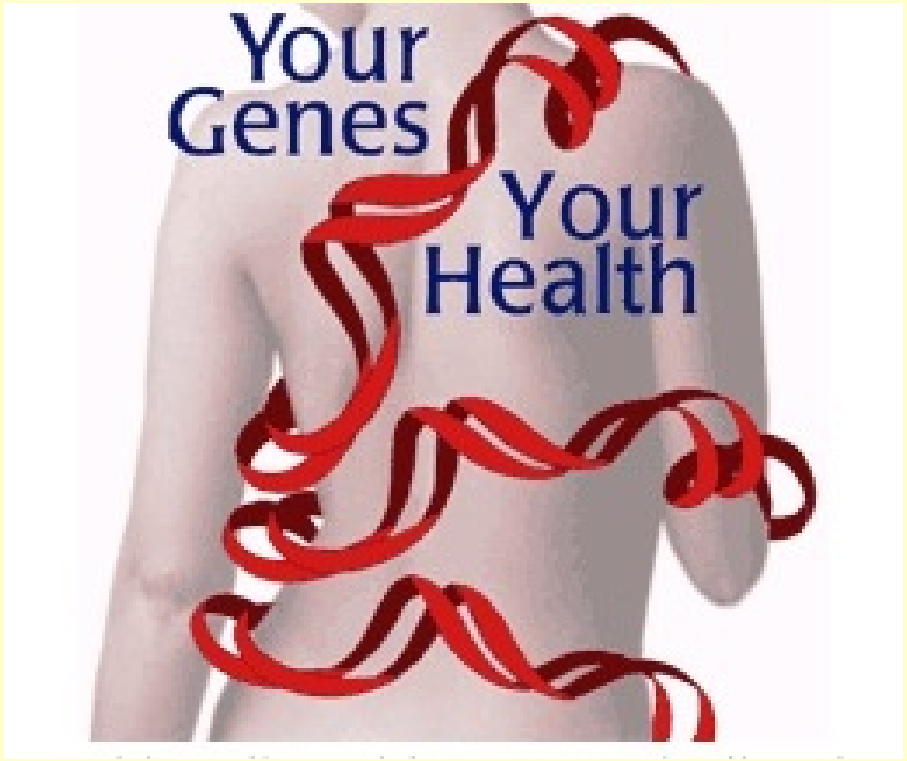
Your Genes and Your Health

http://bio84.stanford.edu/

Stanford Continuing Studies

http://continuingstudies.stanford.edu/

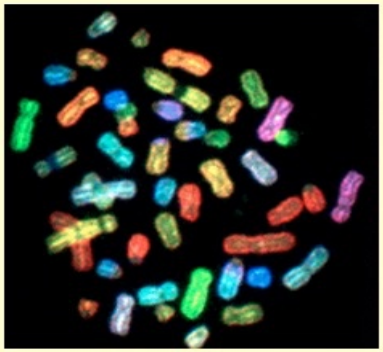


Doug Brutlag, Professor Emeritus
Biochemistry and Medicine (by courtesy)
brutlag@stanford.edu

Your Genes and Your Health

http://bio84.stanford.edu/

Your Genes and Your Health



Stanford Continuing Studies

Welcome to Bio 84: Your Genes and Your Health

Doug Brutlag

2016

Course Summary

Analyzing your DNA can reveal a lot about your potential health and future well being. Although knowledge of your genetic frailties may suggest a predisposition to a disease, genetics alone does not seal your fate. Most common diseases are affected by your behavior and your environment. Changing one's lifestyle can alleviate or even prevent disease. Hence, understanding genetic indicators can alert one to the importance of being vigilant about lifestyle and medical follow-ups.

In this course, we will empower you with tools to learn more about diseases, treatments and genetic tests that will help you to understand any genetic disease. The instructor will use data from his own genome to show you how you can look into your ancestry, family relationships, inherited diseases and response to drugs. You will see how knowing one's genetic profile can lead to reduced health care costs and a new approach to a confident, healthy lifestyle.

Course Syllabus

http://bio84.stanford.edu/

Date	Topic	Videos 2015	Videos 2016	Slide Link 1	Slide Link 2
Jan 13	Introduction: Your Genes and Your Health	Video		Slides	
Jan 20	Disease, Genetics and Clinical Trial Resources	Video		Slides	
Jan 27	The Human Genome Project	Video		Slides	Slides
Feb 3	Genome Variations: SNPs and CNVs	Video		Slides	Slides
Feb 10	Gene Variations Associated with Disease	Video		Slides	
Feb 17	What You Can Learn from Personal Genomics	Video		Slides	
Feb 24	The Potential of Genetic and Stem Cell Therapies to Treat Disease	Video		Slides	Slides
Mar 2	Gene Expression and Cancer Diagnostics	Video		Slides	
Mar 9	MicroRNA and Epigenetic Regulation of Gene Expression	Video		Slides	Slides
Mar 16	The Impact of Genomics on Drug Discovery, Development and Delivery	Video		Slides	Slides

Course Requirements

- No scientific or medical background is necessary for this course.

- For those enrolled for a grade, the grade will be determined based on attendance (9 out of 10 lectures) and a short (4-5 page paper) paper due at the end of the course on a topic of interest to you and relevant to genomics and medicine. See example papers on the course web site:

  http://bio84.stanford.edu

- For those taking the course for Credit/No Credit, credit will depend on attendance (9 out of 10 lectures) and a short, 1-2 page paper due at the end of the course. You may choose one of the following paper topics:

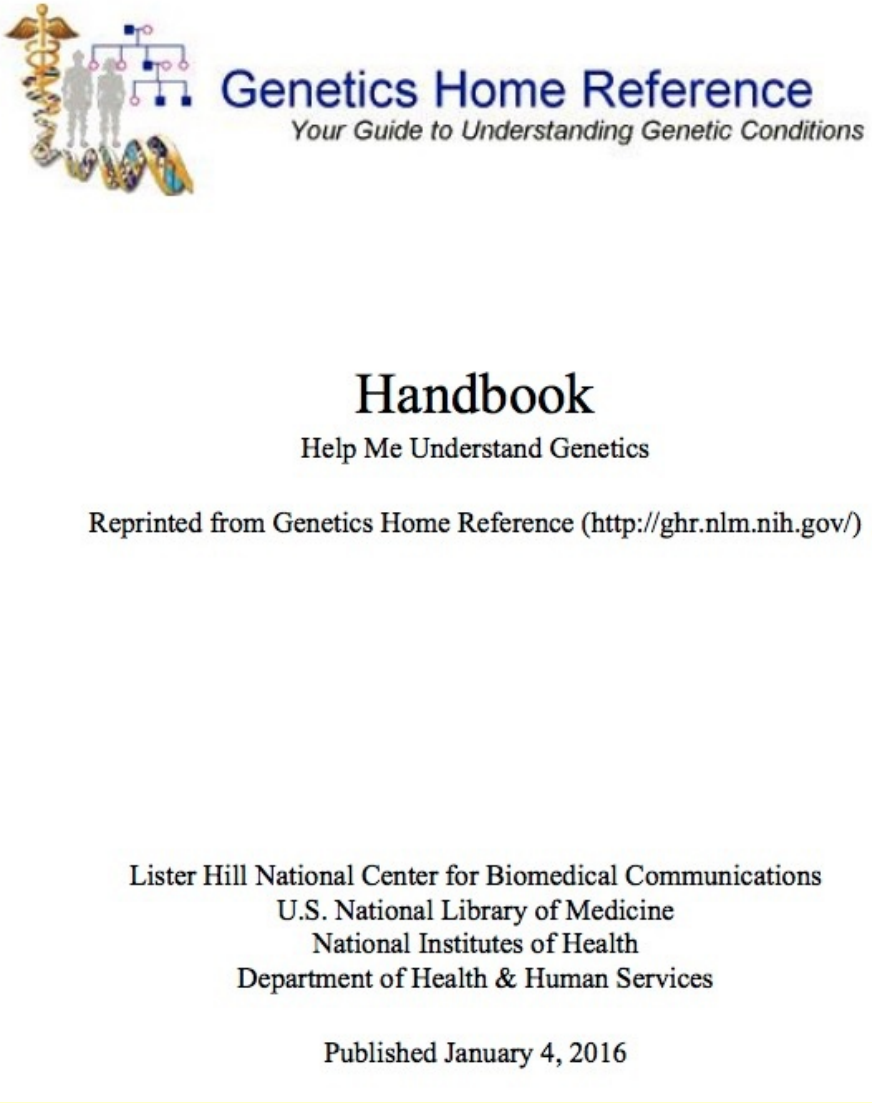
Suggested Final Paper Topics

Suggestion 1: Describe an inherited disease
Suggestion 2: Describe the role of a gene in an inherited disease
Suggestion 3: Describe a genome wide association study
Suggestion 4: Describe a novel stem cell or gene therapy for a disease
Final project ideas for students wishing a letter grade

- If you miss a lecture, you may get credit for attending that lecture by watching it on the video link in the course web page and sending me an email with a question about the lecture.

Genetics Home Reference Handbook

http://ghr.nlm.nih.gov/handbook.pdf



Genetics Home Reference

Your Guide to Understanding Genetic Conditions

Handbook

Help Me Understand Genetics

Reprinted from Genetics Home Reference (http://ghr.nlm.nih.gov/)

