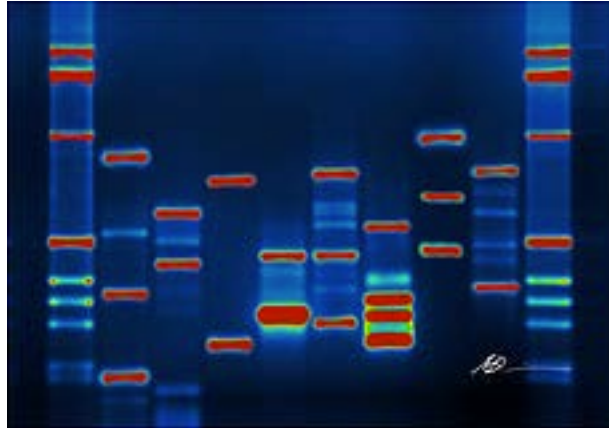


Spotlight! On Genetics



Genetics Resources

Presented October 26, 2011

Dana Abbey, MLS

Health Information Literacy Coordinator

National Network of Libraries of Medicine, MidContinental Region

University of Colorado – Anschutz Medical Campus

Dana.abbey@ucdenver.edu



National Network of Libraries of Medicine
MidContinental Region

Colorado • Kansas • Missouri • Nebraska • Utah • Wyoming

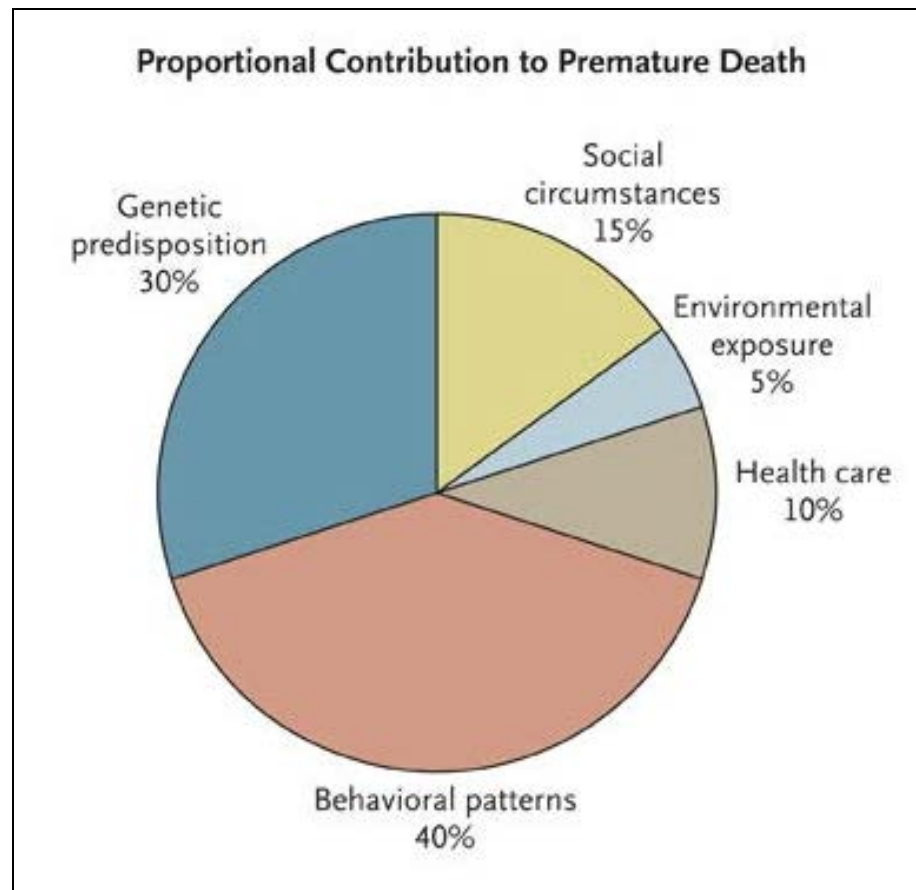
Image courtesy of Micah Baldwin



Objectives

- Learn about genetic resources for consumers, students, healthcare providers, and researchers
- Be familiar with resources for understanding genetic testing and counseling
- Know resources to explore the ethical, legal and social issues of human genetics
- Learn about resources to document family health history
 - All links @ <http://delicious.com/dabbey1/genetics>

