

Colorado Association of Libraries



Genes: The Future of Personalized Medicine

Thursday - October 22, 2015

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Objectives

- Understand the basic concepts of and concerns surrounding precision medicine.
- Understand the role of pharmacogenomics in personalized medicine.
- Learn about genetic resources for consumers, patients, K-12+, healthcare providers, and researchers.

White House Initiative



Personalized Medicine

- Intervention based on unique genetic makeup
 - Waiting for disease to occur **vs.** predicting and preventing.
 - Recently discovered genetic variants affecting bone density and fracture risk – engrailed homeobox-1 gene (*EN1*).
 - Paying for costs and procedures **vs.** paying for outcomes and value.
 - Payers willing to reimburse for Hepatitis C treatment costing \$1,000 per pill because of its effectiveness.

Genetic vs. Genomic

- Often used interchangeably, but to a scientist they are different.
 - Genetic testing – refers to a specific sequence of DNA on a single chromosome.
 - Genomic testing – looks for variations within large segments across the entirety of genetic material.
 - Genomics-based healthcare will help us make more informed health decisions, based on our genetic makeup.

Diagnosing Disease

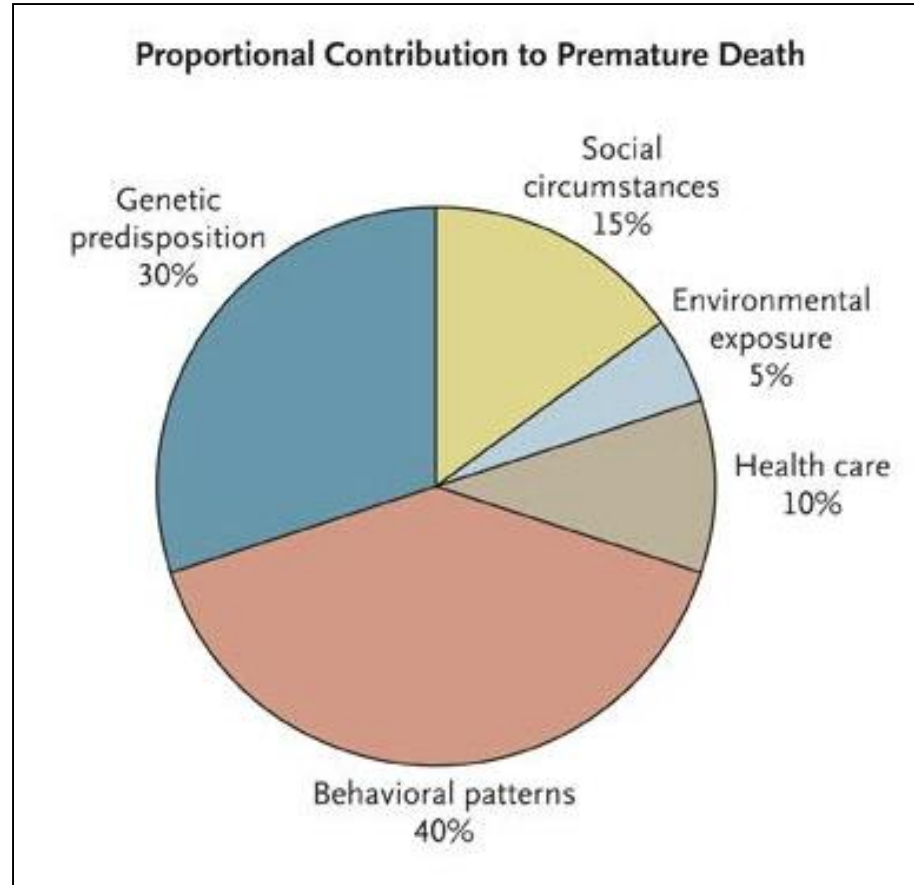


Precision Medicine

Why Now?

INNOVATION	2004	2014
Amount of time to sequence the human genome	2 years	2 days
Cost of human sequencing	\$22 million	\$1,000 to \$5,000
Number of smart phones	1 million	160 million
Computing power	n	n ¹⁶

Contributors to Premature Death



McGinnis JM, Williams-Russo P, Knickman JR. The case for more active policy attention to health promotion. *Health Aff (Millwood)* 2002;21:78-93

Genetic Testing

- Diagnostic – identifies current disease states (e.g., prenatal, newborn screenings).
- Carrier – determines if you carry a certain genetic trait (Cystic Fibrosis, Sickle Cell Anemia, Tay-Sachs Disease).
- Predictive – determines if you are at risk for developing a disorder based on family history (Huntington's disease, colorectal cancer).