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services

Published January 4, 2016

Free to download

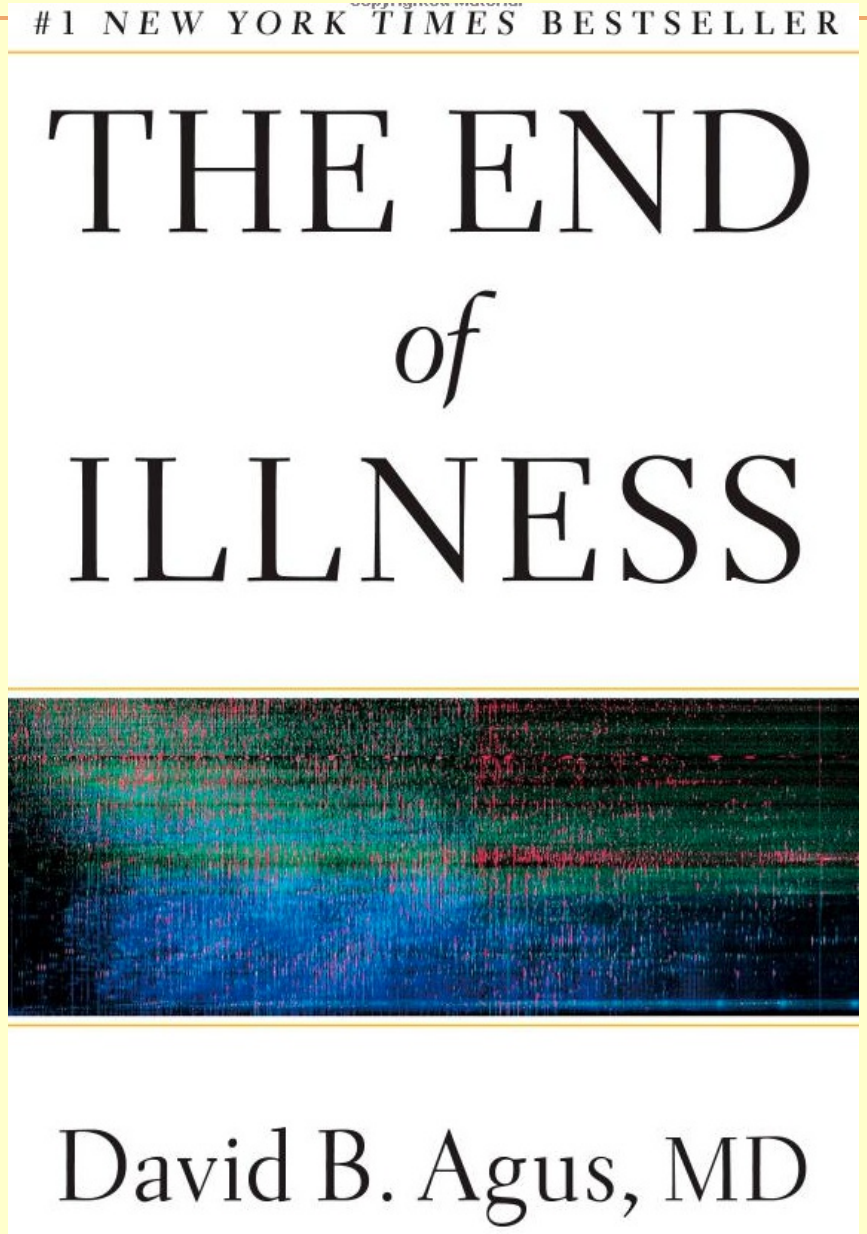
Genetics Home Reference Handbook

http://ghr.nlm.nih.gov/handbook.pdf

Table of Contents

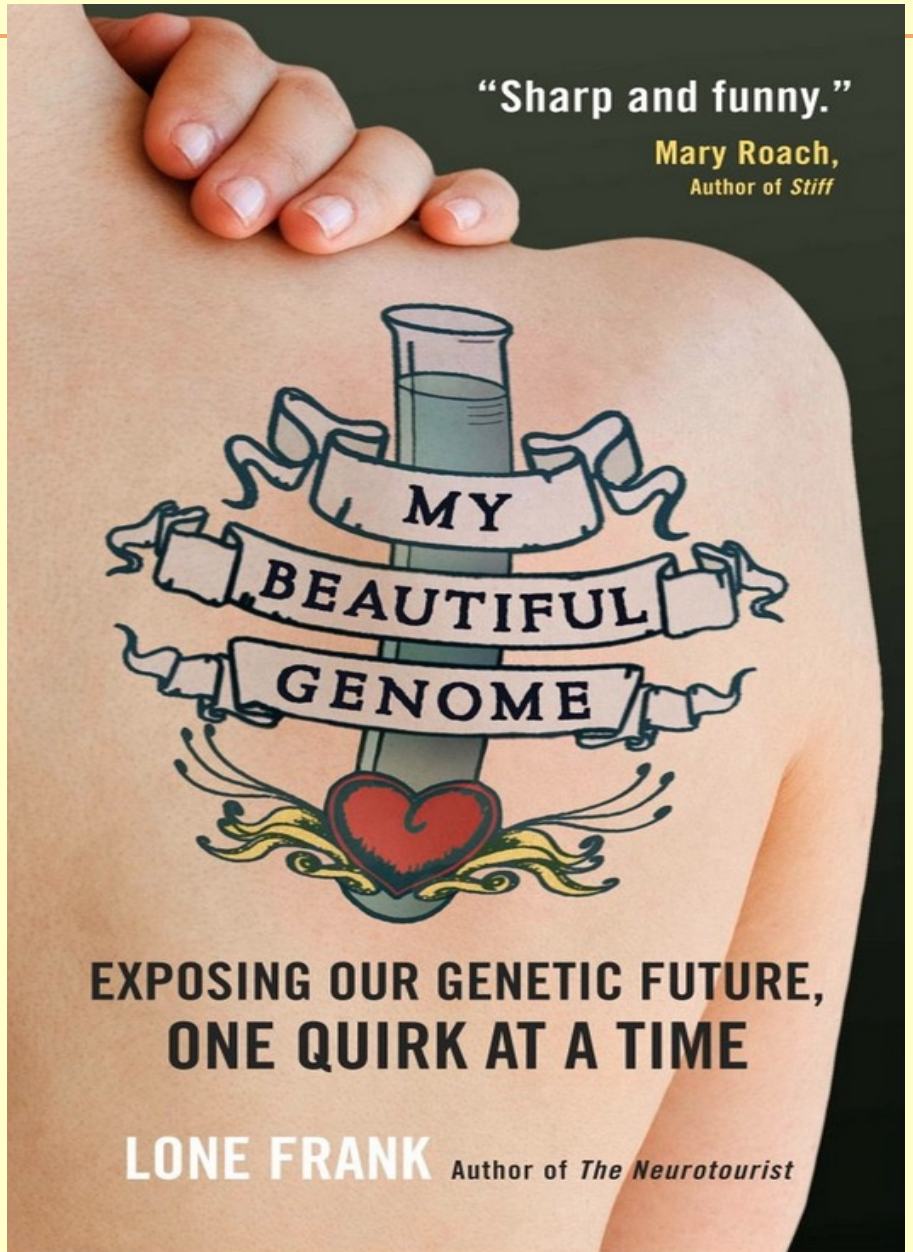
Free to download

The End of Illness David B. Agus



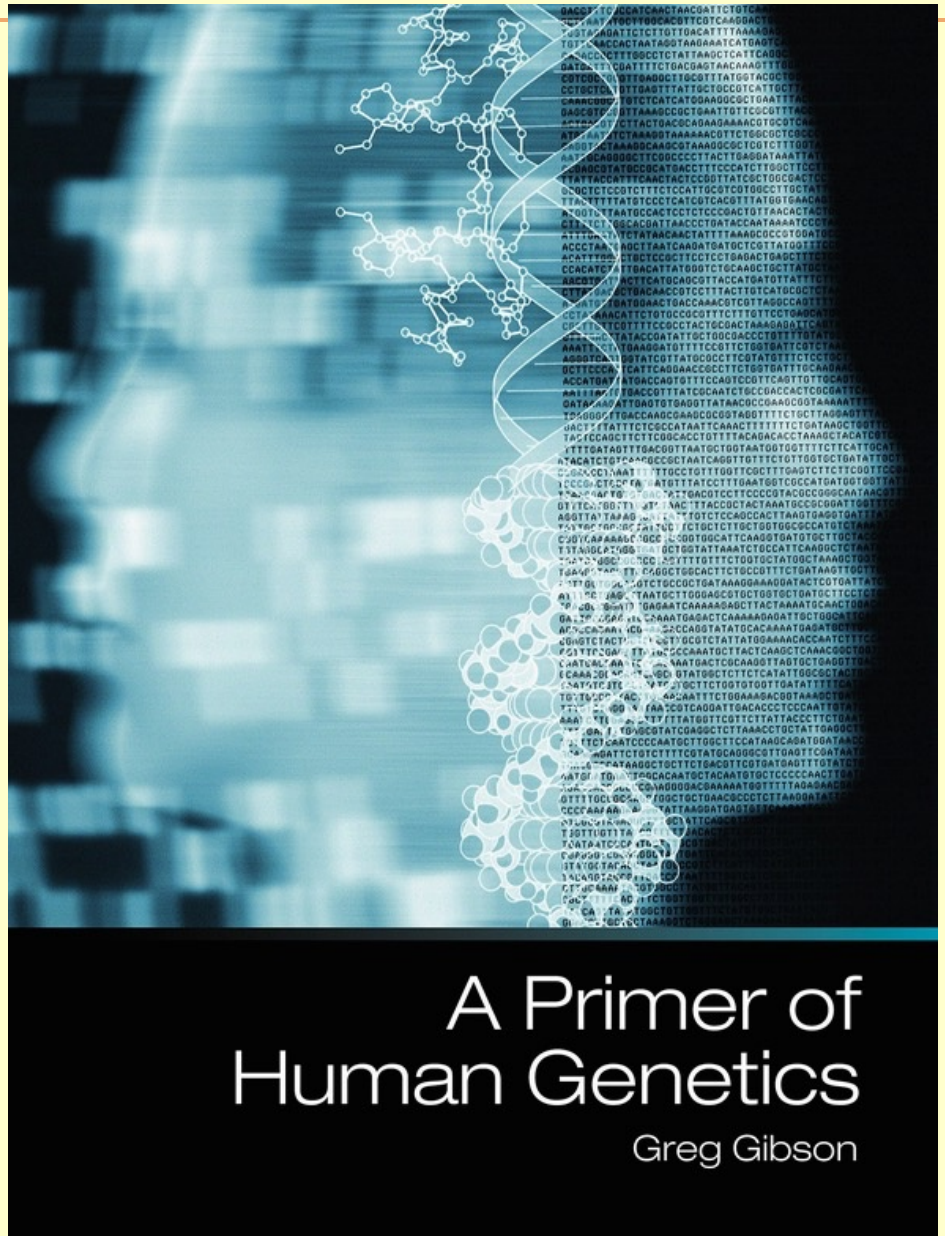
$12 download on Amazon

My Beautiful Genome by Lone Frank

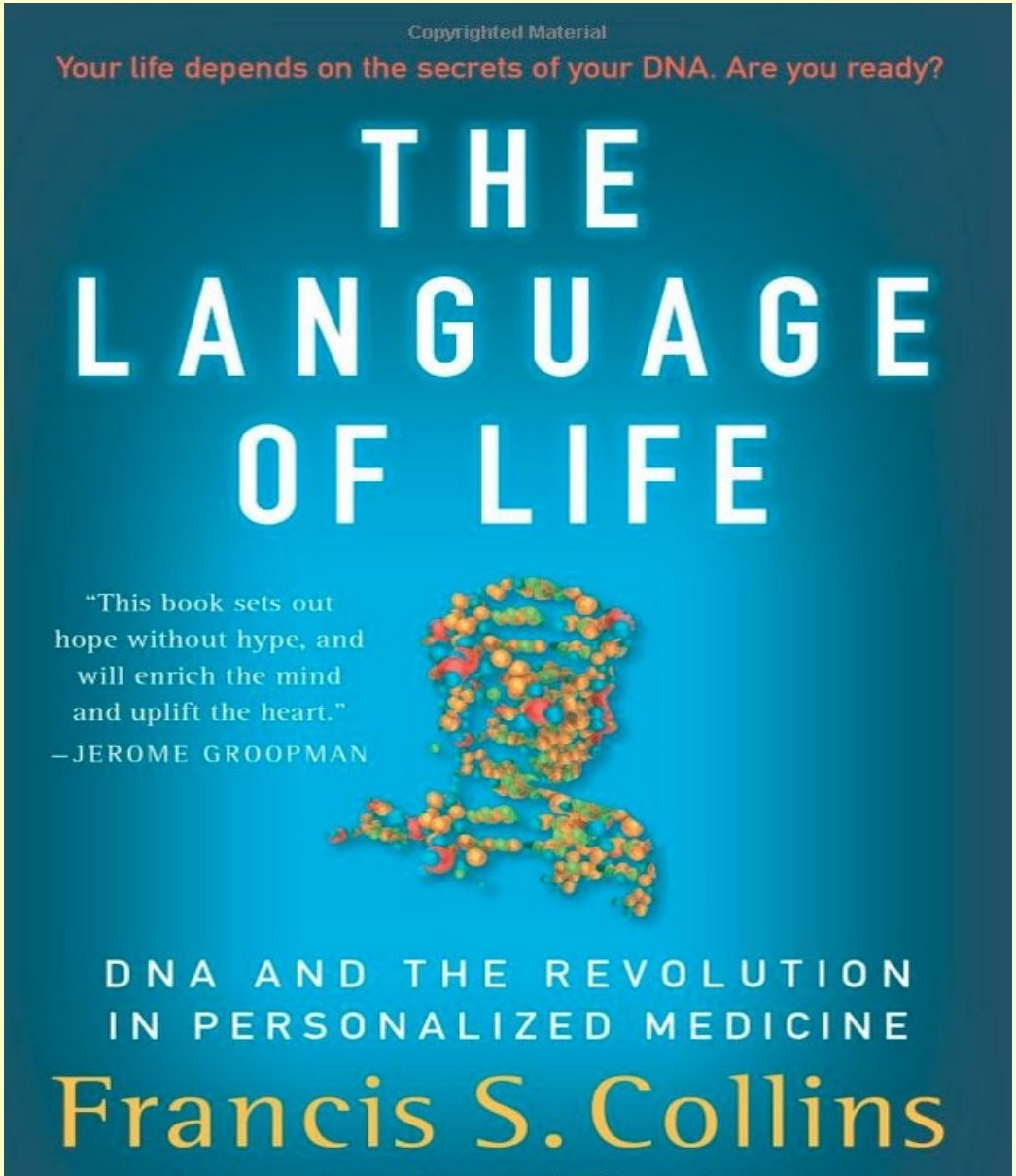


Sinauer Associates

Gibson: A Primer of Human Genetics



The Language of Life: DNA and the Revol



Genome TV – YouTube Videos

http://www.genome.gov/GenomeTV/

GenomeTV

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Welcome

GenomeTV is the National Human Genome Research Institute (NHGRI) collection of video resources. A wide variety of videos is available, from lectures, to news documentaries, to full video collections of meetings that tackle the research, issues and clinical applications of genomic research.

View All Videos

National Advisory Council for Human Genome Research

Event starts Mon, Feb 8 2016 7:00 AM PST

GET NOTIFIED

livestream

Go to: GenomeTV Live page

Note: Click on the title for the video, on the icon for more information.

NHGRI Advisory Council

- 09/2015 Council Open Session
- 05/2015 Council Open Session
- 02/2015 Council Open Session
- 09/2014 Council Open Session
- 05/2014 Council Open Session

View All

Symposia

- 05/2015 TCGA Fourth Annual Symposium
- 08/2014 GCTP Alumni Research Symposium
- 05/2014 TCGA Third Annual Symposium
- 09/2013 The African Diaspora

News and Documentary

- 11/2015 Family Health History Day
- 09/2015 1000 Genomes Project
- 09/2015 Impact of HGP, 25 Years From Launch
- 09/2015 G2C2 Overview
- 09/2015 UDN Accepts Patient

Lectures

- 05/2015 Genomics and Health Disparities
- 02/2015 Jeffrey M. Trent Lecture
- 07/2014 Current Topics in Genome Analysis
- 06/2014 Genomics in Medicine (All Videos)

Education and Training

- 08/2015 NHGRI Three-minute Talks (TmT)
- 08/2015 Genetics and Genomics Primer
- 07/2015 (Factor) Analyze This: PCA or EFA
- 03/2015 Pre-Application Webinar: PA-14-015

Workshops and Meetings

- 12/2015 A Quarter Century After the HGP ...
- 11/2015 NSIGHT Public Webinar
- 10/2015 ENCODE/Roadmap ASHG Tutorial
- 09/2015 CSER and Beyond
- 07/2015 ENCODE 2015 Users Meeting

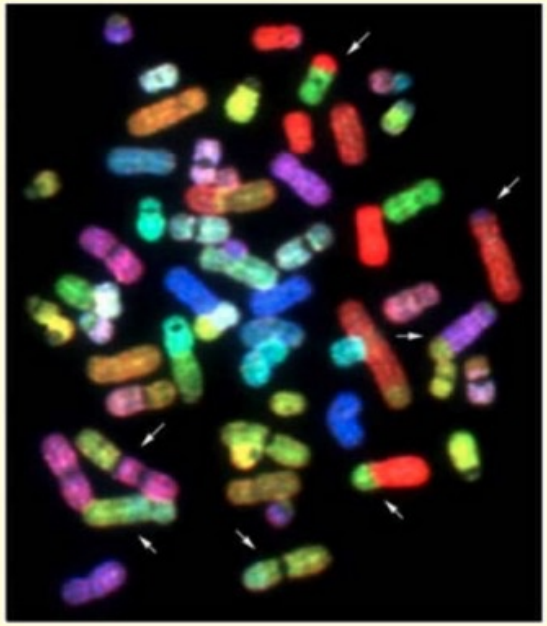
Genomics, Bioinformatics & Medicine

http://biochem158.stanford.edu/

Biochemistry 158/258 - HumBio 158G

BioMedical Informatics 258

Genomics, Bioinformatics & Medicine



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Course Topics

http://bio84.stanford.edu/

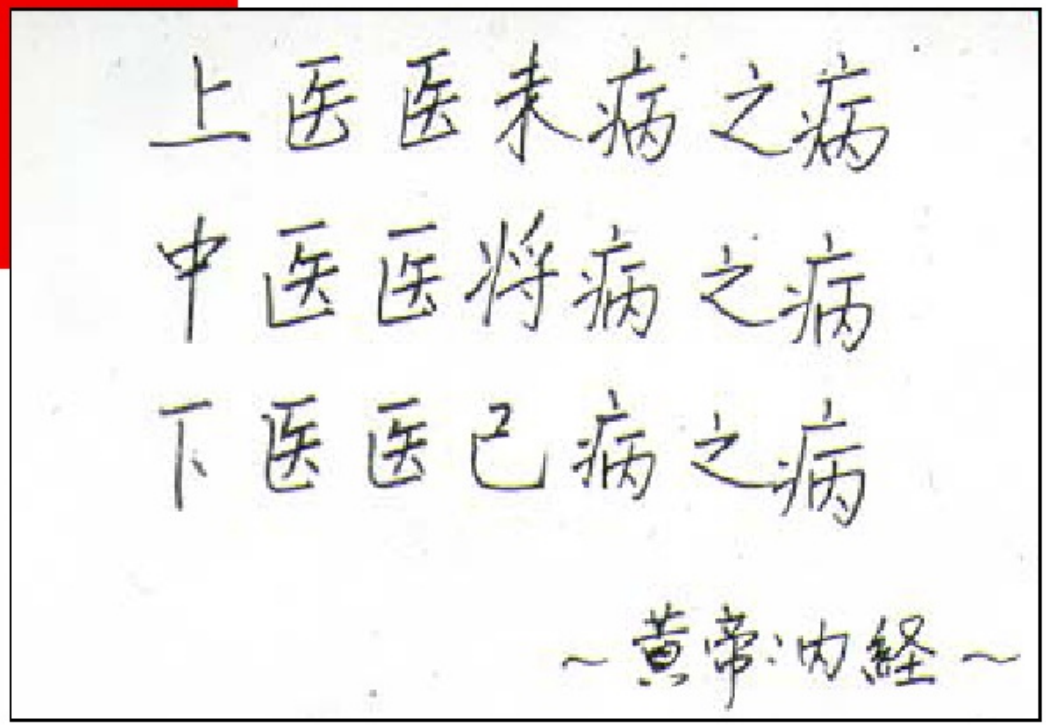
- Inherited diseases and traits
  - Disease databases and websites
  - Case presentations
- Genomics and novel diagnostics
  - Sequencing human genome
  - Genomic databases and websites
  - Genome variations leading to diseases and traits
  - Genome wide association studies
- Personal genomics
  - Carrier Status
  - Predisposition to disease
  - Pharmacogenomics
  - Family, genealogies and ancestry
- Gene expression and disease
  - Cell signaling and transcriptional regulation
  - MicroRNAs and translational regulation
  - Protein modification and regulation

Course Topics (continued)

- Drug development and pharmacogenomics
  - Analyzing the function of genes and regulatory regions
  - Drug target identification and validation
  - Identifying new potential drug targets via bioinformatics methods
  - Drug development paradigm and effect of genomics
  - Personalized medicine and medications
- Novel Therapies
  - Gene therapy approaches
  - Stem cell therapies
    - Pluripotent human embryonic stem cells
    - Adult stem cell approaches
    - Induced pluripotent stem cells
- Ethical issues
  - Discrimination for employment and insurability
  - Privacy of genetic information
  - Genetic selection and eugenics
  - Interventions in procreation

Introductions

Preventive Medicine



"Superior Doctors Prevent the Disease.
Mediocre Doctors Treat the Disease Before Evident.
Inferior Doctors Treat the Full Blown Disease."

-Huang Dee: Nai - Ching (2600 B.C. 1st Chinese Medical Text

Founder of Preventive Medicine: Louis Pasteur

When thinking about diseases, I never think about how to cure them, but instead I think about how to prevent them.

Immunization: A Fragile Fortress

Preventive Medicine

- The goal is to prevent disease from occurring.
- First one must identify the cause of the disease.
- Treat the cause of the disease rather than the symptoms
  - Example 1: Peptic Ulcers
  - Example 2: Pyrogens
- Genomics identifies genetic causes of inherited disease.
- When Paul Wise (a Stanford pediatrician) heard that we may soon sequence every child's genome at birth, he stated:
  - **"… all medicine may soon become pediatrics!"**
- Overlooked acquired diseases such as infections, accidents, environmental diseases and aging.
- Health care costs can be greatly reduced if we:
  - Invest in preventive medicine
  - Target the cause of disease rather than symptoms
  - Eliminate the need for expensive therapies.

Health Care Policy

- Current health care treats symptoms and full blown disease (illness care?)
- Future health care should be focused on preventive medicine
- Reduce need for expensive treatments (interventions).
- Need policies that incentivize patients and doctors to prevent disease.