Good News



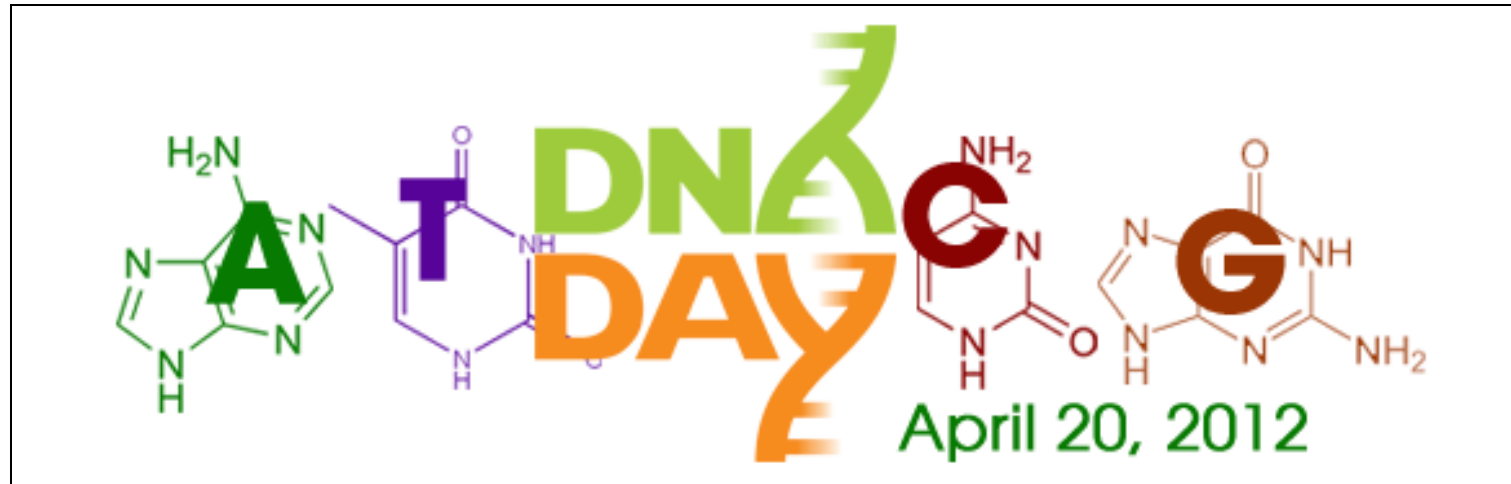
McGinnis JM, Williams-Russo P, Knickman JR. The case for more active policy attention to health promotion. *Health Aff (Millwood)* 2002;21:78-93



Genetics in the News

- DNA sequenced of a woman who lived to 115 – revealed rare genetic changes
- Prenatal blood test that can detect Down syndrome in a fetus in early pregnancy is now available to doctors in 20 U.S. cities
- OXTR gene variants (2 copies of the guanine allele) might contribute to increased optimism, mastery and self esteem
- Study finds pregnant women with a particular gene variation at higher risk of having child with autism – offers possibility of genetic test
- Breast cancer study of Bahamian women finds genetic mutation in 25% of relatives – raising lifetime risk 70 to 80% of developing breast cancer
- Prisoner exonerated after serving 25 years when DNA links killing to another man

National DNA Day





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
- Easy-to-read summaries (529) and full technical reports
- Resources to understand research results
- Medical encyclopedia (A.D.A.M.) with links to MedlinePlus
- <http://www.ncbi.nlm.nih.gov/pubmedhealth/>



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 115,026 trials, with 65,000 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**
- 25-30 million people in the U.S. have a rare disease (disease or condition affecting fewer than 200,000)
- 6,800 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 12,000 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>



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 only about 2,500 trained genetic counselors and 1,100 clinical geneticists in North America
 - National Society of Genetic Counselors @ <http://www.nsgc.org/FindaGeneticCounselor/tabid/64/Default.aspx>
 - 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
- Example: Heart disease and stroke
- Tools
 - My Family Health Portrait - <https://familyhistory.hhs.gov/fhh-web/home.action>
 - Family Health History guides in English/Spanish - <http://www.geneticalliance.org/fhh>



Resources for Parents

- Prenatal diagnostic testing and newborn screening
- State Newborn Screening Programs - <http://www.cdc.gov/genomics/resources/a.htm#newborn>
- National Newborn Screening Status Report - <http://genes-r-us.uthscsa.edu/nbsdisorders.pdf>

Questions/Comments

Dana Abbey, MLS

Dana.abbey@ucdenver.edu

303-724-2110