Your Genes and Your Health

http://bio84.stanford.edu/

Diseases and Disease Databases

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Biochemistry & Medicine
Stanford University School of Medicine

Stanford at The Tech: Understanding Genetics

http://genetics.thetech.org/

Genomics as the Basis of Preventive Medicine

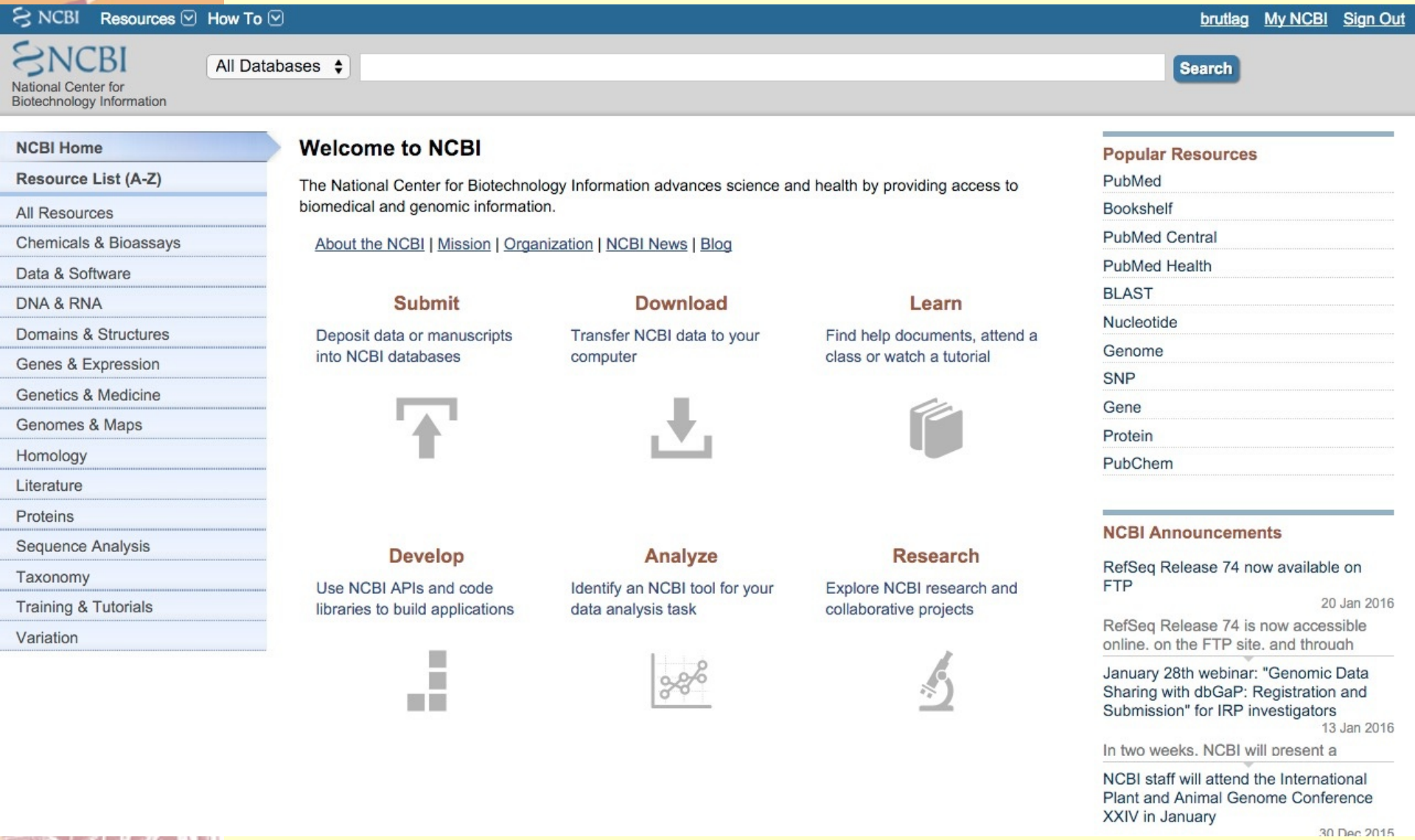
- If we know the gene that causes an inherited disease
- And we know the function of that gene
- Then we can understand the cause of the disease at the molecular level,
- This knowledge permits development of better diagnoses, treatments, drugs, therapies and other interventions to cure the disease.

Where would you go for information on inherited diseases?

- Google or Google Scholar?
- National Center for Biotechnology Information
- Genes and Disease
- Genetics Home Reference
- Medline Plus
- Gene Reviews
- Online Mendelian Inheritance in Man (OMIM)
- MedGen – human medical genetics
- PubMed Keywords or PubMed MeSh search?
- PubMed Central
- PubMed Health
- Clinical Trials Database

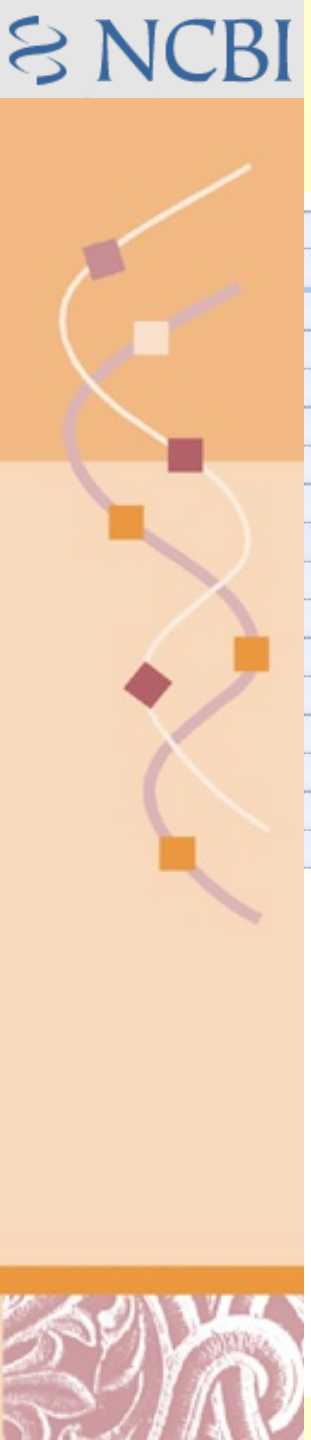
NCBI: National Center for Biotechnology Information

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



NCBI: Genetics and Medicine

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



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Genetics & Medicine

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Databases

Bookshelf
A collection of biomedical books that can be searched directly or from linked data in other NCBI databases. The collection includes biomedical textbooks, other scientific titles, genetic resources such as *GeneReviews*, and NCBI help manuals.