Is Medicine a Science or an Art?

If it were not for the great variability among individuals, medicine might well be a science, not an art.

- Sir William Osler, Physician 1892
- Johns Hopkins School of Medicine
- Johns Hopkins Hospital
- Father of modern medicine

Personalized Medicine

Courtesy of Felix W. Frueh US FDA

Personalized Medicine

- Medicine is personal:
  - We are all different.
  - Some of our genetic differences translate into how we react to drugs as individuals.
  - This is why personalized medicine is important to everyone.
- Why does someone need twice the standard dose to be effective?
- Why does this drug work for you but not me?
- Why do I have side-effects and you don't?
- Why do some people get cancer and others don't?
- Why is anecdotal information irrelevant to your own health and treatment?

NIH Precision Medicine Initiative

http://www.nih.gov/precisionmedicine/

National Institutes of Health
Turning Discovery Into Health

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NIH Home > Research & Training

PRECISION MEDICINE INITIATIVE

Precision Medicine Initiative

- What are the near-term goals?
- What are the longer-term goals?
- How is it different?
- Who will participate?
- NIH Workshop

Precision Medicine Initiative

Far too many diseases do not have a proven means of prevention or effective treatments. We must gain better insights into the biology of 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 significant advances in precision medicine have been made for select cancers, the practice is not currently in use for most diseases. Many efforts are underway to help make precision medicine the norm rather than the exception. To accelerate the pace, President Obama has now unveiled the Precision Medicine Initiative – a bold new enterprise to revolutionize medicine and generate the scientific evidence needed to move the concept of precision medicine into every day clinical practice.

Email Updates

To sign up for updates please enter your e-mail address.

Submit

Related Links

- NEJM Perspective: A New Initiative on Precision Medicine
- White House Precision Medicine Web Page
- White House Fact Sheet: President Obama's Precision Medicine Initiative
- Precision Medicine Initiative and Cancer Research

NHS 100,000 Genomics England Project

http://www.genomicsengland.co.uk/

Genomics england

Home | About Genomics England | The 100,000 Genomes Project | GeCIP | Library and resources | News | Contact us

Genomics England, with the consent of participants and the support of the public, is creating a lasting legacy for patients, the NHS and the UK economy through the sequencing of 100,000 genomes: the 100,000 Genomes Project.

Genomics England was set up by the Department of Health to deliver the 100,000 Genomes Project. Initially the focus will be on rare disease, cancer and infectious disease. The project is currently in its pilot phase and will be completed by the end of 2017.

Read more...

Huntington Disease

- Autosomal Dominant
  - On the tip of the short arm of chromosome 4
  - One bad gene causes disease (dominant)
  - Brain degeneration over 10-15 years until death
- Neurodegenerative disease
  - Loss of movement control
  - Loss of cognitive skills (dementia) and hallucinations
  - Depression, hostility, aggression and loss of inhibitions
- Dyskinesias
  - Chorea: uncontrollable tics and involuntary movements of extremities, hyperkinesias
  - Dystonia uncontrollable muscle contractions
  - Dysphagia (difficulty in swallowing) and uncontrollable oral buccal dyskinesia
  - Bradykinesia, slow uncertain movements

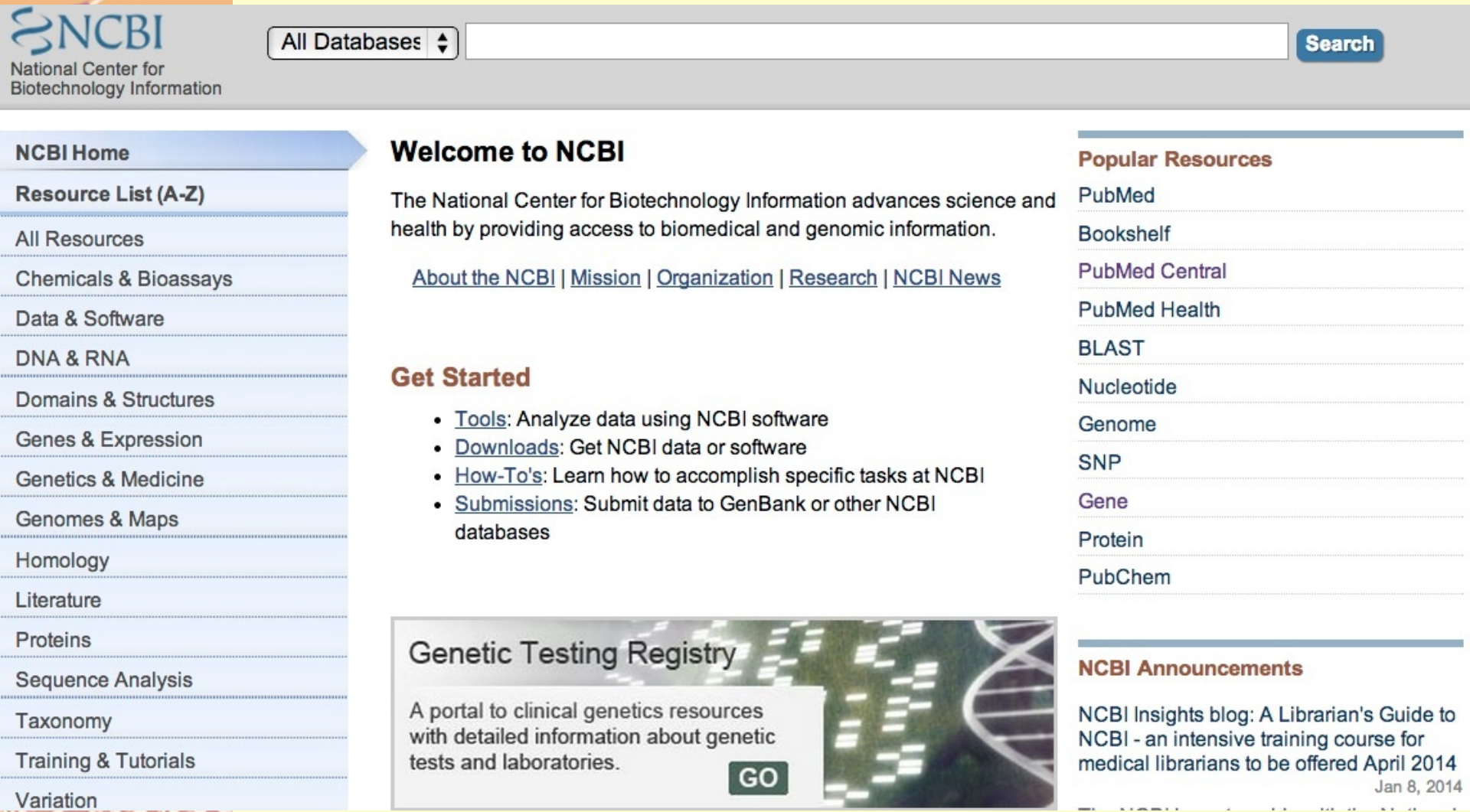
Huntington Disease

The Inheritance

- You are 20 years old.
- Your father abandoned you and your mother when you only 3 years old.
- Your father died this year and left you an inheritance.
- He died from an autosomal dominant disease known as Huntington's Chorea or Huntington's Disease.
- You have a 50% chance of inheriting this invariably fatal neurodegenerative disease.
- But there is a genetic test for this disease that can tell you not only if you have the disease, and if you do, when you will die from it.
- Would you take the genetic test or not?
- Why?

NCBI: National Center for Biotechnology Information

http://www.ncbi.nlm.nih.gov/



NCBI: Genetics and Medicine

http://www.ncbi.nlm.nih.gov/guide/genetics-medicine/

Gene

A searchable database of genes, focusing on genomes that have been completely sequenced and that have an active research community to contribute gene-specific data. Information includes nomenclature, chromosomal localization, gene products and their attributes (e.g., protein interactions), associated markers, phenotypes, interactions, and links to citations, sequences, variation details, maps, expression reports, homologs, protein domain content, and external databases.

GeneReviews

A collection of expert-authored, peer-reviewed disease descriptions on the NCBI Bookshelf that apply genetic testing to the diagnosis, management, and genetic counseling of patients and families with specific inherited conditions.

Genes and Disease

Summaries of information for selected genetic disorders with discussions of the underlying mutation(s) and clinical features, as well as links to related databases and organizations.

Genetic Testing Registry (GTR)

A voluntary registry of genetic tests and laboratories, with detailed information about the tests such as what is measured and analytic and clinical validity. GTR also is a nexus for information about genetic conditions and provides context-specific links to a variety of resources, including practice guidelines, published literature, and genetic data/information. The initial scope of GTR includes single gene tests for Mendelian disorders, as well as arrays, panels and pharmacogenetic tests.

NCBI: Genetics and Medicine

http://www.ncbi.nlm.nih.gov/guide/genetics-medicine/

MedGen
A portal to information about medical genetics. MedGen includes term lists from multiple sources and organizes them into concept groupings and hierarchies. Links are also provided to information related to those concepts in the NIH Genetic Testing Registry (GTR), ClinVar, Gene, OMIM, PubMed, and other sources.

Online Mendelian Inheritance in Animals (OMIA)
A database of genes, inherited disorders and traits in animal species (other than human and mouse), with textual information and references, as well as links to relevant records from other NCBI databases, such as PubMed and Gene.

Online Mendelian Inheritance in Man (OMIM)
A database of human genes and genetic disorders. NCBI maintains current content and continues to support its searching and integration with other NCBI databases. However, OMIM now has a new home at omim.org, and users are directed to this site for full record displays.

PubMed
A database of citations and abstracts for biomedical literature from MEDLINE and additional life science journals. Links are provided when full text versions of the articles are available via PubMed Central (described below) or other websites.

PubMed Central (PMC)
A digital archive of full-text biomedical and life sciences journal literature, including clinical medicine and public health.

PubMed Health
A collection of clinical effectiveness reviews and other resources to help consumers and clinicians use and understand clinical research results. These are drawn from the NCBI Bookshelf and PubMed, including published systematic reviews from organizations such as the Agency for Health Care Research and Quality, The Cochrane Collaboration, and others (see complete listing). Links to full text articles are provided when available.

Genes & Disease

http://www.ncbi.nlm.nih.gov/books/NBK22183/

Bookshelf ID: NBK22183

Genes and Disease

National Center for Biotechnology Information (US)

Bethesda (MD): National Center for Biotechnology Information (US); 1998-.

Copyright notice.

Genes and Disease is a collection of articles that discuss genes and the diseases that they cause. These genetic disorders are organized by the parts of the body that they affect. As some diseases affect various body systems, they appear in more than one chapter.

With each genetic disorder, the underlying mutation(s) is discussed, along with clinical features and links to key websites.

Contents

Genes and Disease Table of Contents

http://www.ncbi.nlm.nih.gov/books/NBK22183/

Contents

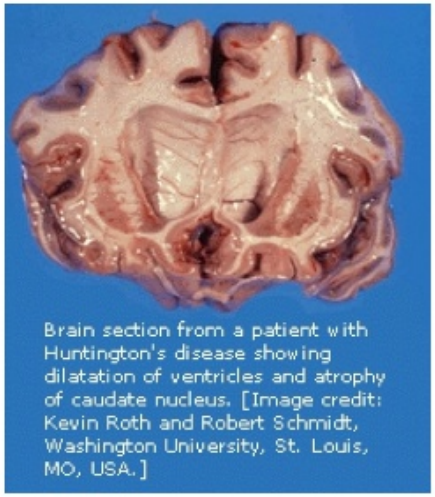
Genes & Disease: Huntington Disease

http://www.ncbi.nlm.nih.gov/books/NBK22226/

Contents | Print View | < Prev | Next >

Bookshelf ID: NBK22226

Huntington disease



Brain section from a patient with Huntington's disease showing dilatation of ventricles and atrophy of caudate nucleus. [Image credit: Kevin Roth and Robert Schmidt, Washington University, St. Louis, MO, USA.]

Huntington disease (HD) is an inherited, degenerative neurological disease that leads to dementia. About 30,000 Americans have HD and about 150,000 more are at risk of inheriting the disease from a parent.

The HD gene, whose mutation results in Huntington disease, was mapped to chromosome 4 in 1983 and cloned in 1993. The mutation is a characteristic expansion of a nucleotide triplet repeat in the DNA that codes for the protein huntingtin. As the number of repeated triplets - CAG (cytosine, adenine, guanine) - increases, the age of onset in the patient decreases. Furthermore, because the unstable trinucleotide repeat can lengthen when passed from parent to child, the age of onset can decrease from one generation to the next. Since people who have those repeats always suffer from Huntington disease, it suggests that the mutation causes a gain-of-function, in which the mRNA or protein takes on a new property or is expressed inappropriately.

Genes and Disease [Internet].
National Center for Biotechnology Information (US).
Bethesda (MD): National Center for Biotechnology Information (US); 1998-.
[Table of Contents Page]

Download

PDF version of this page (265K)

Gene sequence

Genome view see gene locations

Entrez Gene collection of gene-related information

BLink related sequences in different organisms

The literature

Research articles online full text

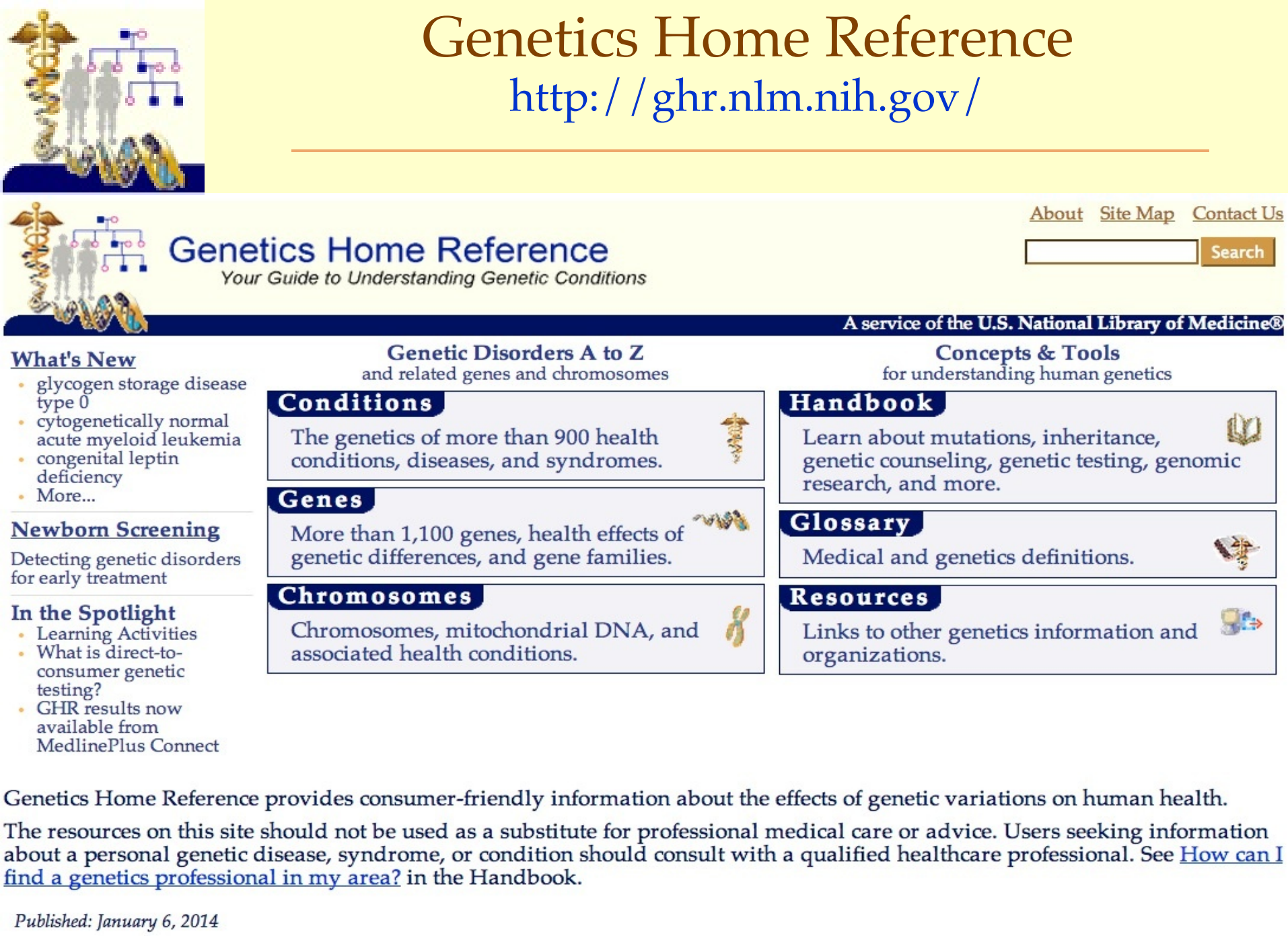
Books online books section

OMIM catalog of human genes and disorders

GeneReviews a medical genetics resource

Genetics Home Reference

http://ghr.nlm.nih.gov/



Huntington Disease in Genetics Home Reference

http://ghr.nlm.nih.gov/condition/huntington-disease

Huntington disease

Print version

- Related Gene(s)
- References
- Quick links to this topic

MedlinePlus
Health information

Genetic and Rare Diseases Information Center
Information about genetic conditions and rare diseases

Additional NIH Resources
National Institutes of Health

Educational resources
Information pages

Patient support
For patients and families

Gene Reviews
Clinical summary

Gene Tests
DNA test labs

ClinicalTrials.gov
Research studies

PubMed
Recent literature

Online Books
Medical and science texts

OMIM
Genetic disorder catalog

On this page: Description Genetic changes Inheritance Treatment Additional information Other names Glossary definitions

Reviewed October 2008

What is Huntington disease?