Other Influencers

- Behavioral patterns
- Social determinants of health
- Health care-related mortality
- Environmental factors

Ethical, Legal, Regulatory Issues

- Who should have access to personal genetic information – Insurers? Employers? Courts? Military?
- Who owns and controls genetic information?
- How does genomic information affect members of minority communities?
- How reliable and useful is fetal genetic testing?
- How will genetic tests be evaluated and regulated for accuracy?
- Should testing be performed when no treatment is available?
- Who should regulate the safety of diagnostic devices, the risk to patients (or fetus), standards for testing?

Privacy

- Data about more than 120 million people has been compromised in more than 1,100 separate breaches at organizations handling protected health data since 2009.
- Discrimination by insurance companies, employers, society.

Cost

- Ability to pay.
 - Daraprim – from \$13.50 to \$750 (+5455%)
 - Cycloserine – from \$500 to \$10,800 (+2060%)
 - Doxycycline – from \$20 to \$1,849 (+9145%)
- Disparities in coverage and access.
- Does the treatment work?

Quality and Safety

- Quality assurance
- Validity of testing
 - Direct to consumer testing issues
- Regulation
- Society

Legal and Regulatory Developments

- 1996 – Health Insurance Portability and Accountability Act (HIPAA).
- 2009 – American Recovery and Reinvestment Act (ARRA).
- 2010 – Health Information Technology for Economic and Clinical Health Act (HITECH).
- 2005 – Genetic Information and Nondiscrimination Act (GINA).

Pharmacogenomics



Genetics Home Reference

- **Target Audience: consumers, students, teachers.**
- Starting point to learn about human genetics and inherited disorders.
- Conditions are chosen based on whether they have a known genetic basis – gene mutations or chromosomal changes.
- Entries come from MedlinePlus.
- Help Me Understand Genetics – basic genetics information in clear language.
- Links to relevant articles from the biomedical literature.
- <http://ghr.nlm.nih.gov/>

MedlinePlus

- **Target audience: consumers, caregivers, healthcare providers, students, teachers.**
- Patient handouts in English and Spanish.
- Interactive tutorials from the Dorlan DNA Learning Center (Cold Spring Harbor Laboratory)– the nation's first science center dedicated to DNA education
- Preformulated PubMed searches for patient education handouts.
- Newborn screening information.
- Clinical trials
- www.medlineplus.gov

PubMed Health

- **Target audience: consumers, researchers, healthcare providers.**
- Specializes in the review of clinical effectiveness research, running search simultaneously in PubMed.
- Over 500 genetic-related articles, many geared at consumers/patients.
- Resources to understand research results.
- Medical encyclopedia (A.D.A.M.) with links to MedlinePlus.
- <http://www.ncbi.nlm.nih.gov/pubmedhealth/>

PubMed

- **Target audience: consumers, researchers, healthcare providers.**
- Tool for locating medical literature from around the world.
- New MeSH terms: Genetic Therapy; Individualized Medicine; Genetic Privacy.
- Filter for Patient Education Handouts.
- <http://www.ncbi.nlm.nih.gov/pubmed>

National Human Genome Research Institute

- **Target audience: consumers, students, educators, health professionals.**
- Patient information on diseases, treatments, and research.
- English and Spanish talking glossary of genetic terms.
- Competency resources for health professionals.
- Policy and ethics issues.
- Genetic Testing Registry
 - Data from 770 labs, over 9,000 genetic tests
 - Indications for use, validity data, and **evidence of the test's usefulness**
 - Launch in early 2012
- <http://www.genome.gov/>

ClinicalTrials.gov

- **Target Audience: consumers, researchers, health care providers.**
- Currently contains over 200,000 trials, with 61,000 daily visitors from around the world.
- Locate studies by condition, drug intervention, topic, location.
- FDA drug and device information.
- Animated tutorials.
- <http://clinicaltrials.gov/>

researchMatch

- **Target Audience : consumers, researchers.**
- Non-profit match site for people trying to find research studies and researchers looking for participants.
- Matches created based on non-identifying data provided by volunteer.
- Links to MedlinePlus and ClinicalTrials.gov.
- Register to create profile.
- <https://www.researchmatch.org/>

NCBI Bookshelf: Genes and Diseases

Target Audience: consumers, students

- 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.
- <http://www.ncbi.nlm.nih.gov/books/NBK22183/>

NIH: Office of Rare Diseases Research

- **Target Audience: consumers, researchers, health care providers, educators, students.**
- 10% of the U.S. have a rare disease (disease or condition affecting fewer than 200,000).
- 7,000 known rare diseases.
- Site serves as a portal to information in the rare diseases community, including genetic diseases.
- Resources available in Spanish.
- <http://rarediseases.info.nih.gov/>