ClinVar
A resource to provide a public, tracked record of reported relationships between human variation and observed health status with supporting evidence. Related information in the NIH Genetic Testing Registry (GTR), MedGen, Gene, OMIM, PubMed and other sources is accessible through hyperlinks on the records.

ClincialTrials.gov
A registry and results database of publicly- and privately-supported clinical studies of human participants conducted around the world.

Database of Genotypes and Phenotypes (dbGaP)
An archive and distribution center for the description and results of studies which investigate the interaction of genotype and phenotype. These studies include genome-wide association (GWAS), medical resequencing, molecular diagnostic assays, as well as association between genotype and non-clinical traits.

Database of Major Histocompatibility Complex (dbMHC)
An open, publicly accessible platform where the HLA community can submit, edit, view, and exchange data related to the human major histocompatibility complex. It consists of an interactive Alignment Viewer for HLA and related genes, an MHC microsatellite database, a sequence interpretation site for Sequencing Based Typing (SBT), and a Primer/Probe database.

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.

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.

NCBI Pathogen Detection Project

A project involving the collection and analysis of bacterial pathogen genomic sequences originating from food, environmental and patient isolates. Currently, an automated pipeline clusters and identifies sequences supplied primarily by public health laboratories to assist in the investigation of foodborne disease outbreaks and discover potential sources of food contamination.

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-.

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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

NCBI

Genes and Disease

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

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Huntington Disease

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

NCBI

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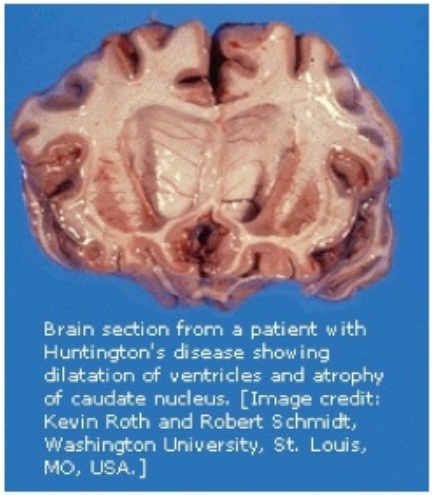
Bookshelf ID: NBK22226

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.

Related diseases

See other Diseases of the Nervous System

Genes and Disease [Internet].
National Center for Biotechnology Information (US).
Bethesda (MD): National Center for Biotechnology Information (US); 1998-.
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Gene sequence

Genome view see gene locations

Entrez Gene collection of gene-related information

BLink related sequences in different organisms

The literature

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Books online books section

OMIM catalog of human genes and disorders

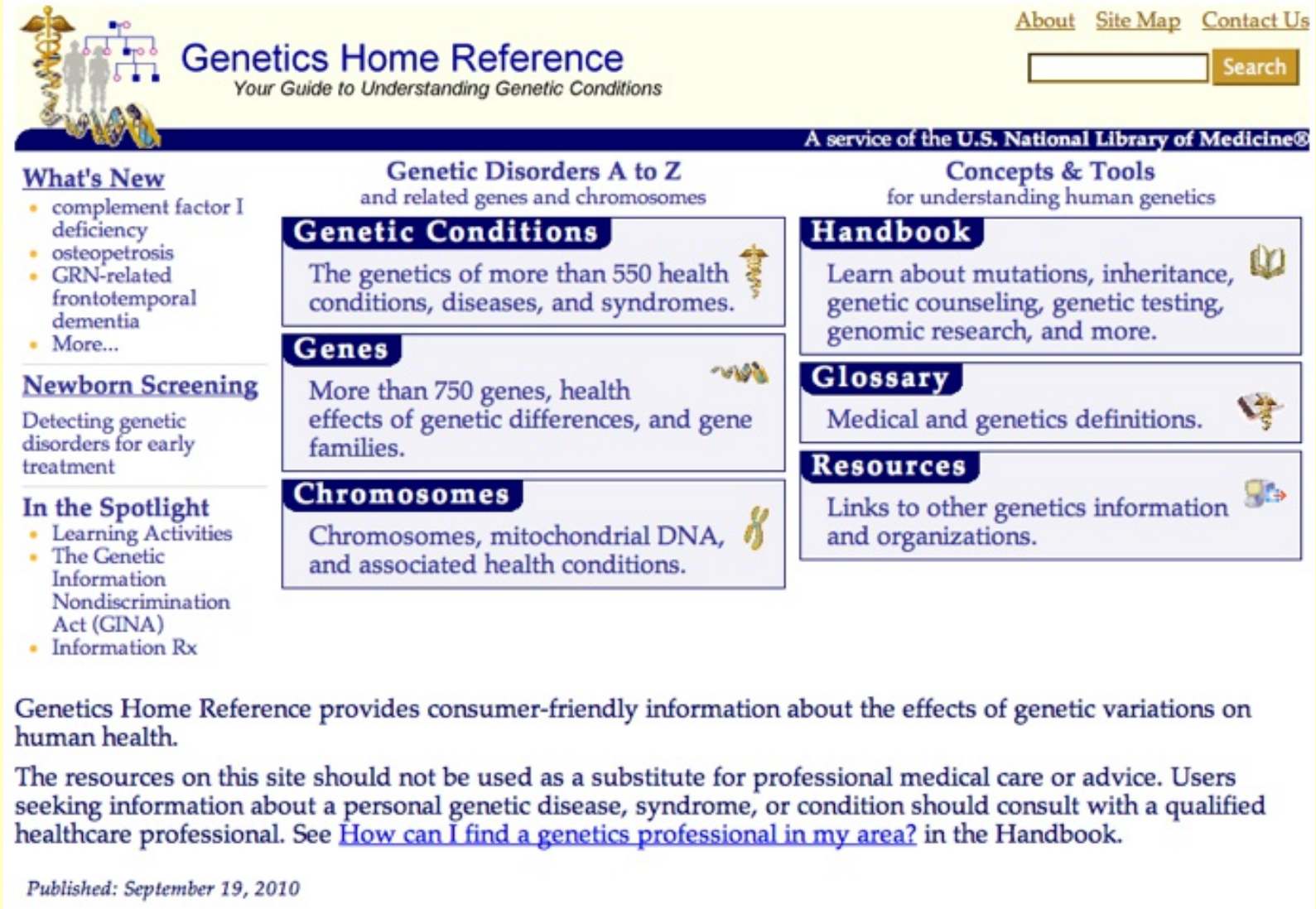
GeneReviews a medical genetics resource

Related information

Gene

Genetics Home Reference

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



Huntington Disease in Genetics Home Reference

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

Genetic Conditions >

Huntington disease

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MedlinePlus
Health information

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

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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.