Huntington disease is a progressive brain disorder that causes uncontrolled movements, emotional problems, and loss of thinking ability (cognition).

Adult-onset Huntington disease, the most common form of this disorder, usually appears in a person's thirties or forties. Early signs and symptoms can include irritability, depression, small involuntary movements, poor coordination, and trouble learning new information or making decisions. Many people with Huntington disease develop involuntary jerking or twitching movements known as chorea. As the disease progresses, these movements become more pronounced. Affected individuals may have trouble walking, speaking, and swallowing. People with this disorder also experience changes in personality and a decline in thinking and reasoning abilities. Individuals with the adult-onset form of Huntington disease usually live about 15 to 20 years after signs and symptoms begin.

A less common, early-onset form of Huntington disease begins in childhood or adolescence. It also involves movement problems and mental and emotional changes. Additional signs of the early-onset form include slow movements, clumsiness, frequent falling, rigidity, slurred speech, and drooling. School performance often declines as thinking and reasoning abilities become impaired. Seizures occur in 30 percent to 50 percent of children with this condition. Early-onset Huntington disease tends to progress more quickly than the adult-onset form; affected individuals usually live 10 to 15 years after signs and symptoms appear.

How common is Huntington disease?

Huntington disease affects an estimated 3 to 7 per 100,000 people of European ancestry. The disorder appears to be less common in some other populations, including people of Japanese, Chinese, and African descent.

MedlinePlus

http://www.nlm.nih.gov/medlineplus/

Huntington's in Medline Plus

MedlinePlus
Trusted Health Information for You

A service of the U.S. National Library of Medicine
NIH National Institutes of Health

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Search MedlinePlus: Huntingtons GO

Health Topics | Drugs & Supplements | Videos & Cool Tools | ESPAÑOL

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- Medical Encyclopedia (13)
- Videos and Tutorials
- News (2)
- MedlinePlus Magazine (6)
- Other Resources (10)
- Multiple Languages

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- Brain (12)
- Research (12)
- Harvard School of Public Health (15)
- Chorea (8)
- America (8)
- Dementia (11)
- Multiple | Parkinson (7)
- Huntington Beach, CA (6)
- Nerve Diseases (5)

more

Huntington's Disease Search Help

Huntington's disease (HD) is an inherited disease that causes certain nerve cells in the brain to waste away. People are born with the defective gene, but symptoms usually don't appear until middle age. Early symptoms of HD may include uncontrolled movements, clumsiness or balance problems. Later, HD can take away the ability to walk, talk or swallow. Some people stop recognizing family members. Others are aware of their environment and are able to express emotions.

If one of your parents has Huntington's disease, you have a 50-50 chance of getting it. A blood test can tell if you have the HD gene and will develop the disease. Genetic counseling can help you weigh the risks and benefits of taking the test. (Read more)

Results **1 - 10** of **129** for **Huntingtons**

1. **Huntington's Disease** (National Library of Medicine)
   Huntington's disease (HD) is an inherited disease that causes certain nerve cells in the brain to waste ... express emotions. If one of your parents has **Huntington**'s disease, you have a 50-50 chance of ...
   www.nlm.nih.gov/medlineplus/huntingtonsdisease.html - Health Topics
2. **Huntington's disease**
   Huntington chorea ... American doctor George **Huntington** first described the disorder in 1872. **Huntington**'s disease is caused by a genetic defect on chromosome #4. The defect ...
   www.nlm.nih.gov/medlineplus/ency/article/000770.htm - Medical Encyclopedia
3. Genetics Home Reference: **Huntington** disease NIH (National Library of

g 2016

Huntington Disease Gene Review

http://www.ncbi.nlm.nih.gov/books/NBK1305/

Huntington Disease

Huntington Chorea

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Initial Posting: October 23, 1998; Last Update: April 22, 2010.

GeneReviews [Internet].
Pagon RA, Bird TC, Dolan CR, et al., editors.
Seattle (WA): University of Washington, Seattle; 1993-.
[Table of Contents Page]

Summary

Go to: Top▲

Disease characteristics. Huntington disease (HD) is a progressive disorder of motor, cognitive, and psychiatric disturbances. The mean age of onset is 35 to 44 years and the median survival time is 15 to 18 years after onset.

Diagnosis/testing. The diagnosis of HD rests on positive family history, characteristic clinical findings, and the detection of an expansion of 36 or more CAG trinucleotide repeats in *HTT*.

Management. *Treatment of manifestations:* pharmacologic therapy including typical neuroleptics (haloperidol), atypical

GeneReviews Links

GeneTests Home Page
GeneReviews Advanced Search
About GeneTests

Gene Reviews of Huntington Disease

http://www.ncbi.nlm.nih.gov/books/NBK1305/

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National Institutes of Health

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GeneReviews by Title Disable Glossary Links Print View < Prev Next >

Bookshelf ID: NBK1305 PMID: 20301482

Huntington Disease

Synonym: Huntington Chorea

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Initial Posting: October 23, 1998; Last Update: April 22, 2010.

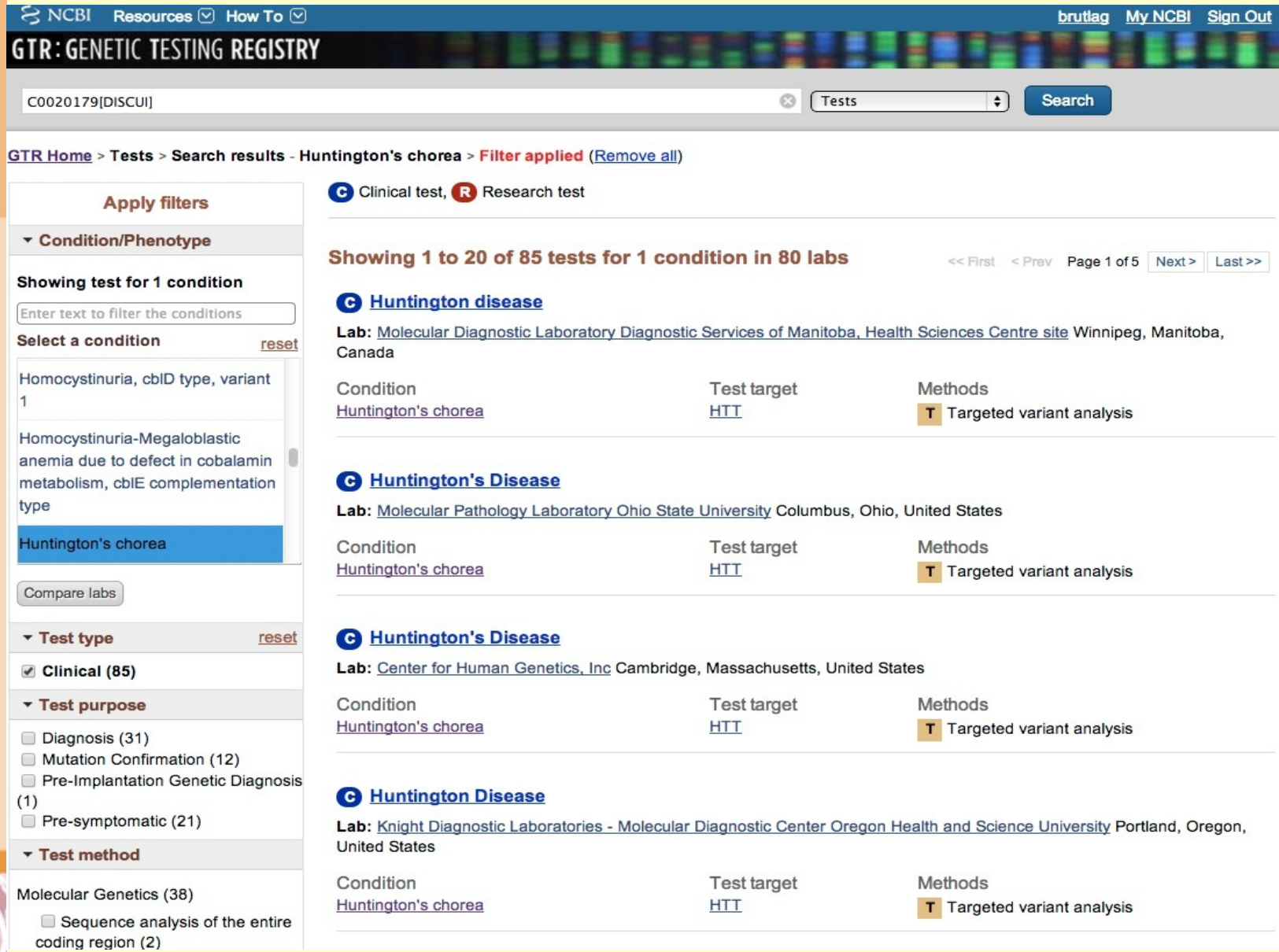
GeneReviews [Internet].
Pagon RA, Bird TD, Dolan CR, editors.
Seattle (WA): University of Wash Seattle; 1993-.
[All GeneReviews]

In this GeneReview

Summary

Genetic Testing Registry for Huntington

http://www.ncbi.nlm.nih.gov/gtr/



Clinical Trials for Huntington
http://clinicaltrials.gov/

ClinicalTrials.gov
A service of the U.S. National Institutes of Health

ClinicalTrials.gov is a registry and results database of publicly and privately supported clinical studies of human participants conducted around the world. Learn more about clinical studies and about this site, including relevant history, policies, and laws.

Find Studies | About Clinical Studies | Submit Studies | Resources | About This Site

ClinicalTrials.gov currently lists 159,585 studies with locations in all 50 states and in 185 countries.

Text Size

Search for Studies
Example: "Heart attack" AND "Los Angeles"
Huntington Diseas [Search]
Advanced Search | See Studies by Topic
See Studies on a Map

Search Help
- How to search
- How to find results of studies
- How to read a study record

Locations of Recruiting Studies

Total N = 31,986 studies
Data as of January 21, 2014

- See more trends, charts, and maps

For Patients & Families
- How to find studies
- See studies by topic
- Learn about clinical studies
- Learn more…

For Researchers
- How to submit studies
- Download content for analysis
- About the results database
- Learn more…

For Study Record Managers
- Why register?
- How to register study records
- FDAAA 801 Requirements
- Learn more…

Learn More
- ClinicalTrials.gov Online Training
- Glossary of common site terms

For the Press
Using our RSS Feeds

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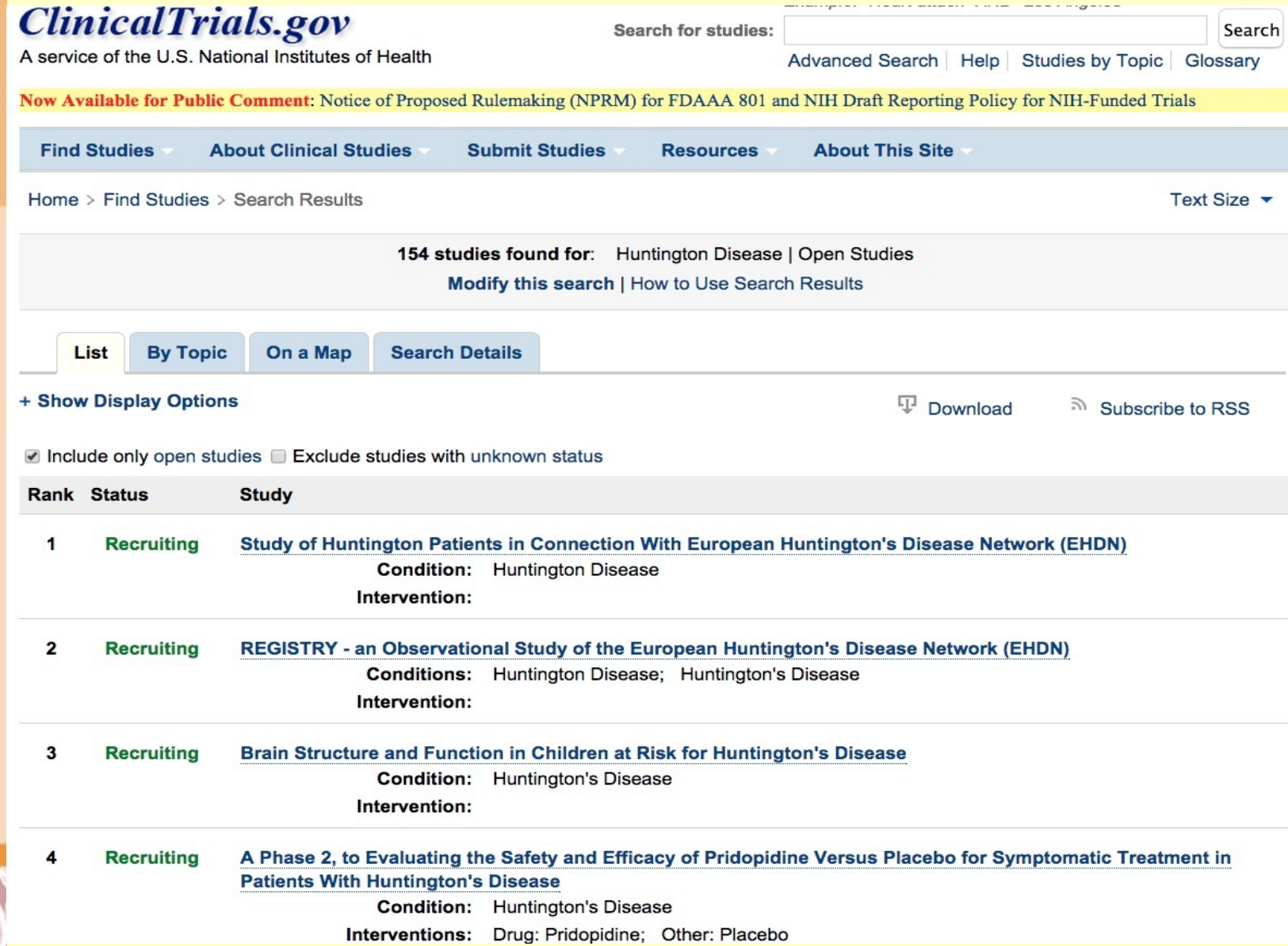
Clinical Trials for Huntington Disease

http://clinicaltrials.gov/



ClinicalTrials.gov
A service of the U.S. National Institutes of Health

Search for studies: Search

Advanced Search | Help | Studies by Topic | Glossary

Now Available for Public Comment: Notice of Proposed Rulemaking (NPRM) for FDAAA 801 and NIH Draft Reporting Policy for NIH-Funded Trials

