button next to the name in the list.
  - Remove a family member from your history by pressing the Remove button next to the name in the list. (You cannot remove yourself, your parents, or grandparents.)

To find out more about what you can do, click on the “Get Help” link on the menu bar above.

<table>
<thead>
<tr>
<th>Name</th>
<th>Relationship to me</th>
<th>Add History</th>
<th>Update History</th>
<th>Remove Relative</th>
</tr>
</thead>
<tbody>
<tr>
<td>My Family</td>
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<tr>
<td>undefined</td>
<td>Self</td>
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<td>Father</td>
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<td>Mother</td>
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<td>My Father's Side of the Family</td>
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<tr>
<td>Paternal Grandfather</td>
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<td>Paternal Grandmother</td>
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<td>My Mother's Side of the Family</td>
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<tr>
<td>Maternal Grandfather</td>
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<tr>
<td>Maternal Grandmother</td>
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<tr>
<td>Recently Added Family Members</td>
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</table>
Consumer Websites
MedlinePlus

- Health Topic pages:
  - Genetics
  - Genetic testing
  - Genetic counseling
  - Genetic disorders
  - Genes and therapy
- text word search
Results 1 - 10 of 945 for genomics

1. Frequently Asked Questions about Genetic and Genomic Science (National Human Genome Research Institute)
   ... this page Frequently Asked Questions About Genetic and Genomic Science What are genetics and genomics? Why are ... genomics? Genetics is a term that refers to the ...
   https://www.genome.gov/19016904 - External Health Links

2. Genomics and Health Impact Update (Centers for Disease Control and Prevention)
   ... Knowledge Base. What's New in the Public Health Genomics Knowledge Base New Implementation Tools Alzheimer's Disease Cardiomyopathy HIV/AIDS About the Genomics & Health Impact Update The Office of Public Health ...
   www.cdc.gov/genomics/update/current.htm - External Health Links

3. Brief Guide to Genomics: DNA, Genes and Genomes (National Human Genome Research Institute)
   ... Breve guía de genómica A Brief Guide to Genomics DNA, Genes and Genomes Deoxyribonucleic acid (DNA) is ... genetic basis for health and disease. Implications of Genomics for Medical Science Virtually every human ailment has ...
   https://www.genome.gov/18016863 - External Health Links

4. Genomic Testing (Centers for Disease Control and Prevention)
Genetics Home Reference

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

Health Conditions

Explore the signs and symptoms, frequency, genetic cause, and inheritance pattern of various conditions, diseases, and syndromes.

Browse by Category

All Health Categories

Browse by First Letter

A B C D E F G H I J K L M N O P
Q R S T U V W X Y Z

- anemia, hereditary
- anemia, sensorineural
- anemia, thalassemia
- anemia, vitamin B12 deficiency
- anemia, vitamin B6 deficiency
- anemia, vitamin C deficiency
- anemia, zinc deficiency

Learn More about Health Conditions

- What does it mean if a disorder seems to run in my family?
- What are the different ways in which a genetic condition can be inherited?
- What are complex or multifactorial disorders?
- What does it mean to have a genetic predisposition to a disease?
Genetics Home Reference

Resources

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
Genetics Home Reference

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
Genetics Home Reference

Search Results

Search for: genomic medicine

Viewing: All

What is precision medicine?
The Centers for Disease Control and Prevention covers precision medicine in its Genomics and Health

What is the difference between precision medicine and personalized medicine? What about
There is a lot of overlap between the terms "precision medicine" and "personalized medicine

What was the Human Genome Project and why has it been important?
GeneEd from the National Library of Medicine and the National Human Genome Research Institute

What were some of the ethical, legal, and social implications addressed by the Human Genome Project?
of the Human Genome Project. The mission of the ELSI program was to identify and address issues

What is a genome?
A genome is an organism's complete set of DNA, including all of its genes. Each genome contains all
NIH National Human Genome Research Institute

For Patients and the Public
- Community Engagement and Community Health
- Family History
- Genetics & Genomics Science & Research
- Genetic & Rare Diseases Information Center
- Genomic Medicine and Health Care
- Online Health and Support Resources
- Specific Genetic Disorders

For Health Professionals
- Competency & Curricular Resources
- Genetics 101
- Inter-Society Coordinating Committee (ISCC)
- New Horizons and Research
- Patient Management
- Policy and Ethics Issues
- What is Genomic Medicine?

Feature
Apply now for the "short course" health professionals track

The Genomic Healthcare Branch is offering the NHGRI Short Course in Genomics: Nurse, Physician Assistant and Faculty Track from August 1 - August 3, 2016. This year's course is for nurses, physician assistants, and the faculty who educate these health professionals in genomics. Read more

Webinars for Health Insurers and Payers: Understanding Genetic Testing

Highlights
Video: A G2C2 Website Overview

This introduction to the Genetics/Genomics Competency Center (G2C2) website at http://g2c2.org/ gives new users an opportunity to view key features of this centralized collection of genomics educational resources for healthcare educators and providers. Read more

Genomic knowledge is power in the fight against obesity

Although many doctors are

NHGRI https://www.genome.gov/
Direct to Consumer Testing

Genetic Testing
Direct to Consumer Testing
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
Example of a 23andMe test result
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.
# American College of Medical Genetics and Genomics