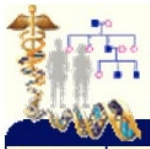
A less common, early-onset form of Huntington disease begins in childhood or adolescence. It also involves movement problems and mental and emotional changes. Additional signs of the early-onset form include slow movements, clumsiness, frequent falling, rigidity, slurred speech, and drooling. School 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.

Huntingtin Gene HTT in Genetics Home Reference

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



Genetics Home Reference

Your Guide to Understanding Genetic Conditions

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Genes > ***HTT***

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Tools for researchers

External link disclaimer

On this page: Name | Normal function | Gene families | Genetic changes | Gene location | Additional information | Other names | About genes | Glossary definitions

Reviewed October 2008

What is the official name of the *HTT* gene?

The official name of this gene is "huntingtin."

HTT is the gene's official symbol. The *HTT* gene is also known by other names, listed below.

Read more about gene names and symbols on the About page.

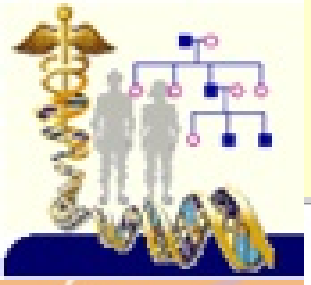
What is the normal function of the *HTT* gene?

The *HTT* gene provides instructions for making a protein called huntingtin. Although the exact function of this protein is unknown, it appears to play an important role in nerve cells (neurons) in the brain and is essential for normal development before birth. Huntingtin is found in many of the body's tissues, with the highest levels of activity in the brain. Within cells, this protein may be involved in chemical signaling, transporting materials, attaching (binding) to proteins and other structures, and protecting the cell from self-destruction (apoptosis).

One region of the *HTT* gene contains a particular DNA segment known as a CAG trinucleotide repeat. This segment is made up of a series of three DNA building blocks (cytosine, adenine, and guanine) that appear multiple times in a row. Normally, the CAG segment is repeated 10 to 35 times within the gene.

Newborn Screening in Genetics Home Reference

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



Newborn Screening

Quick Reference:

- Description of disorders detected through newborn screening

External link disclaimer

Newborn screening is the practice of testing all babies for certain disorders and conditions that can hinder their normal development. Babies with these conditions appear healthy at birth but can develop serious medical problems later in infancy or childhood. Early detection and treatment can help prevent intellectual and physical disabilities and life-threatening illnesses.

Newborn screening usually begins with a blood test 24 to 48 hours after the baby is born. The test is performed by pricking the baby's heel to collect a few drops of blood. The blood is placed on a special piece of paper and sent to a laboratory for analysis. Parents can ask for a copy of the test results, which are sent to the baby's doctor or clinic.

Sometimes a repeat blood test is required, particularly if the first test was done before the baby was 24 hours old. If the results of the test are abnormal, additional testing is required to confirm the result. Parents are notified within a few days of the first test if retesting is necessary. The blood test should be repeated as soon as possible.

Newborn screening varies from state to state. All states must screen for at least 21 disorders by law, and some states test for 30 or more. Parents can ask their doctor about expanded (supplemental) screening if they live in an area that screens for a limited number of disorders.

To encourage uniform and comprehensive newborn screening throughout the United States, the Health Resources and Services Administration (HRSA) issued a report that recommends screening for 29 specific conditions. The recommendations include a test for hearing loss in newborns. The hearing test uses a soft earphone or other instrument that is placed in the baby's ear.

Please use the links below to learn more about newborn screening.

- Description of disorders detected through newborn screening (Genetics Home Reference)
- Newborn screening resources (U.S. National Library of Medicine)
- Frequently Asked Questions about Newborn Bloodspot Screening (National Newborn

Handbook

Help Me Understand Genetics

Help Me Understand Genetics presents basic information about genetics in clear language and provides links to online resources.

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2. What are proteins and what do they do?
3. How many chromosomes do people have?
4. What is a gene mutation and how do mutations occur?
5. What kinds of gene mutations are possible?

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Articles and images for diseases, symptoms, tests, treatments

Medical Dictionary from Merriam-Webster

Is there more to see in your family tree? Glaucoma runs in families.

Learn more during National Glaucoma Awareness Month.

1 2 3 4 ▶

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Huntington Disease in Medline Plus

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Huntington disease

Huntington disease is a disorder in which nerve cells in certain parts of the brain waste away, or degenerate. The disease is passed down through families.

Causes

Huntington disease is caused by a genetic defect on chromosome 4. The defect causes a part of DNA, called a CAG repeat, to occur many more times than it is supposed to. Normally, this section of DNA is repeated 10 to 28 times. But in persons with Huntington disease, it is repeated 36 to 120 times.

As the gene is passed down through families, the number of repeats tend to get larger. The larger the number of repeats, the higher your chance of developing symptoms at an earlier age. Therefore, as the disease is passed along in families, symptoms develop at younger and younger ages.

There are two forms of Huntington disease.

- Adult-onset Huntington disease is the most common. Persons with this form usually develop symptoms in their mid 30s and 40s.
- Early-onset Huntington disease affects a small number of cases and begins in childhood or the teens.

If one of your parents has Huntington disease, you have a 50% chance of getting the gene. If you get the gene from your parents, you can also pass it on to your children, who will also have a 50% change of getting the gene. If you do not get the gene from your parents, you cannot pass the gene on to your children.