Find Studies | About Clinical Studies | Submit Studies | Resources | About This Site

Home > Find Studies > Search Results

Text Size

154 studies found for: Huntington Disease | Open Studies

Modify this search | How to Use Search Results

List | By Topic | On a Map | Search Details

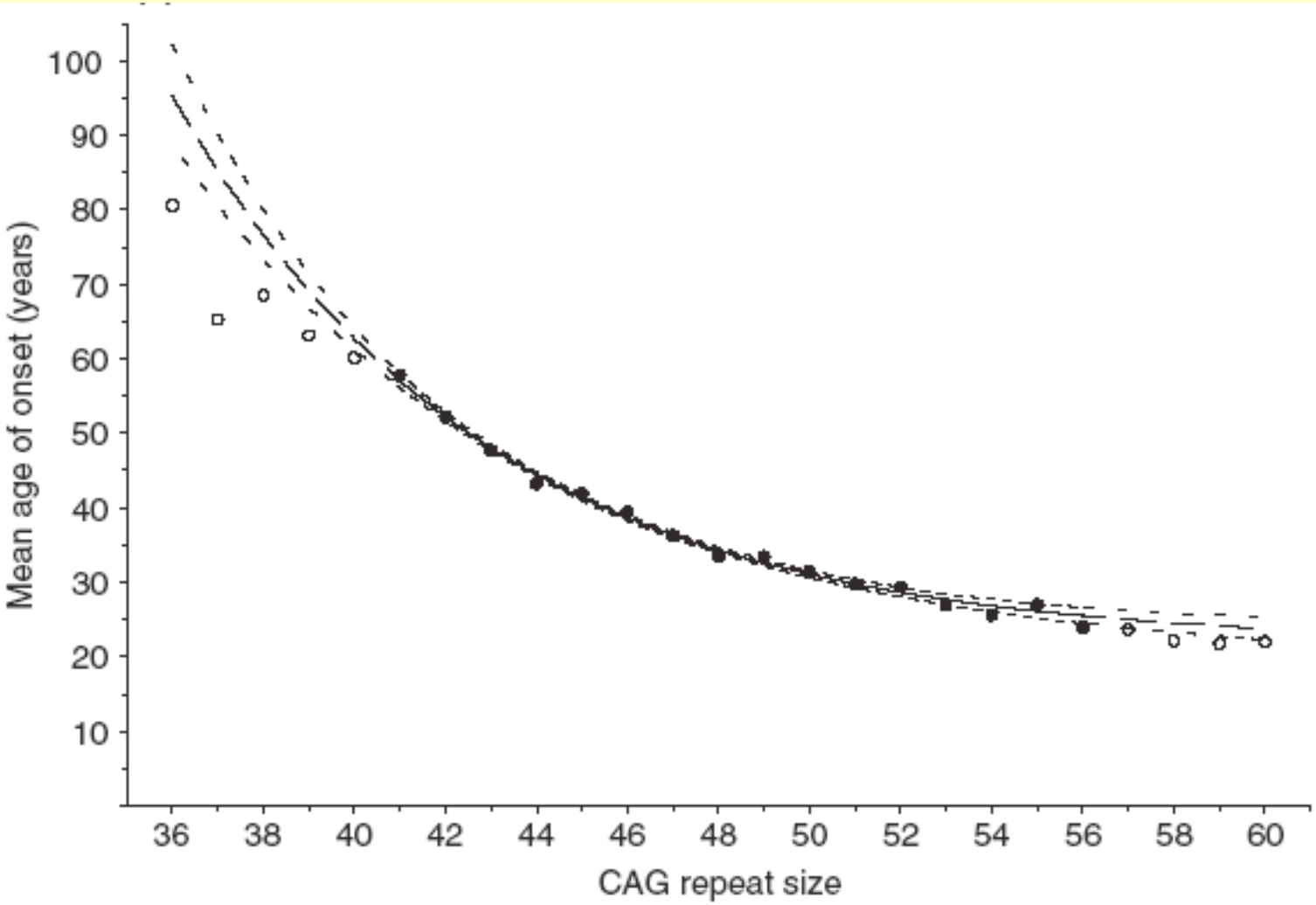
+ Show Display Options

Download | Subscribe to RSS

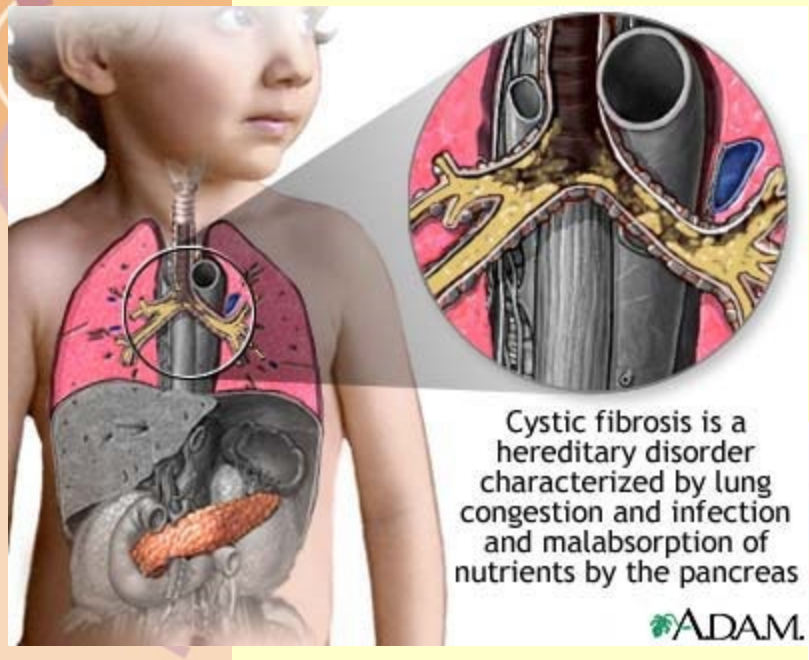
Include only open studies | Exclude studies with unknown status

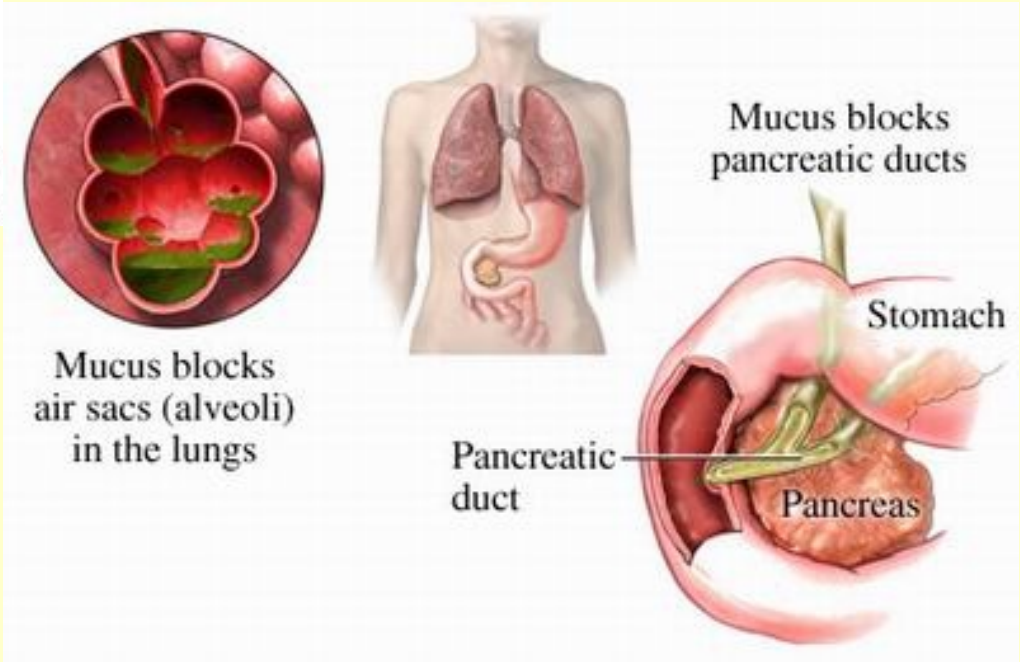
Rank	Status	Study
1	Recruiting	Study of Huntington Patients in Connection With European Huntington's Disease Network (EHDN) **Condition:** Huntington Disease **Intervention:**
2	Recruiting	REGISTRY - an Observational Study of the European Huntington's Disease Network (EHDN) **Conditions:** Huntington Disease; Huntington's Disease **Intervention:**
3	Recruiting	Brain Structure and Function in Children at Risk for Huntington's Disease **Condition:** Huntington's Disease **Intervention:**
4	Recruiting	A Phase 2, to Evaluating the Safety and Efficacy of Pridopidine Versus Placebo for Symptomatic Treatment in Patients With Huntington's Disease **Condition:** Huntington's Disease **Interventions:** Drug: Pridopidine; Other: Placebo

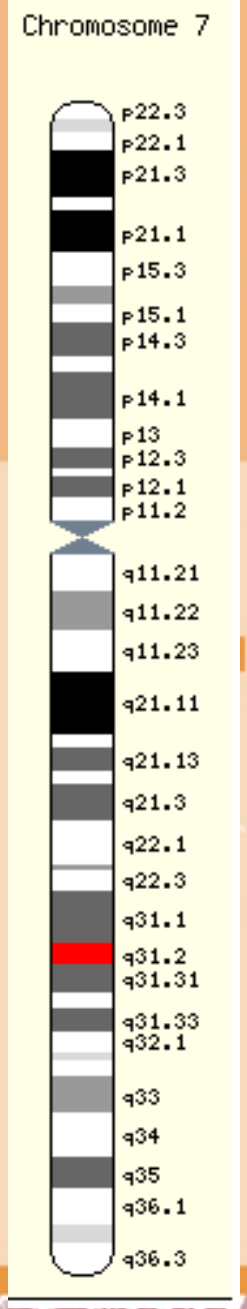
Age of Onset and Repeat Length



Cystic Fibrosis



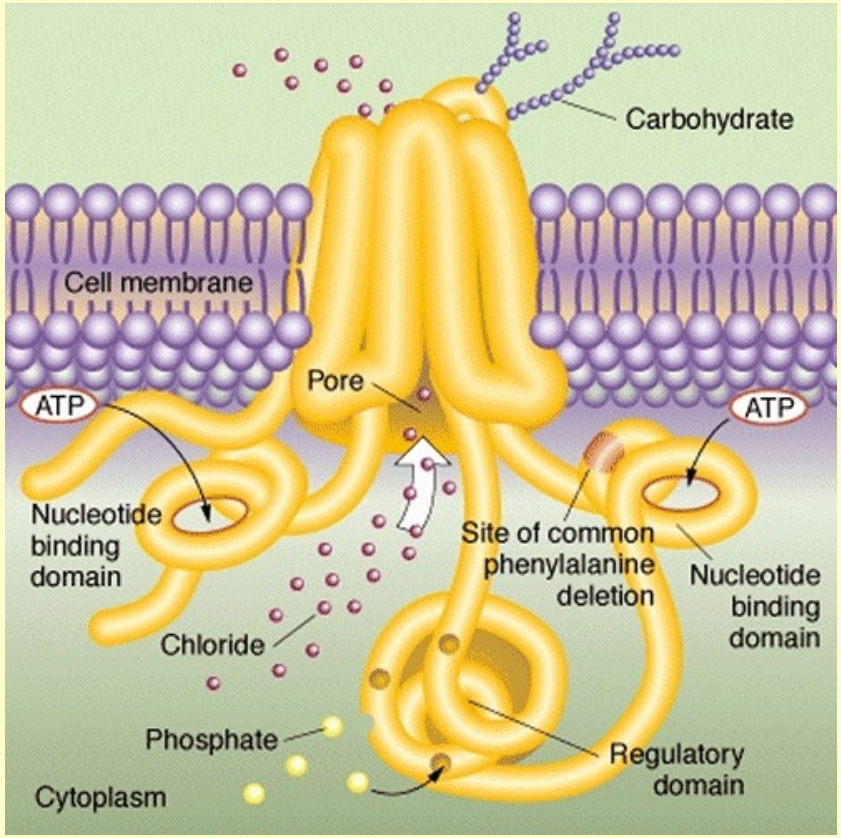




Cystic Fibrosis

- Autosomal (chromosome 7q31.2) recessive
- 3% of North American Caucasians are carriers
- 1.5% of African Americans are carriers
- Inhibits many bodily secretions
  - Pancreatic digestive enzymes
  - Sweat glands
  - Lung mucosa in alveoli and bronchi
  - Infertility in males (>97%)
- Caused by mutations in the CFTR gene that encodes a chloride ion channel that pumps chloride ion and water out of cells.

Cystic Fibrosis Transmembrane Conductance Regulator



Mutations Causing Cystic Fibrosis

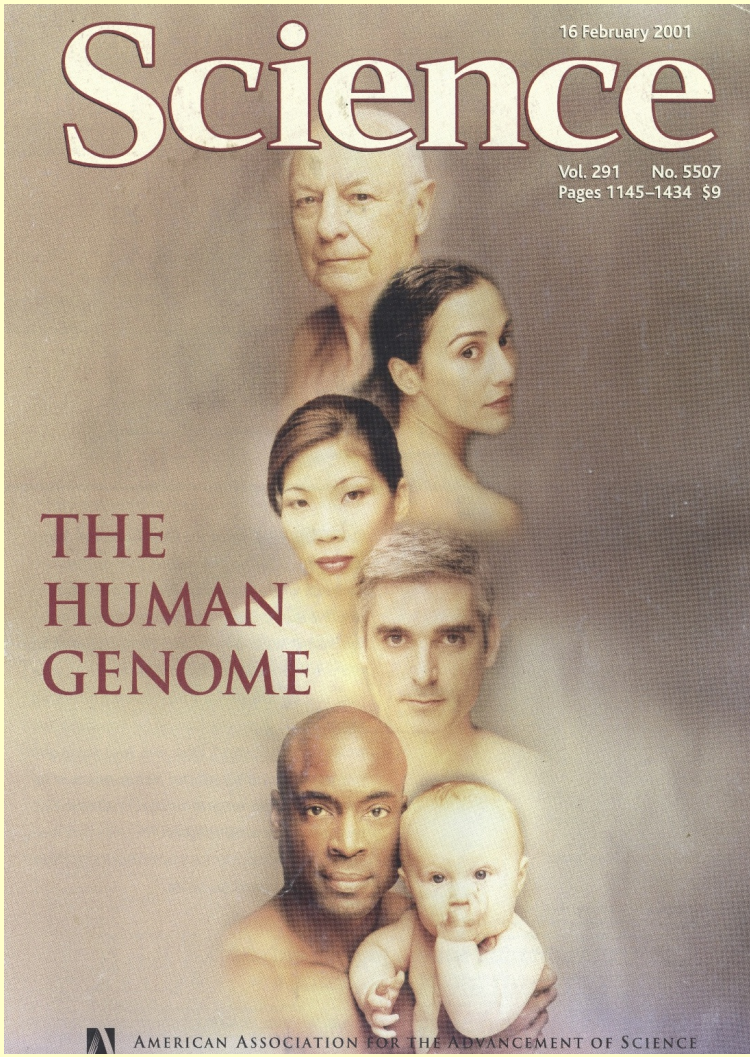
Mutation	Relative Frequency	Mutation Functional Class [1]
ΔF508	66.0%	II
G542X	2.4%	I
G551D	1.6%	III
N1303Lys	1.3%	II
W1282X	1.2%	I
R553X	0.7%	I
621+1G>T	0.7%	I
1717-1G>A	0.6%	I
R117H	0.3%	IV
R1162X	0.3%	Not clear [4]

Population Group	Approximate Carrier Frequency
Ashkenazi Jewish	1:29
North American Caucasian	1:28
African American	1:61