<table>
<thead>
<tr>
<th>Topic</th>
<th>Year</th>
<th>Journal</th>
<th>Volume</th>
<th>Pages</th>
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<tbody>
<tr>
<td>Folic Acid: Folic Acid and Neural Tube Defects</td>
<td>2010</td>
<td>ACMG Med</td>
<td>13:8</td>
<td>593-596</td>
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<td>Genetic Discrimination: Points to Consider in Preventing Unfair Discrimination Based on Genetic Disease</td>
<td>2001</td>
<td>ACMG Med</td>
<td>3:6</td>
<td>436-437</td>
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<td>Genetic Services: Clinical utility of genetic and genomic services: a position statement of the American College of Medical Genetics and Genomics</td>
<td>2015</td>
<td>ACMG Med</td>
<td>7:6</td>
<td>505-507</td>
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<tr>
<td>ACMG position statement on prenatal/ preconception expanded carrier screening</td>
<td>2013</td>
<td>ACMG Med</td>
<td>15:8</td>
<td>482-483</td>
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<td>Technical report: ethical and policy issues in genetic testing and screening of children</td>
<td>2013</td>
<td>ACMG Med</td>
<td>15:3</td>
<td>234-245</td>
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<tr>
<td>Risk categorization for oversight of laboratory-developed tests for inherited conditions</td>
<td>2013</td>
<td>ACMG Med</td>
<td>15:4</td>
<td>314-315</td>
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<tr>
<td>Genetic Testing in Adoption (ACMG/ASHG)</td>
<td>2000</td>
<td>ASHG Genet</td>
<td>66:761</td>
<td>767</td>
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<tr>
<td>ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and</td>
<td>2013</td>
<td>ACMG Med</td>
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</table>

**ACMG** [https://www.acmg.net/](https://www.acmg.net/)
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?
GINA Help  http://ginahelp.org/

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

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Choosing Wisely

Making Smart Decisions About Genetic Testing (ACMG)

Sometimes a genetic test is not the best way to find an inherited condition or disease risk. A routine blood test or procedure might be just as good, and it might be less costly and more easily available.

ACMG

https://www.acmg.net/docs/ACMG Choosing Wisely_Final.pdf
### Issues in Genetics

#### Policy, legal and ethical issues in genetic research

<table>
<thead>
<tr>
<th>Feature</th>
<th>Highlights</th>
</tr>
</thead>
<tbody>
<tr>
<td>Advisory committee addresses building a medical information commons</td>
<td>Investigational Device Exemptions (IDE) in Genomic Research Workshop</td>
</tr>
</tbody>
</table>

**While generating large amounts of medical and genomic data may improve health, questions about access and use remain. To address these challenges,**

**On June 10, 2016, the National Human Genome Research Institute (NHGRI) will host a day-long, public workshop - Investigational Device Exemptions (IDE) and Genomic Research.** The purpose of this workshop is educational. The Food and Drug Administration (FDA) may require

**See Also**

- Policy and Program Analysis Branch
- Policy Staff Contact Information
- Ethical, Legal and Social Implications Research Program
- NHGRI Extramural Research Program
- GenomeTV
- Issues in Genetics Archive
- Past Web content from the Issues in Genetics Section
Ethical and Policy Issues in Genetic Testing and Screening of Children

COMMITTEE ON BIOETHICS, COMMITTEE ON GENETICS, AND, THE AMERICAN COLLEGE OF MEDICAL GENETICS AND, GENOMICS SOCIAL, ETHICAL, AND LEGAL ISSUES COMMITTEE

Abstract

The genetic testing and genetic screening of children are commonplace. Decisions about whether to offer genetic testing and screening should be driven by the best interest of the child. The growing literature on the psychosocial and clinical effects of such testing and screening can help inform best practices. This policy statement represents recommendations developed collaboratively by the American Academy of Pediatrics and the American College of Medical Genetics and Genomics with respect to many of the scenarios in which genetic testing and screening can occur.

ACMG

Informing the Public
Precision Medicine

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

PMI announcement https://www.whitehouse.gov/precision-medicine
Precision Medicine Initiative

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.

PMI announcement https://www.whitehouse.gov/precision-medicine
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

[PMI Infographic](http://syndication.nih.gov/multimedia/pmi/infographics/PMI-infographic.pdf)
Two ways to participate

1. Through the cohort website
2. With participating health care provider organization
NIH and Precision Medicine Initiative

About the Precision Medicine Initiative Cohort Program

Far too many diseases do not have a proven means of prevention or effective treatments. We must gain better insights into the biological, environmental, and behavioral influences on these diseases to make a difference for the millions of Americans who suffer from them. 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. While some advances in precision medicine have been made, the practice is not currently in use for most diseases.
Informed Consent and the Precision Medicine Initiative
September 21, 2016 1PM

Presenter: Malia Fullerton, Associate Professor of Bioethics and Humanities at the University of Washington School of Medicine
“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.”
The 1000 Genomes Project was undertaken in order to increase the _________ of the genomes represented in public databases.

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

Clinicians are not concerned about all genetic variants – only those that are ____________.

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

True or False? A genetic variant may originally be classified as “likely pathogenic” and later classified as “likely benign.”

What resource would you recommend to consumers who wanted to learn more about a genetic condition?

What is a good starting place for finding genetic information for clinicians?
Questions?

Diana Louden, MLib
Biomedical & Translational Sciences Librarian
UW Health Sciences Library
DKNL@uw.edu

Carolyn Martin, MLS, AHIP
Consumer Health Coordinator
NN/LM PNR
martinc4@uw.edu

Presentation resources
https://nnlm.gov/pnr/training/presentations