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Huntington's Disease

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Movement – unpredictable or jerky
Muscle atrophy

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

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- Evaluating Health Information
- Director's Comments Podcast
- NIHSeniorHealth Videos

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-.
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Summary

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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
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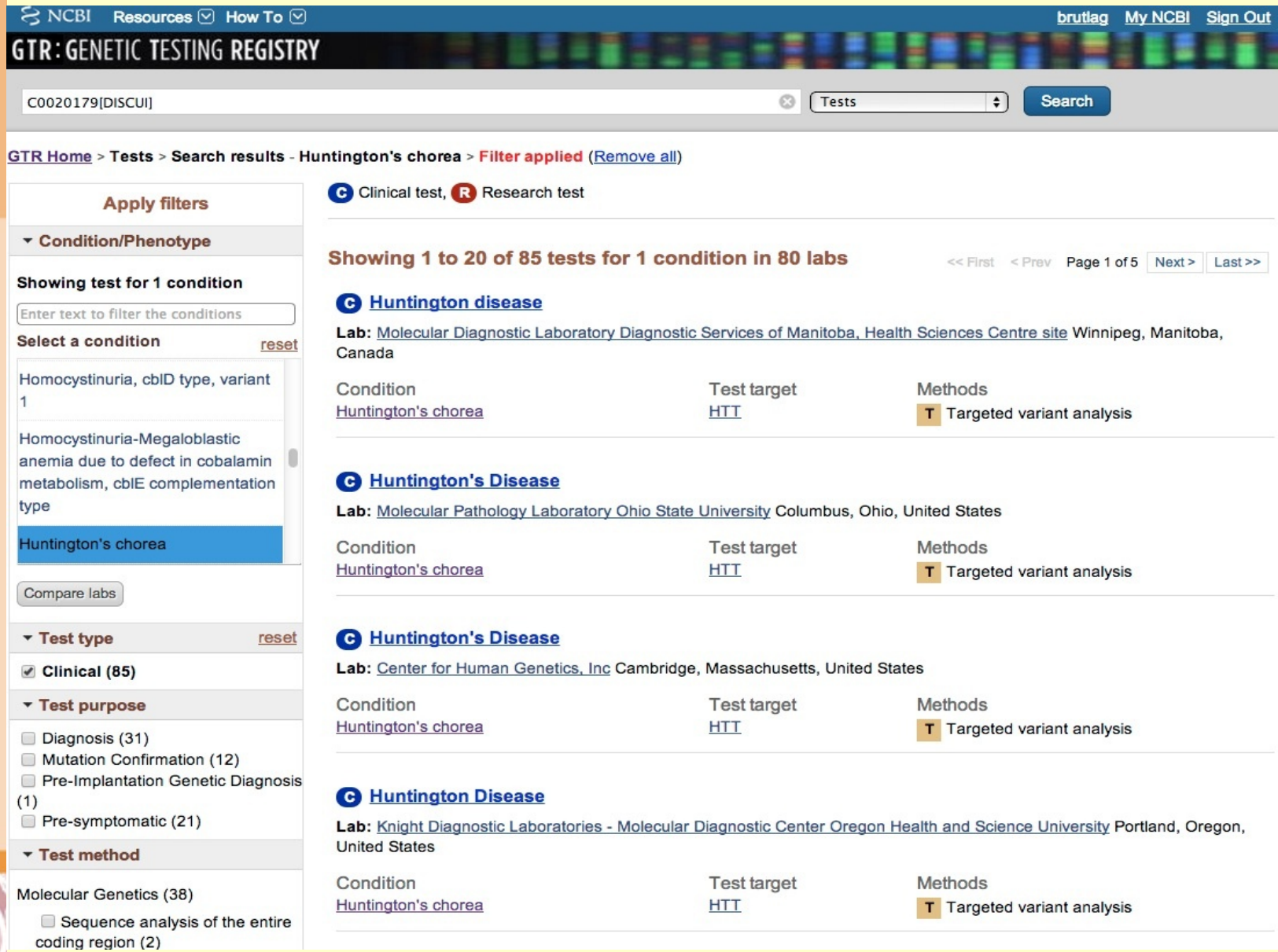
Genetic Testing Registry for Huntington

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



Huntington Disease Gene

http://www.ncbi.nlm.nih.gov/gene/3064

NCBI

Entrez Gene
Genes and mapped phenotypes

Search: Gene | Limits | Advanced search | Help

Search | Clear

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HTT huntingtin [*Homo sapiens*]

Gene ID: 3064, updated on 3-Jan-2011

Summary

Official Symbol	HTT provided by HGNC
Official Full Name	huntingtin provided by HGNC
Primary source	HGNC:4851
See related	Ensembl:ENSG00000197386; HPRD:00883; MIM:613004
Gene type	protein coding
RefSeq status	REVIEWED
Organism	Homo sapiens
Lineage	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as	HD; IT15; HTT
Summary	Huntingtin is a disease gene linked to Huntington's disease, a neurodegenerative disorder characterized by loss of striatal neurons. This is thought to be caused by an expanded, unstable trinucleotide repeat in the huntingtin gene, which translates as a polyglutamine repeat in the protein product. A fairly broad range in the number of trinucleotide repeats has been identified in normal controls, and repeat numbers in excess of 40 have been described as pathological. The huntingtin locus is large, spanning 180 kb and consisting of 67 exons. The huntingtin gene is widely expressed and is required for normal development. It is expressed as 2 alternatively polyadenylated forms displaying different relative abundance in various fetal and adult tissues. The larger transcript is approximately 13.7 kb and is expressed predominantly in adult and fetal brain whereas the smaller transcript of approximately 10.3 kb is more widely expressed. The genetic defect leading to Huntington's disease may not necessarily eliminate transcription, but may confer a new property on the mRNA or alter the function of the protein. One candidate is the huntingtin-associated protein-1, highly expressed in brain, which has increased affinity for huntingtin protein with expanded polyglutamine repeats. This gene contains an upstream open reading frame in the 5' UTR that inhibits expression of the huntingtin gene product through translational repression. [provided by RefSeq]