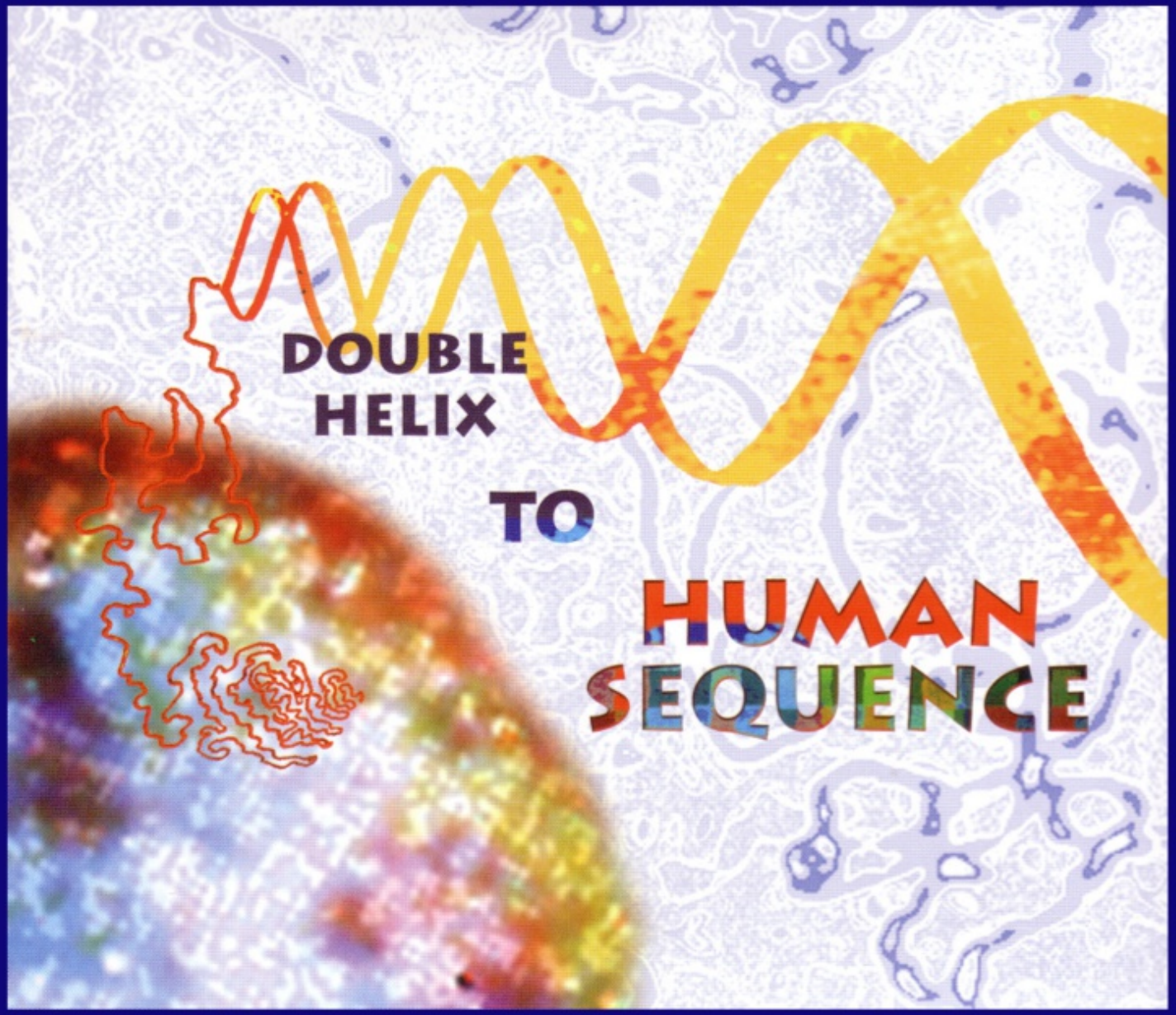
Human Genome First draft February 2001

Public Effort

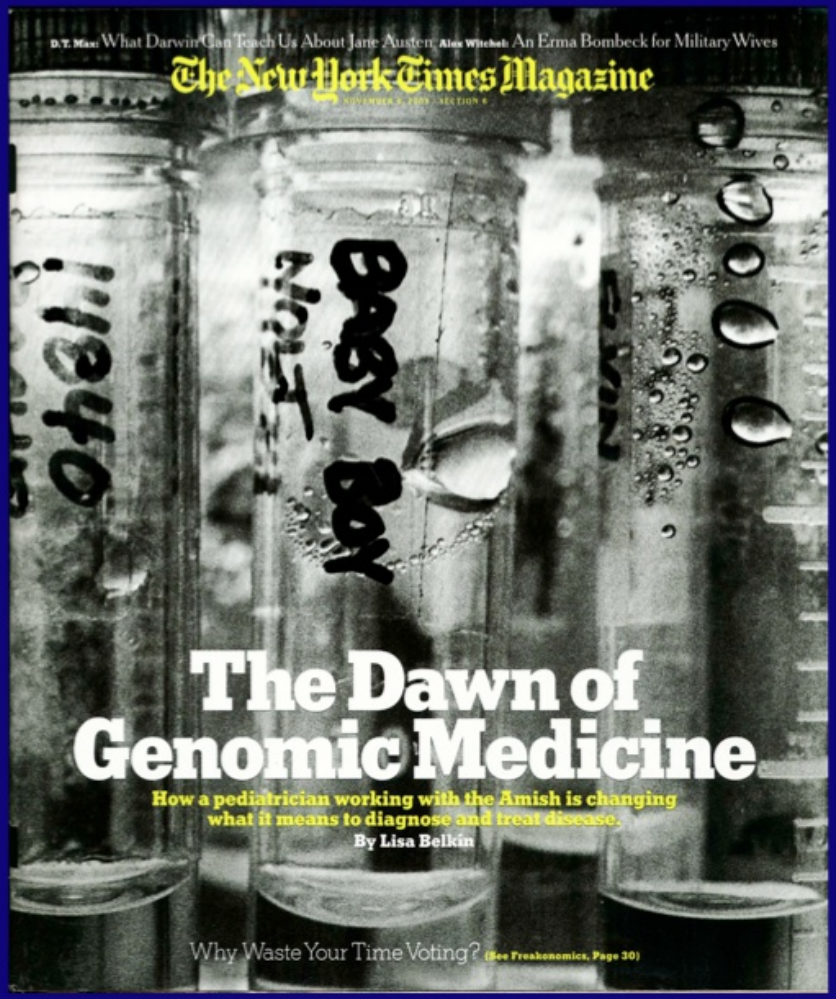
Private Effort (Celera)



April, 2003 Completion



Genomic Medicine



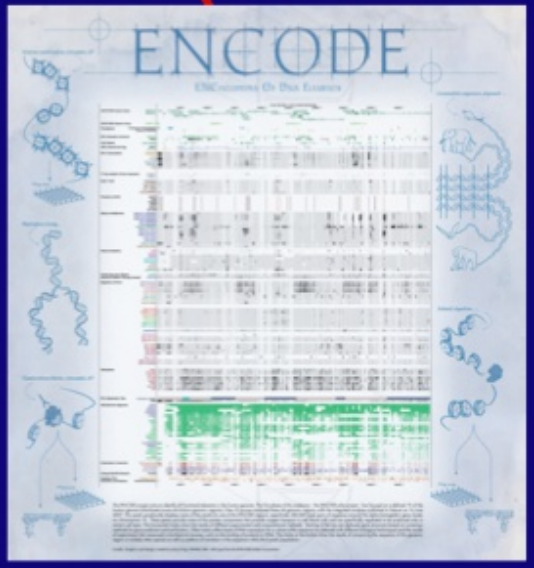
Genomic Medicine

Healthcare tailored to the individual based on genomic information

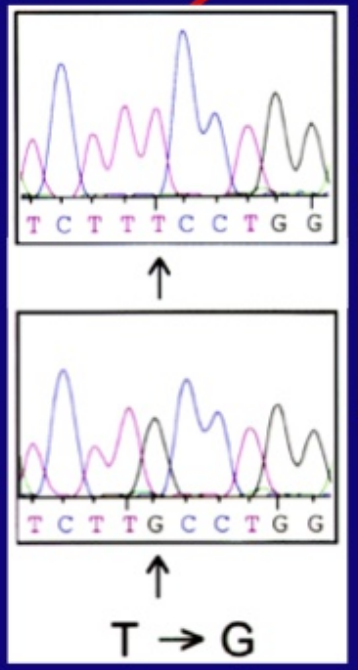
The Pathway to Genomic Medicine

Interpreting the Human Genome Sequence

HGP



Implicating Genetic Variants with Human Disease



Realization of Genomic Medicine

National Human Genome Research Institute

http://www.genome.gov/

genome.gov
National Human Genome Research Institute
National Institutes of Health

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Highlights RSS

Carla Easter to lead NHGRI Education and Community Involvement Branch

Carla Easter, Ph.D., a biologist and science educator, has been named chief of the National Human Genome Research Institute (NHGRI) Education and Community Involvement Branch (ECIB). The branch is a part of NHGRI's Division of Policy, Communication and Education (DPCE). She will lead the division's program of genomics education and outreach activities that engage a variety of communities, from pre-college students, to teachers to members of the general public. **Read more**

The Genomics Landscape

Next Phase: NHGRI's Genome Sequencing Program

Large-scale genome sequencing has been a central component of NHGRI's Extramural Research Program since the NHGRI's inception, starting with the Human Genome Project. In this month's *The Genomics Landscape*, I describe the next phase of NHGRI's Genome Sequencing Program and highlight additional items that I hope will be of interest to you.
Read more

For You

- Students
- Educators
- Patients
- Health Professionals
- Grant Applicants
- News Media

GenomeTV View All

Newsroom RSS

NIH grants aim to decipher the language of gene regulation
January 5, 2015

Researchers conduct comprehensive genomic study of sub-Saharan Africans
December 3, 2014

New comprehensive view of the mouse genome finds many similarities and striking differences

Quick Links

Genome: Unlocking Life's Code

http://www.genome.gov/Smithsonian/

genome.gov
National Human Genome Research Institute
National Institutes of Health

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Research Funding | Research at NHGRI | Health | **Education** | Issues in Genetics | Newsroom | Careers & Training | About | For You

Home > Education > Smithsonian NHGRI Genome Exhibition

Education

- All About The Human Genome Project (HGP)
- Education Archive
- Fact Sheets
- Genetic Education Resources for Teachers
- National DNA Day
- Online Genetics Education Resources
- Smithsonian NHGRI Genome Exhibition
- Talking Glossary
- Understanding the Human Genome Project

Genome: Unlocking Life's Code

Share Print

The Genome Unlocking Life's Code Exhibition

On June 14, 2013, the Smithsonian Institution in Washington, D.C. opened the high-tech, high-intensity exhibition *Genome: Unlocking Life's Code* to celebrate the 10th anniversary of researchers producing the first complete human genome sequence - the genetic blueprint of the human body - in April 2003. The exhibition is a collaboration between the Smithsonian's National Museum of Natural History (NMNH) and the National Human Genome Research Institute (NHGRI) of the National Institutes of Health.

2007 SCIENTIFIC BREAKTHROUGH OF THE YEAR

Science Magazine, December 21, 2007

"It's all about me!"

Single Nucleotide Polymorphisms (SNPs)

The Great Wave of GWAS Studies

http://www.genome.gov/gwastudies/

Hokusai, K. *The Great Wave*

Published Genome-Wide Associations through 12/2013

Published GWA at $p \leq 5X10^{-8}$ for 17 trait categories

NHGRI GWA Catalog
www.genome.gov/GWAStudies
www.ebi.ac.uk/fgpt/gwas/

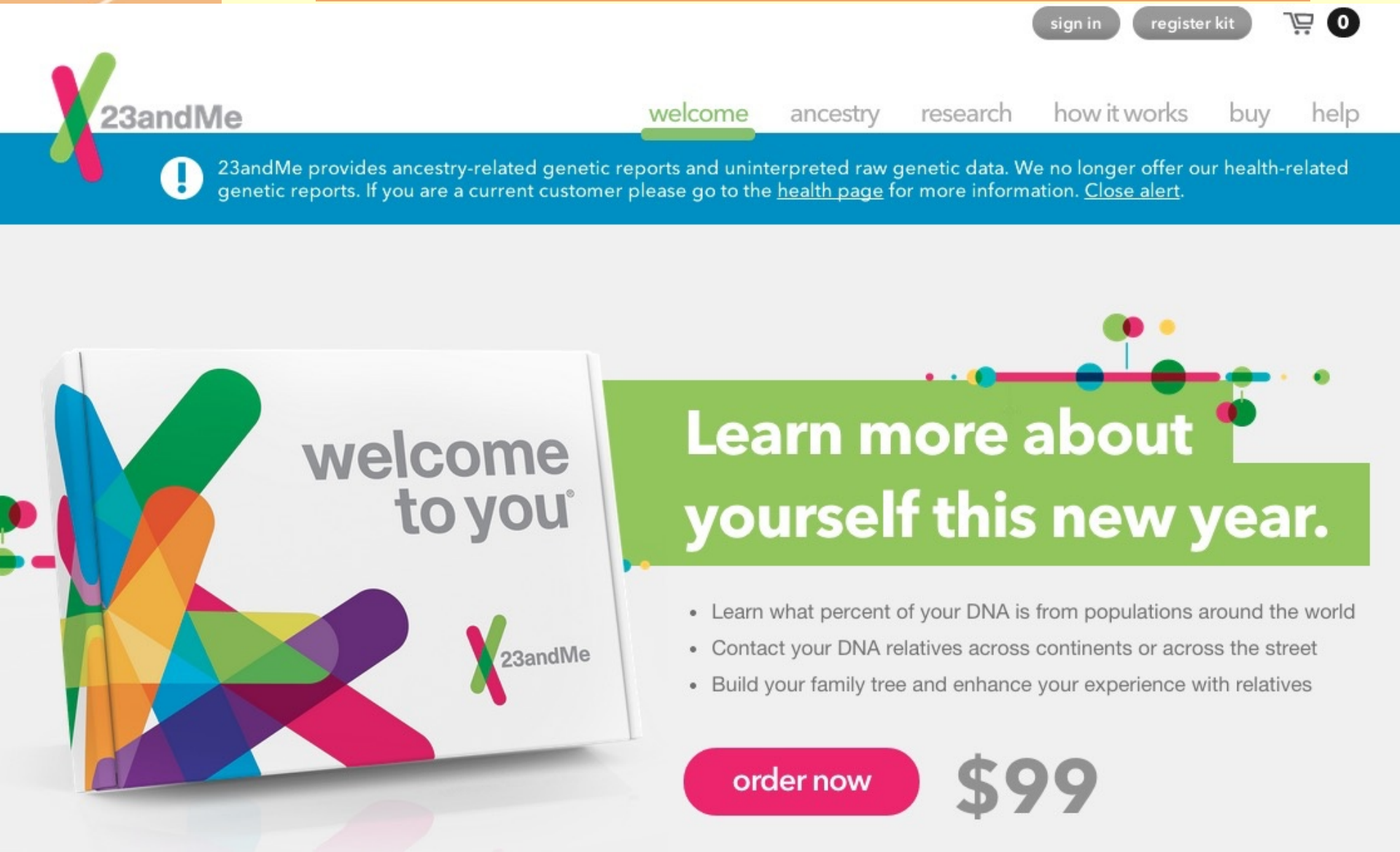
National Human Genome Research Institute