Genetic Alliance

- **Target audience: consumers, advocates, health professionals, government agencies, think tanks.**
- Network of over 1,000 disease-specific advocacy organizations.
- Integrates individual, family and community perspectives.
- InfoSearch locates organizations, research and links to NLM resources.
- Resource repository locates documents, links, audio and video files.
- <http://www.geneticalliance.org/>

OMIM – Online Mendelian Inheritance in Man

- **Target audience: physicians, genetic disorder professionals, genetic researchers, advanced students in science/medicine.**
- Clinical features, diagnosis, mapping, molecular genetics, history.
- All known hereditary disorders and over 20,000 genes.
- Updated daily – copious links to genetic resources.
- <http://www.ncbi.nlm.nih.gov/omim>

GenETHX

- **Target audience: consumers, students, educators, anyone involved in genetics research/counseling.**
- Bioethics Research Library – Georgetown University.
- Ethic issues on counseling, testing, gene therapy, behavioral genetics, sociobiology, genome mapping, eugenics, human ancestry.
- Scans bibliographic resources, portals and databases.
- Scholarly and popular resources.
- <http://bioethics.georgetown.edu/index.html>

Lab Tests Online

- **Target audience: consumers, patients, caregivers, health professionals.**
- In 14 languages.
- Searchable by test, condition/disease, and screening.
- Helps patients understand how to decipher a lab report, make informed decisions, and formulate questions for their care provider.
- <https://labtestsonline.org/>

CDC Public Health Genomics

- **Target audience: consumers, public health workforce, policy makers.**
- Genomic news and publications relevant to public health.
- Information in English and Spanish.
- Genomics translation – bench to bedside.
- Autism, breast/ovarian cancer, heart disease, hemochromatosis/iron overload, mental health, obesity, sickle cell disease, stroke.
- <http://www.cdc.gov/genomics/>

Genetic Testing

- Clinical testing
 - Follow-up with healthcare provider and/or genetic counselor
 - Cost ranges from under a \$100 to more than \$2,000 (pre-natal \$15-60)
- Direct-to-consumer testing
 - Health related, Nutrigenetic, Non-Medical
 - Cost ranges from several \$100 to over a \$1,000
 - Often no follow-up guidance for treatment/prevention
- Resources
 - http://rarediseases.info.nih.gov/Resources/Genetics_Information_Services.aspx
 - <http://bioethics.od.nih.gov/geneticstesting.html>
 - <http://ghr.nlm.nih.gov/handbook/testing>
 - <http://vsearch.nlm.nih.gov/vivisimo/cgi-bin/query-meta?v%3Aproject=medlineplus&query=genetic+testing&x=0&y=0>
 - <http://www.cdc.gov/genomics/gtesting/index.htm>

Genetic Counseling

- There are 3,500 trained genetic counselors in North America.
 - National Society of Genetic Counselors @ <http://nsgc.org/p/cm/ld/fid=164>
 - National Cancer Institute @ <http://www.cancer.gov/cancertopics/genetics/directory>
 - NCBI Genetic Testing Clinic Directory @ <http://www.ncbi.nlm.nih.gov/sites/GeneTests/clinic?db=GeneTests>

Family History

- A comprehensive family health history is the best “genetic test.”
 - Learn if you are at risk for future health problems.
 - Learn if there are ways to reduce risk.
- Tools
 - My Family Health Portrait in English/Spanish
<https://familyhistory.hhs.gov/fhh-web/home.action>
 - Family Health History guides in English/Spanish -
<http://www.geneticalliance.org/fhh>

Resources for New Parents

- Prenatal diagnostic testing and newborn screening
 - State Newborn Screening Programs - <http://www.cdc.gov/genomics/resources/a.htm#newborn>
 - National Newborn Screening and Global Resource Center - <http://genes-r-us.uthscsa.edu/>
- Expecting Health - <http://www.geneticalliance.org/programs/expecting-health>
- Your Genes, Your Health - <http://www.ygyh.org/>

Resources for the Classroom

- GeneEd (Grades 9-12)-
<http://geneed.nlm.nih.gov/>
- Genetics Home Reference (Grades 6-12+) -
<http://ghr.nlm.nih.gov/>
 - Help Me Understand Genetics Handbook (Grades 6-12+), <http://ghr.nlm.nih.gov/handbook>
- Harry Potter's World (Grades 9-12) -
<https://www.nlm.nih.gov/exhibition/harrypottersworld/>

Questions/Comments

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