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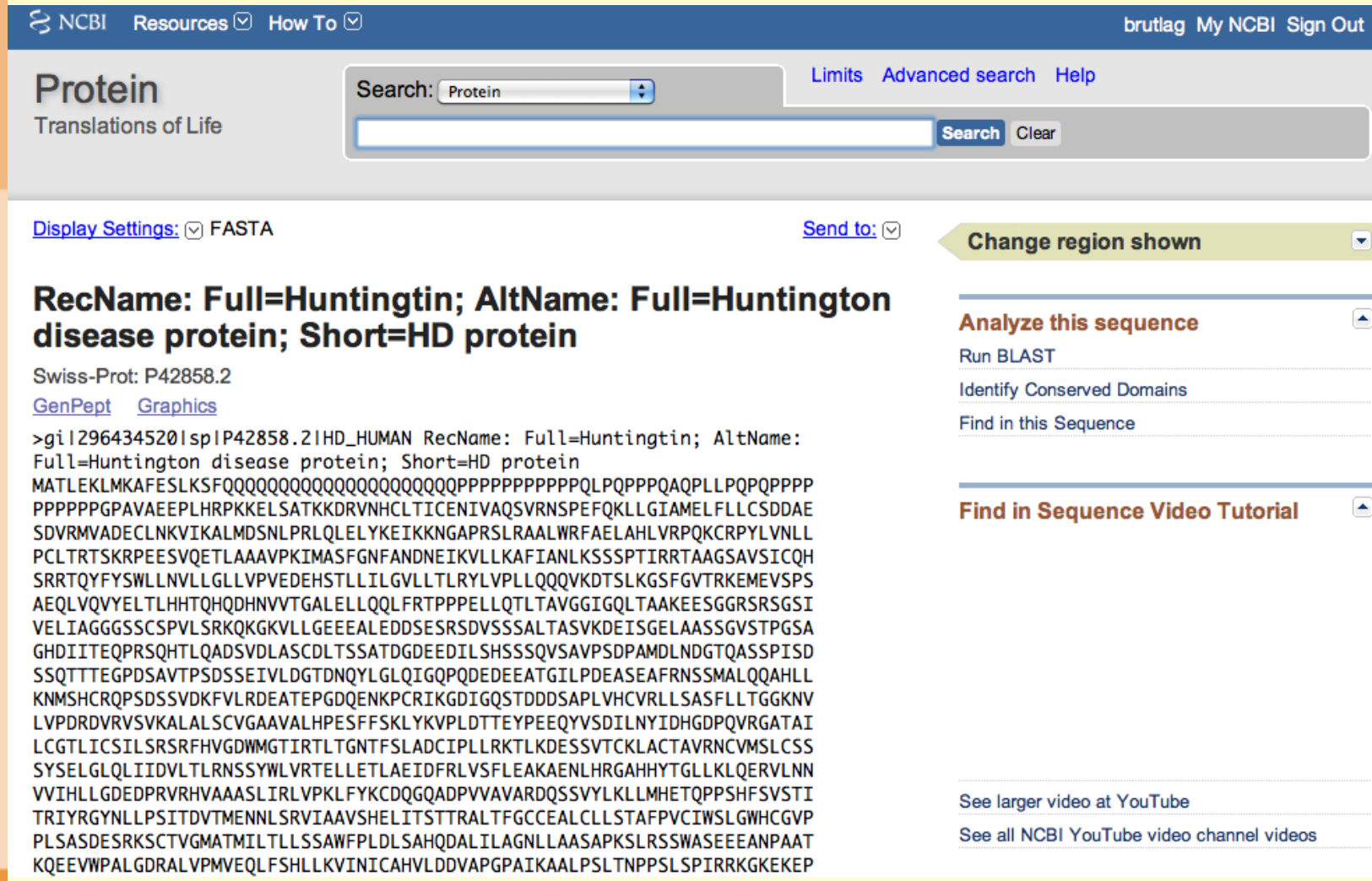
- Map Viewer
- Nucleotide