EMBL-EBI


Abdominal aortic aneurysm
Acute lymphoblastic leukemia
Adhesion molecules
Adiponectin levels
Age-related macular degeneration
AIDS progression
Alcohol dependence
Alopecia areata
Alzheimer disease
Amyloid A levels
Amyotrophic lateral sclerosis
Angiotensin-converting enzyme activity
Ankylosing spondylitis
Arterial stiffness
Asparagus anosmia
Asthma
Atherosclerosis in HIV
Atrial fibrillation
Attention deficit hyperactivity disorder
Autism
Basal cell cancer
Behcet's disease
Bipolar disorder
Biliary atresia
Bilirubin
Bitter taste response
Birth weight
Bladder cancer
Bleomycin sensitivity
Blond or brown hair
Blood pressure
Blue or green eyes
BMI, waist circumference
Bone density
Breast cancer
C-reactive protein
Calcium levels
Cardiac structure/function
Cardiovascular risk factors
Carnitine levels
Carotenoid/tocopherol levels
Celiac disease
Celiac disease and rheumatoid arthritis
Cerebral atrophy measures
Chronic lymphocytic leukemia
Chronic myeloid leukemia
Cleft lip/palate
Coffee consumption
Cognitive function
Conduct disorder
Colorectal cancer
Corneal thickness
Coronary disease
Creutzfeldt-Jakob disease
Crohn's disease
Crohn's disease and celiac disease
Cutaneous nevi
Cystic fibrosis severity
Dermatitis
DHEA-s levels
Diabetic retinopathy
Dilated cardiomyopathy
Drug-induced liver injury
Drug-induced liver injury (amoxicillin-clavulanate)
Endometrial cancer
Endometriosis
Eosinophil count
Eosinophilic esophagitis
Erectile dysfunction and prostate cancer treatment
Erythrocyte parameters
Esophageal cancer
Essential tremor
Exfoliation glaucoma
Eye color traits
F cell distribution
Fibrinogen levels
Folate pathway vitamins
Follicular lymphoma
Fuch's corneal dystrophy
Freckles and burning
Gallstones
Gastric cancer
Glioma
Glycemic traits
Hair color
Hair morphology
Handedness in dyslexia
HDL cholesterol
Heart failure
Heart rate
Height
Hemostasis parameters
Hepatic steatosis
Hepatitis
Hepatocellular carcinoma
Hirschsprung's disease
HIV-1 control
Hodgkin's lymphoma
Homocysteine levels
Hypospadias
Idiopathic pulmonary fibrosis
IFN-related cytopeni
IgA levels
IgE levels
Inflammatory bowel disease
Insulin-like growth factors
Intracranial aneurysm
Iris color
Iron status markers
Ischemic stroke
Juvenile idiopathic arthritis
Keloid
Kidney stones
LDL cholesterol
Leprosy
Leptin receptor levels
Liver enzymes
Longevity
LP (a) levels
LpPLA(2) activity and mass
Lung cancer
Magnesium levels
Major mood disorders
Malaria
Male pattern baldness
Mammographic density
Matrix metalloproteinase levels
MCP-1
Melanoma
Menarche & menopause
Meningococcal disease
Metabolic syndrome
Migraine
Moyamoya disease
Multiple sclerosis
Myeloproliferative neoplasms
Myopia (pathological)
N-glycan levels
Narcolepsy
Nasopharyngeal cancer
Natriuretic peptide levels
Neuroblastoma
Nicotine dependence
Obesity
Open angle glaucoma
Open personality
Optic disc parameters
Osteoarthritis
Osteoporosis
Otosclerosis
Other metabolic traits
Ovarian cancer
Pancreatic cancer
Pain
Paget's disease
Panic disorder
Parkinson's disease
Periodontitis
Peripheral arterial disease
Personality dimensions
Phosphatidylcholine levels
Phosphorus levels
Photic sneeze
Phytosterol levels
Platelet count
Polycystic ovary syndrome
Primary biliary cirrhosis
Primary sclerosing cholangitis
PR interval
Progranulin levels
Progressive supranuclear palsy
Prostate cancer
Protein levels
PSA levels
Psoriasis
Psoriatic arthritis
Pulmonary funct. COPD
QRS interval
QT interval
Quantitative traits
Recombination rate
Red vs.non-red hair
Refractive error
Renal cell carcinoma
Renal function
Response to antidepressants
Response to antipsychotic therapy
Response to carbamazepine
Response to clopidogrel therapy
Response to hepatitis C treat
Response to interferon beta therapy
Response to metaformin
Response to statin therapy
Restless legs syndrome
Retinal vascular caliber
Rheumatoid arthritis
Ribavirin-induced anemia
Schizophrenia
Serum metabolites
Skin pigmentation
Smoking behavior
Speech perception
Sphingolipid levels
Statin-induced myopathy
Stroke
Sudden cardiac arrest
Suicide attempts
Systemic lupus erythematosus
Systemic sclerosis
T-tau levels
Tau AB1-42 levels
Telomere length
Testicular germ cell tumor
Thyroid cancer
Thyroid volume
Tooth development
Total cholesterol
Triglycerides
Tuberculosis
Type 1 diabetes
Type 2 diabetes
Ulcerative colitis
Urate
Urinary albumin excretion
Urinary metabolites
Uterine fibroids
Venous thromboembolism
Ventricular conduction
Vertical cup-disc ratio
Vitamin B12 levels
Vitamin D insuffiency
Vitiligo
Warfarin dose
Weight
White cell count
White matter hyperintensity
YKL-40 levels

Personal Genomics: 23andMe

https://www.23andme.com/



Navigenics

http://www.navigenics.com/

Navigenics

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(866) 522-1585 / +1 (650) 585-7743

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Gene-ius.
A smart way to look at your health.

Navigenics is the leading provider of clinically guided genetic analysis. Our goal is to empower you with genetic insights to help motivate you to improve your health. We also put a premium on privacy, keeping you in control of your genetic information.

New: Your genes, your medications

Will a new medication be effective for you? Will a treatment cause serious side effects? Now, genetic insights from Navigenics can help you and your doctor select medications that may be right for your genetic makeup.

Learn More

Success Stories

"We hear a lot of different – and sometimes conflicting – opinions about how to take care of our health. I'm very excited about receiving only the most relevant information to me, based on my DNA."

More Success Stories

Find a physician

Find a physician in your area who offers the Navigenics genetic testing services, so you can focus your health plan on prevention.

Find a physician now >

Next Steps

- I'm new to Navigenics
- Adding to family history
- Genetic testing: Myths and truths
- Genetic knowledge can help you

For Physicians

- Free educational webinars
- More personalized care
- Genetic counselors for patients and you
- Foundation that rests on strong science

Our Collaborators

PositScience Your brain will thank you. | MEDCAN CLINIC | Scripps Health Foundation

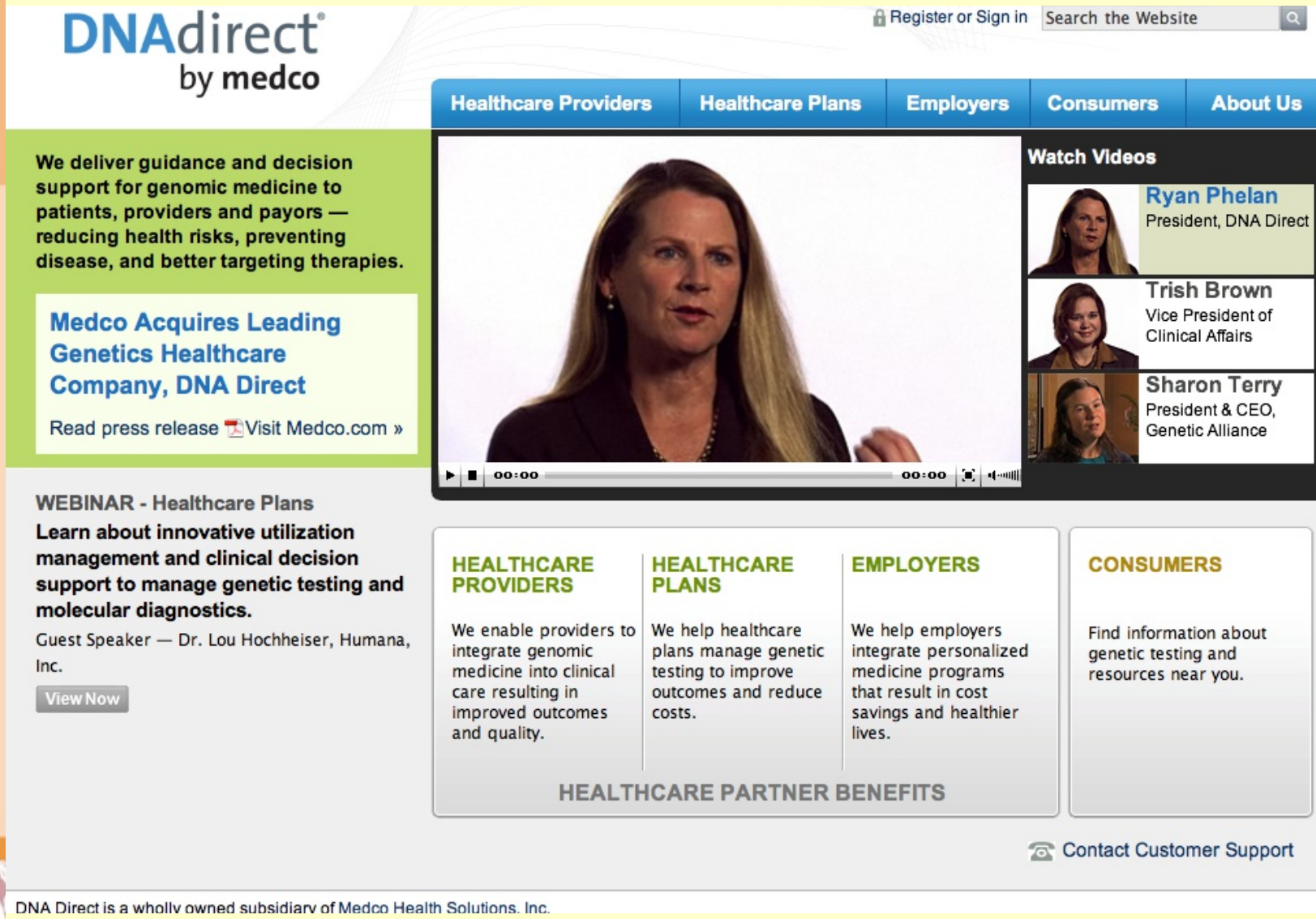
View all collaborators >

Latest Headlines Working with regulators -- the road ahead

Read our blog >

DNAdirect

DNAdirect: Clinical Genetic Testing



http://www.dnadirect.com/web/

INFORMED Medical Decisions

http://informeddna.com/

PMWC 2013
PERSONALIZED MEDICINE WORLD CONFERENCE
- SILICON VALLEY -

Personalized Medicine World Conference

http://2015sv.pmwcintl.com/

PMWC
PERSONALIZED MEDICINE WORLD CONFERENCE
Silicon Valley 2015

Jan. 26-28
Mountain View, CA

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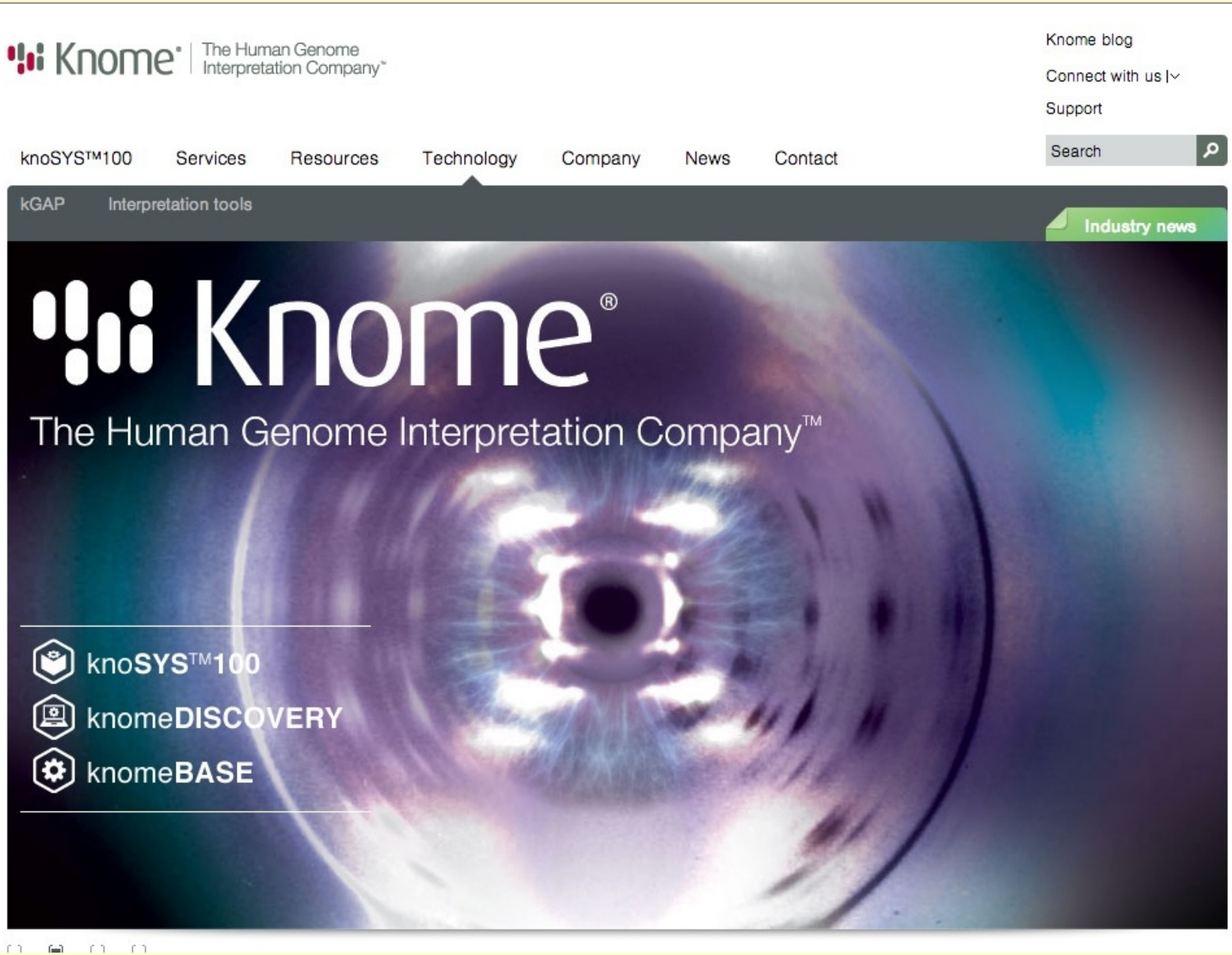
Session Themes

Newborn & Prenatal Dx
Clinical Methodologies & Applications of NGS
Applications of NGS for Non-oncology
Cardiovascular Disease & Biomarkers, Immunotherapy
Consumerism & Crowdsourcing
Coverage & Reimbursement
Commercialization of Genetics of Longevity & Aging
Applying Complimentary Technologies Towards Biomarker Discovery
Large Sequence Data Analysis & Clinical Interpretation
Navigating Reimbursement for Oncology Patients and Care Teams
Pharma Perspective on PM
Risk-Reward & Innovation
Morality, Ethics & Public Law In PM
Clinical Trials in the Era of PM
Genomic Profiling Moving Into Routine Clinical Care
PMWC Most Promising Company Competition