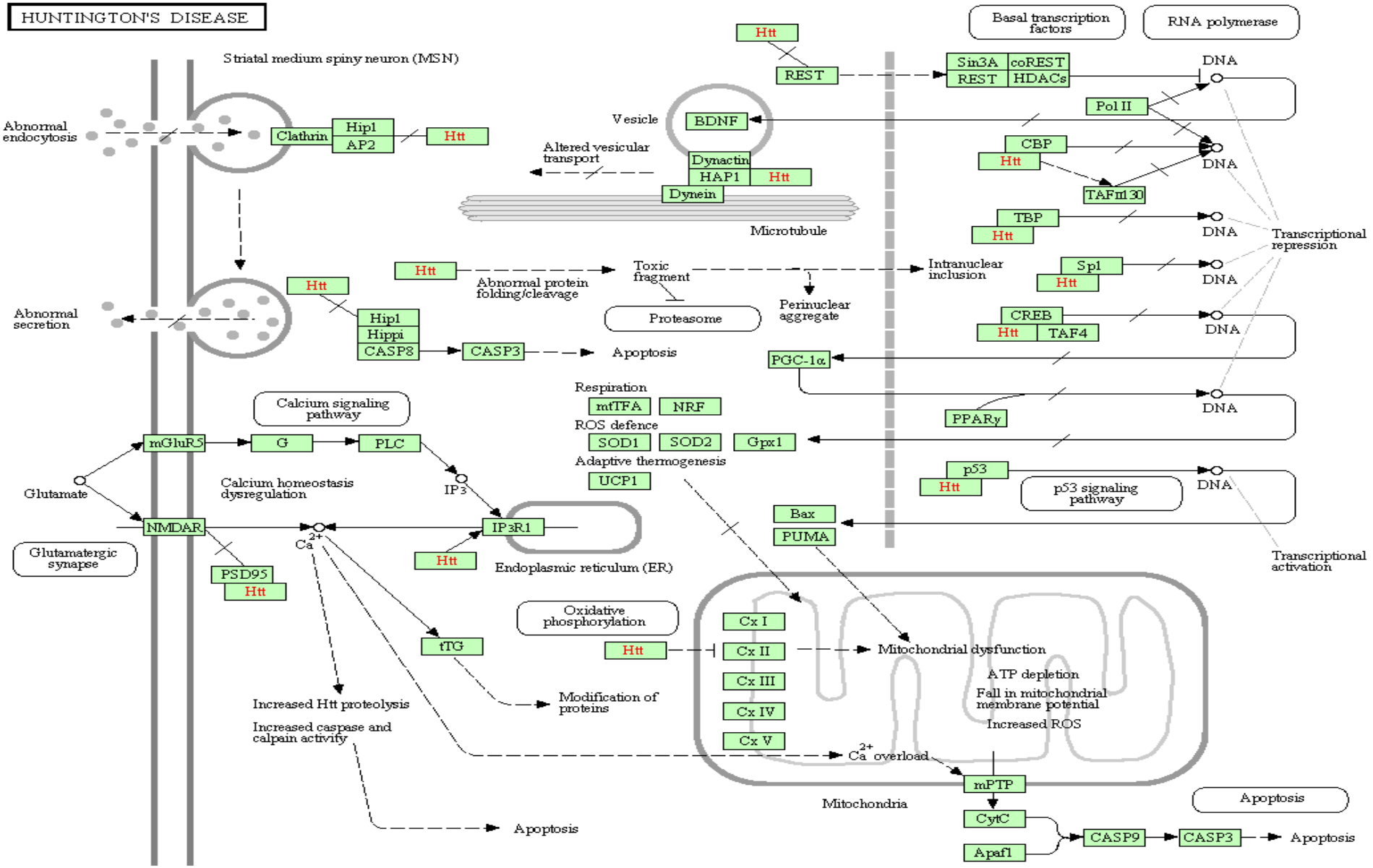
Huntington Disease Protein Sequence

http://www.ncbi.nlm.nih.gov/protein/296434520?report=fasta

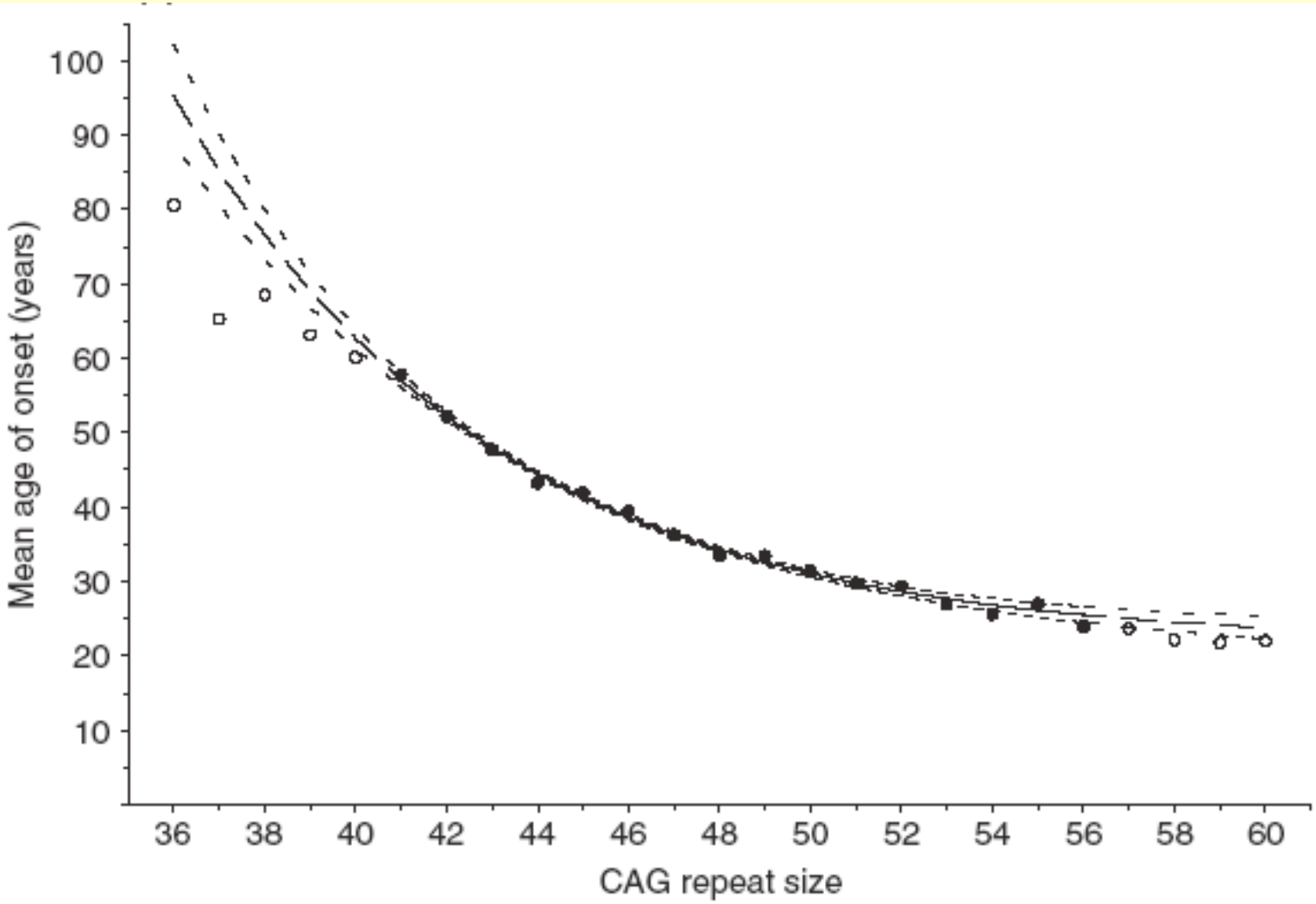


Huntington Disease Biosystem

http://www.ncbi.nlm.nih.gov/biosystems/83100

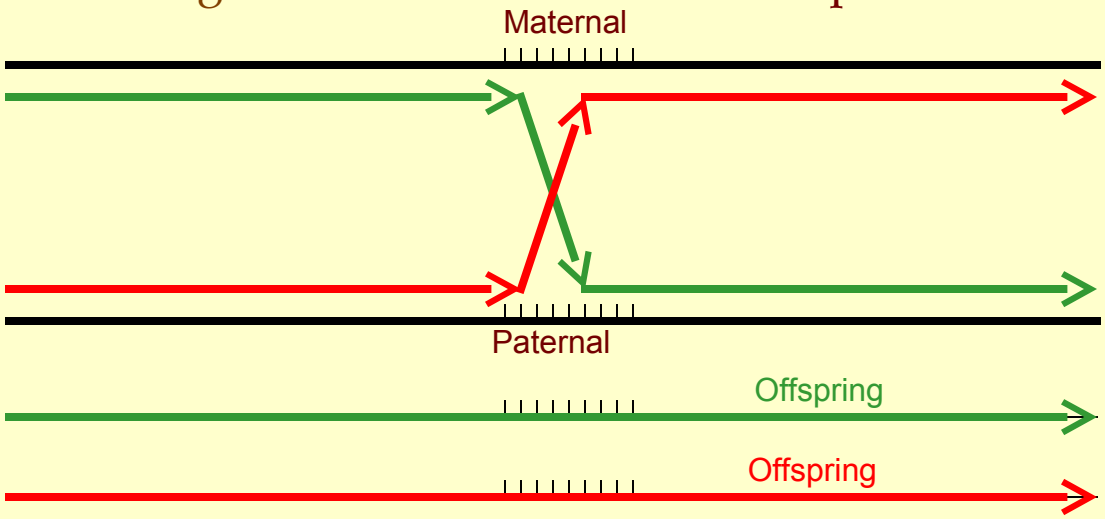


Age of Onset and Repeat Length

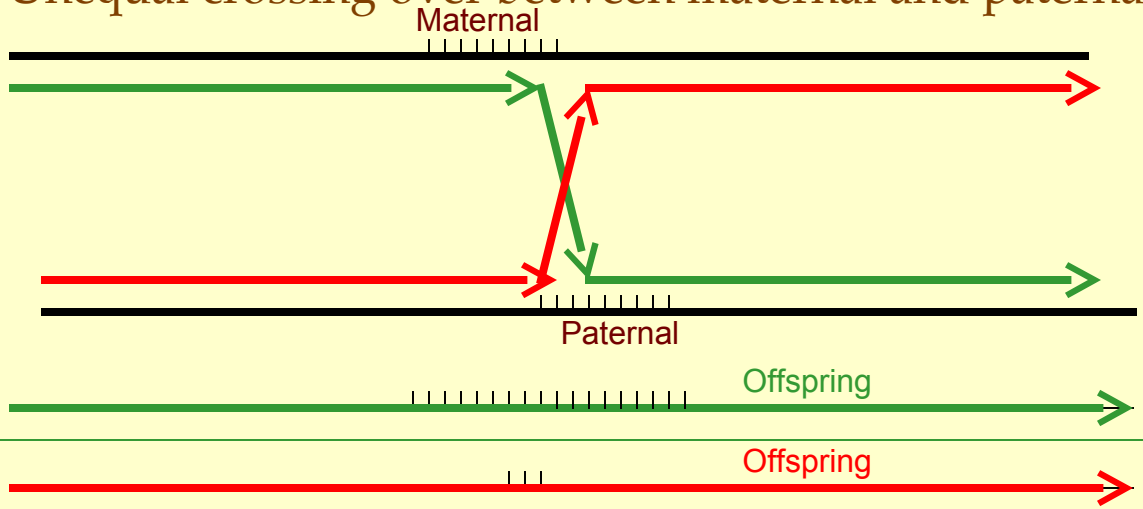


Huntington Disease can Arise from Unequal Crossing Over During Meiosis

- Crossing over between maternal and paternal chromosomes



- Unequal crossing over between maternal and paternal chromosomes



Trinucleotide Repeat Disorders

https://en.wikipedia.org/wiki/Trinucleotide_repeat_disorder

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Trinucleotide repeat disorder

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Trinucleotide repeat disorders (also known as **trinucleotide repeat expansion disorders**, **triplet repeat expansion disorders** or **codon reiteration disorders**) are a set of genetic disorders caused by trinucleotide repeat expansion, a kind of mutation where trinucleotide repeats in certain genes exceed the normal, stable threshold, which differs per gene. The mutation is a subset of unstable microsatellite repeats that occur throughout all genomic sequences. If the repeat is present in a healthy gene, a dynamic mutation may increase the repeat count and result in a defective gene.

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Trinucleotide Repeat Disorders

https://en.wikipedia.org/wiki/Trinucleotide_repeat_disorder

Polyglutamine (PolyQ) Diseases [edit]

Type	Gene	Normal PolyQ repeats	Pathogenic PolyQ repeats
DRPLA (Dentatorubropallidoluysian atrophy)	ATN1 or DRPLA	6 - 35	49 - 88