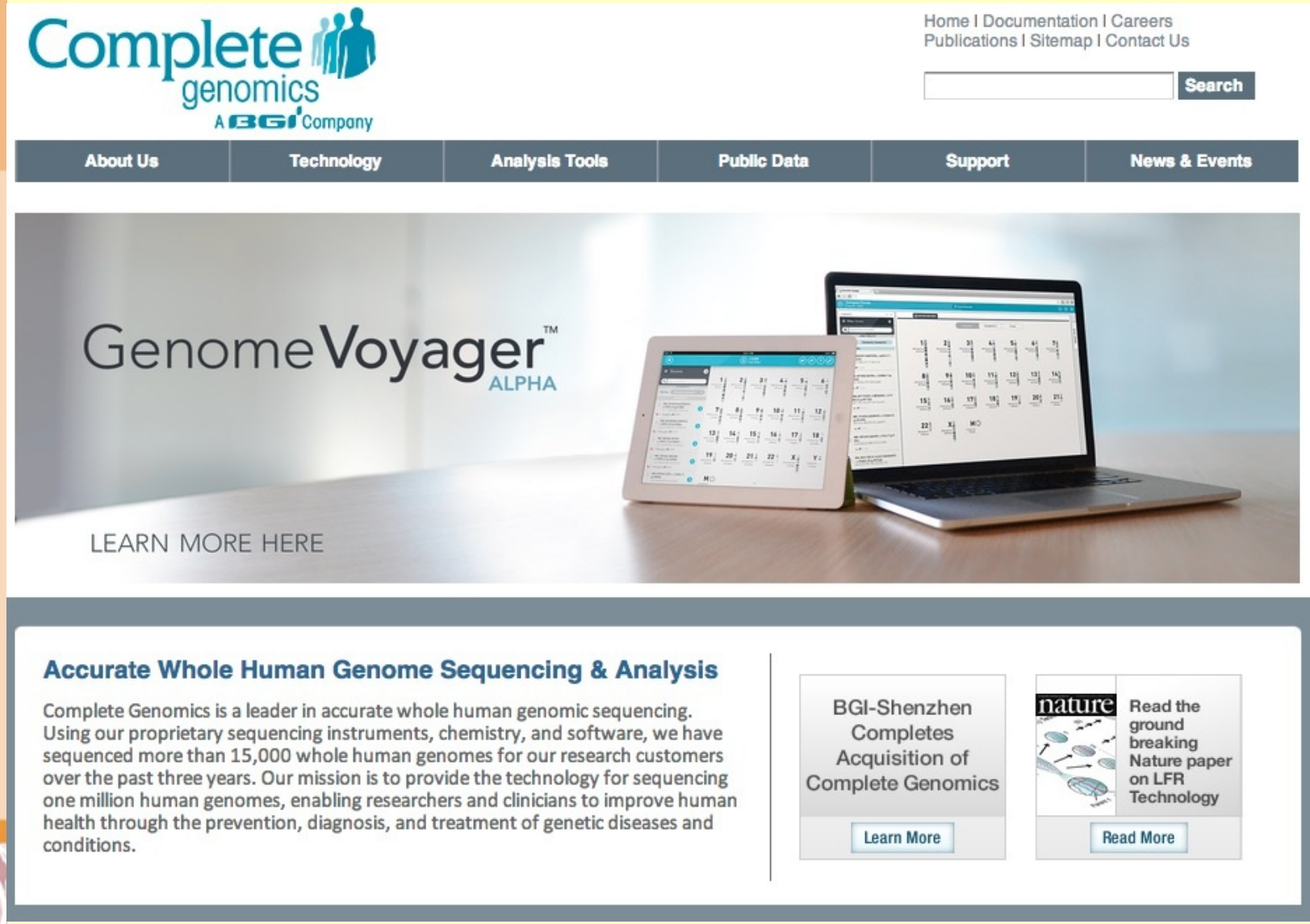
Knome

http://www.knome.com/



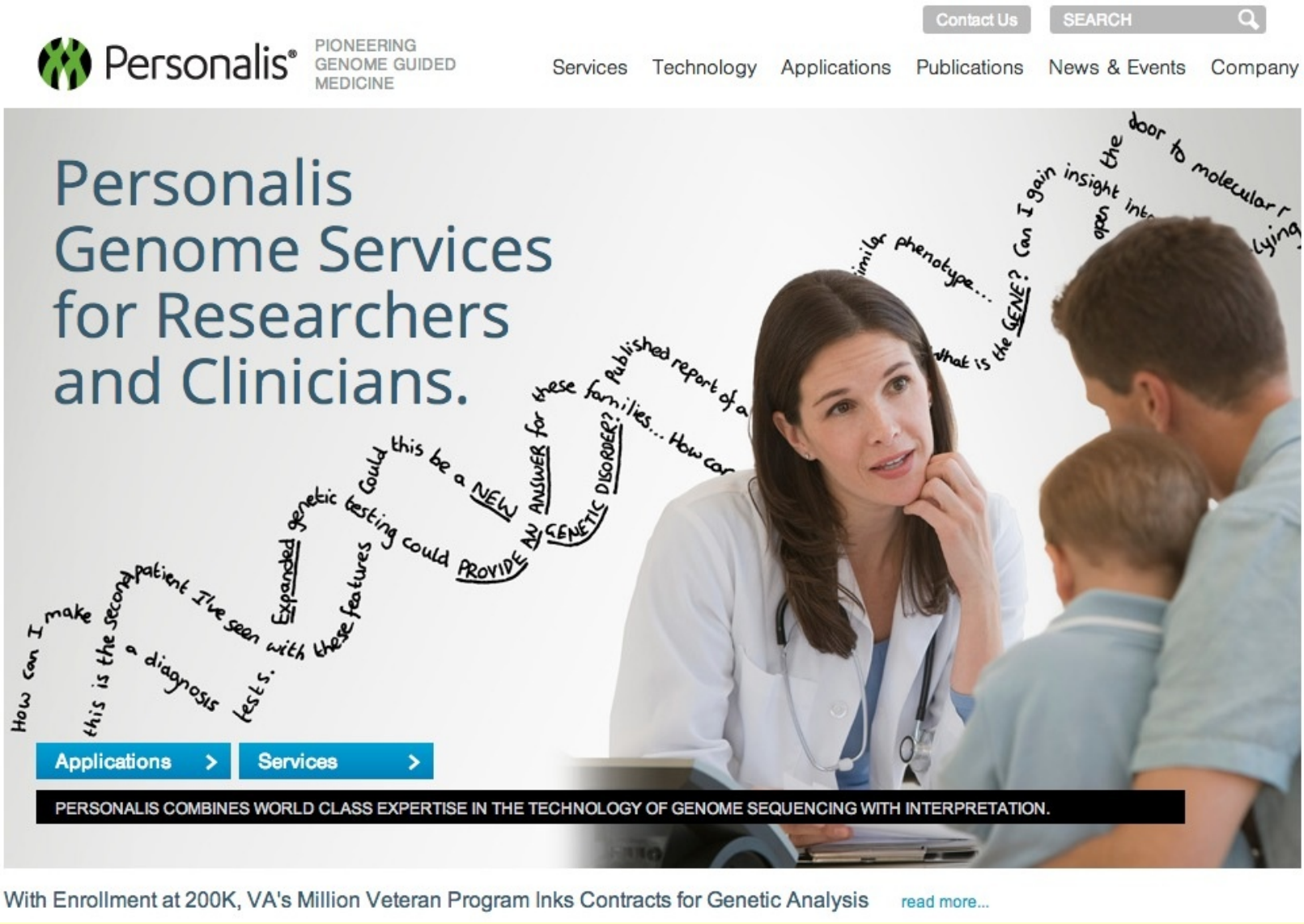
Complete Genomics

http://www.completegenomics.com/



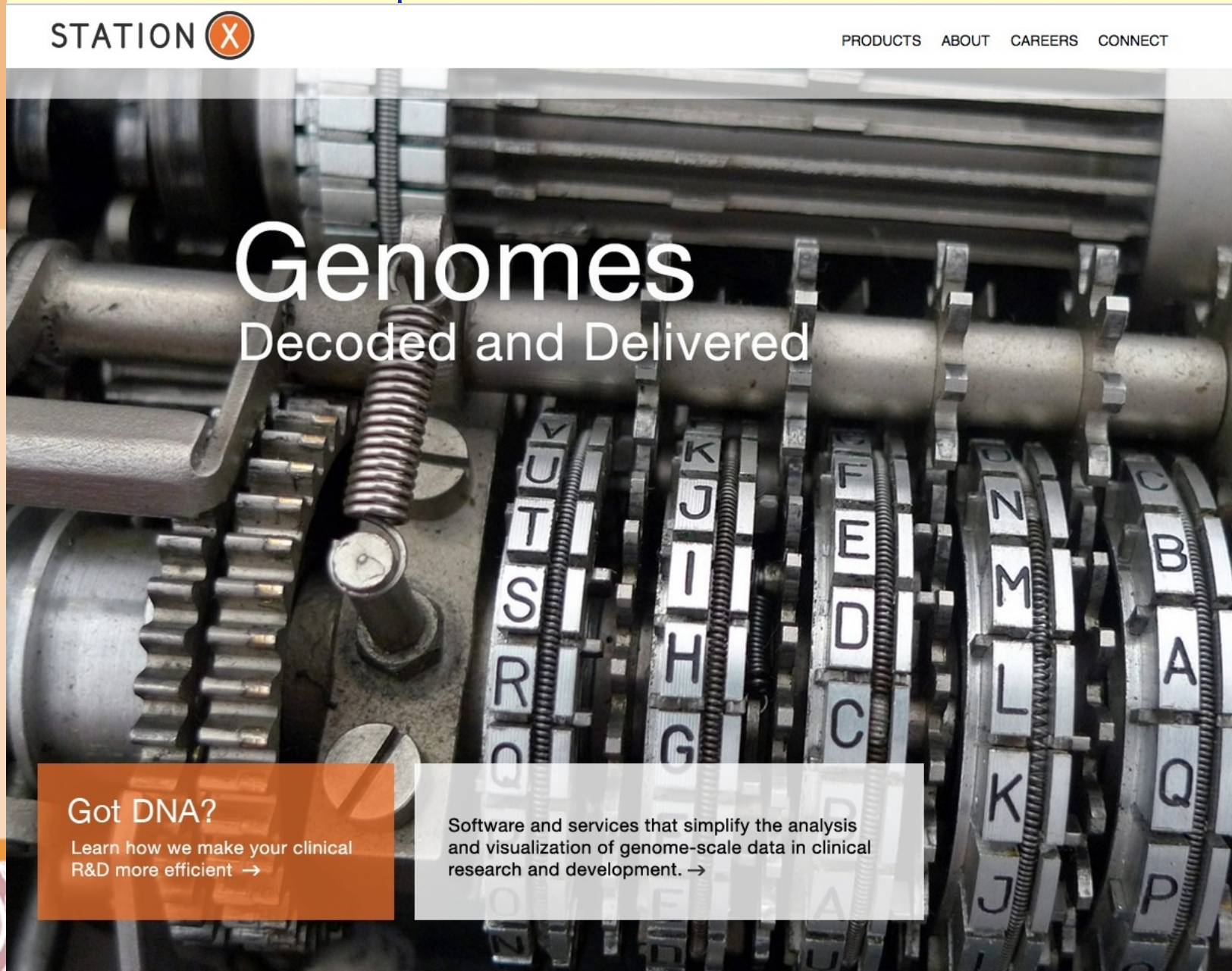
Personalis

http://www.personalis.com/



StationX

http://www.stationxinc.com/



Omicia

http://www.omicia.com/

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Opal: Unlocking individualized medicine

Advances in whole-genome sequencing technology are paving the way for genome analysis to become a routine part of healthcare delivery. Interpretation of genomes is the key factor limiting their utility for clinical applications.

Introducing Omicia Opal

Omicia Opal empowers researchers and clinicians to analyze genomes and prioritize disease-causing variants and genes.

Omicia Opal combines powerful, peer-reviewed analysis tools with proprietary disease gene sets into an interactive genome mining, filtering, prioritizing, and reporting environment.

Omicia Opal is cloud-based, scalable, and secure. Genome interpretation is now at your fingertips.

Stanford Center for Genomics & Personalized Medicine

http://scgpm.stanford.edu/

STANFORD SCHOOL OF MEDICINE

Stanford Center for Genomics and Personalized Medicine

GENOMICS PROMISES TO ADVANCE HEALTH

Our health depends upon both our genes and our environmental exposure. The current revolution in genomics makes it possible not only to determine our entire DNA sequence but also to begin to understand how our specific genome sequence can inform our health. In addition, our Center has recently demonstrated that it is possible to measure tens of thousands of components in blood to obtain a clear picture of our molecular picture during healthy and disease states. A combination of such sequence and molecular omics profiling is expected to be powerful in preventing, detecting, understanding, and treating complex diseases such as cancer and inherited diseases that are otherwise difficult to diagnose. *Learn more ...*

SCGPM FACILITATES TRANSLATION OF GENOMICS INTO PATIENT CENTERED MEDICINE

SCGPM Brochure

The Stanford Center for Genomics and Personalized Medicine (SCGPM) seeks to advance genomic technology so that someday both genetic and molecular profiling will become powerful and routine tools for predicting disease risk and monitoring and treating a wide range of pathologies. Towards this mission, the SCGPM serves to centralize and develop collaborative intellectual and technological resources that promote genomic research and analysis, predict drug response, educate physicians, and examine the ethics of personalized medicine. This includes large basic science projects such as ENCODE that decipher the human genome as well as clinical research projects such as the sequencing of cancer genomes and individuals with inherited diseases. Through these efforts, the Center aims to bring genomics to the clinic.

A MESSAGE FROM THE DIRECTOR

"Genomics is transforming both biological research and medicine. Stanford has long been a leader in this area and continues to develop new approaches to revolutionize the way medicine is practiced, so that disease can be rapidly diagnosed and the right treatment is applied at the right time."

Mike Snyder, PhD
Chair, Stanford Department of Genetics
Stanford W. Ascherman, MD, FACS, Professor of Genetics
Director, SCGPM

ANNOUNCEMENTS

2013 SGTP Symposium

4th Annual Symposium on Genomics and Personalized Medicine

View Announcement and Registration
Note: Registration deadline is March 29, 2013; open to Stanford community only

SCGPM MEMBER RESEARCH AND NEWS

SCGPM Members & Staff
» Executive Committee
» Affiliated Faculty
» Staff

News & Events
» 2013 SGTP Symposium Announcement & Registration
» Members in the News
» Stanford Seminars

Scientific Background
» General Information
» Genome & Genome Variation

Portrait of a Glitch

- Revere La Noue, MFA, Stanford, 2005
- What is this film about?
- What classes of glitches are mentioned?
- What do these glitches cause?
- Why did I show this film?

Genetic and Medical Web Sites

- NLM and NCBI
  - Entrez Gene
    - Protein
    - Biosystems
  - GeneReviews
  - OMIM
  - Genetics Home Reference
  - Genes and Diseases
  - Genetic Testing Registry
  - MedGen
  - Medline Plus

Impact of Genomics on Medicine
I. Diagnostics

- Genomics: Identifying all known human genes
- Functional Genomics: Functional analysis of genes
  - What tissues are they important
  - When in development are the genes used
  - How are they regulated
- Novel diagnostics
  - Linking genes to diseases and to traits
  - Predisposition to diseases
  - Expression of genes and disease
- Personal Genomics
  - Understanding link between genomics and environment
  - Increased vigilance and taking action to prevent disease
  - Improving health care

Impact of Genomics on Medicine
II. Therapeutics

- Novel Drug Development
  - Identifying novel drug targets
  - Validating drug targets
  - Predicting toxicity and adverse reactions
  - Improving clinical trials and testing
- Gene therapy
  - Replacing the gene rather than gene product
- Stem cells therapies
  - Replacing the entire cell to cure a disease
- Pharmacogenomics
  - Personalized medicine
  - Adjusting drug, amounts and delivery to suit patients
  - Maximize efficacy and minimize side effects
  - Identify genetics of adverse reactions
  - Identify patients who respond optimally

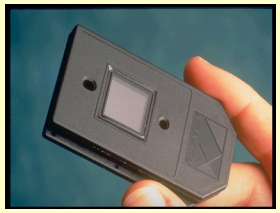
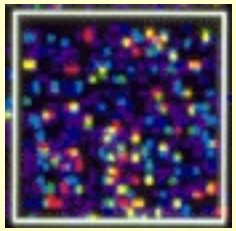
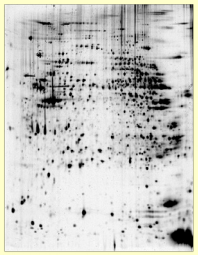
Impact of Genomics on Medicine
III. Ethical, Legal and Social Issues

- Personal Privacy
- Insurability
- Employability
- Discrimination
- Eugenics
- Cosmetic genetics
- Patentability of genes, proteins and other natural products

Leveraging Genomic Information

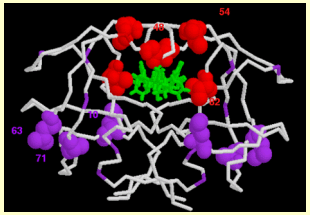
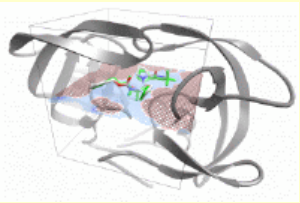
Novel Diagnostics

- Microchips – DNA and variations
- Microarrays Gene Expression - RNA
- Proteomics – Protein and modifications

Novel Therapeutics

- Drug Target Discovery
- Rational Drug Design
- Molecular Docking
- Gene Therapy
- Stem Cell Therapy


Understanding Metabolism

Understanding Disease

- Inherited Diseases –
- Genetics Home Reference
- Infectious Diseases
  - Pathogenic Bacteria
  - Viruses