HD (Huntington's disease)	HTT (Huntingtin)	6 - 35	36 - 250
SBMA (Spinal and bulbar muscular atrophy)	AR	9 - 36	38 - 62
SCA1 (Spinocerebellar ataxia Type 1)	ATXN1	6 - 35	49 - 88
SCA2 (Spinocerebellar ataxia Type 2)	ATXN2	14 - 32	33 - 77
SCA3 (Spinocerebellar ataxia Type 3 or Machado-Joseph disease)	ATXN3	12 - 40	55 - 86
SCA6 (Spinocerebellar ataxia Type 6)	CACNA1A	4 - 18	21 - 30
SCA7 (Spinocerebellar ataxia Type 7)	ATXN7	7 - 17	38 - 120
SCA17 (Spinocerebellar ataxia Type 17)	TBP	25 - 42	47 - 63

Trinucleotide Repeat Disorders

https://en.wikipedia.org/wiki/Trinucleotide_repeat_disorder

Non-Polyglutamine Diseases [edit]

Type	Gene	Codon	Normal/wild type	Pathogenic
FRAXA (Fragile X syndrome)	*FMR1*, on the X-chromosome	**CGG**	6 - 53	230+
FXTAS (Fragile X-associated tremor/ataxia syndrome)	*FMR1*, on the X-chromosome	**CGG**	6 - 53	55-200
FRAXE (Fragile XE mental retardation)	AFF2 or FMR2, on the X-chromosome	**CCG**	6 - 35	200+
FRDA (Friedreich's ataxia)	FXN or X25, (frataxin—reduced expression)	**GAA**	7 - 34	100+
DM (Myotonic dystrophy)	DMPK	**CTG**	5 - 37	50+
SCA8 (Spinocerebellar ataxia Type 8)	OSCA or SCA8	**CTG**	16 - 37	110 - 250
SCA12 (Spinocerebellar ataxia Type 12)	PPP2R2B or SCA12	**nnn** On 5' end	7 - 28	66 - 78

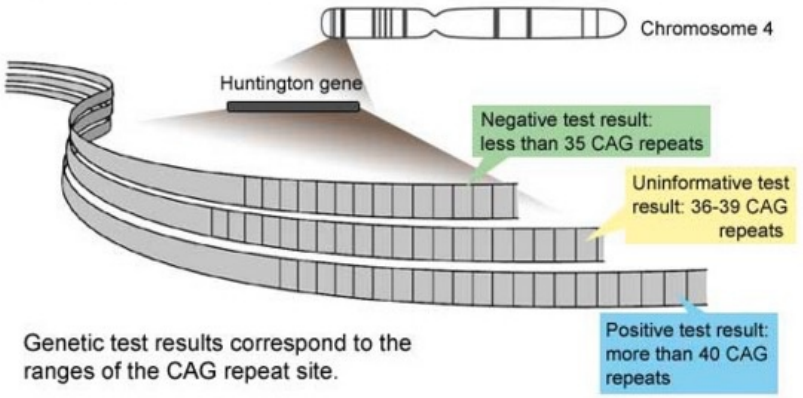
Huntington Outreach Project at Stanford

http://web.stanford.edu/group/hopes/cgi-bin/hopes_test/

HUNTINGTON'S OUTREACH PROJECT FOR EDUCATION, AT STANFORD

Home | About | For Kids | Science of HD | Managing HD | Global HD | Other Resources | Search

Figure S-3: CAG Repeat Counts on the Huntington gene



Genetic test results correspond to the ranges of the CAG repeat site.

The HOPES Brain Tutorial
Cerebral Cortex

Genetic Testing

TRiC and Huntington Protein Aggregation

Trojan Therapy
The Science Behind Trojan Therapy

2016

Huntington Outreach Project at Stanford

http://web.stanford.edu/group/hopes/cgi-bin/hopes_test/trinucleotide-repeat-disorders/

HUNTINGTON'S OUTREACH PROJECT FOR EDUCATION, AT STANFORD

Other neurodegenerative disorders

Trinucleotide Repeat Disorders

By Stephanie Liou | 26 Jun, 2010 | Other neurodegenerative disorders

When the cause of a disease can be traced to having too many copies of a certain nucleotide triplet in the DNA, the disease is said to be a trinucleotide repeat disorder. Today, there are 14 documented trinucleotide repeat disorders that affect human beings**. Huntington's Disease is part of this group.

Some of these 14 trinucleotide repeat disorders are more alike than others. While the symptoms and the affected body parts vary by disease, scientists consider two illnesses to be similar if they share the same repeated codon as their cause. Six of the 14 trinucleotide repeat disorders have little or no apparent similarity to each other, or to the 8 remaining diseases. These 6 are described in brief at the end of this section. The 8 remaining disorders, one of which is Huntington's Disease, all share the same repeated codon as their cause: CAG. Since CAG codes for an amino acid called glutamine, these 8 trinucleotide

Clinical Trials

https://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.

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

ClinicalTrials.gov currently lists 182,594 studies with locations in all 50 states and in 187 countries.

Text Size

Search for Studies

Example: "Heart attack" AND "Los Angeles"

Huntington Disease | 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

- Non-U.S. Only (52%)
- U.S. Only (42%)
- Both U.S. and Non-U.S. (6%)

Total N = 34,362 studies
Data as of January 20, 2015

- 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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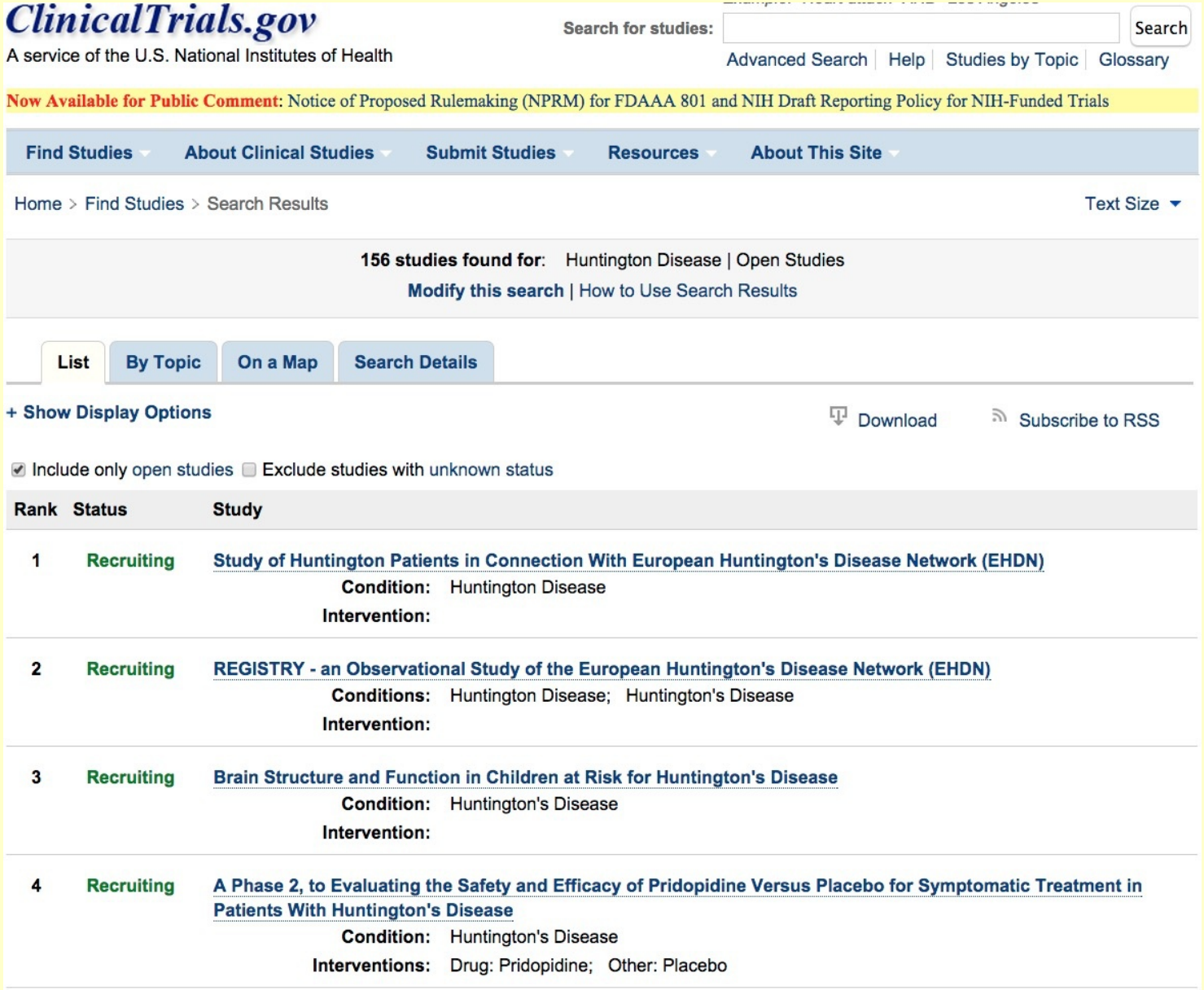
Huntington Disease Clinical Trials

https://clinicaltrials.gov/ct2/results?term=Huntington+Disease&recr=Open/



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

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

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Home > Find Studies > Search Results | Text Size

156 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

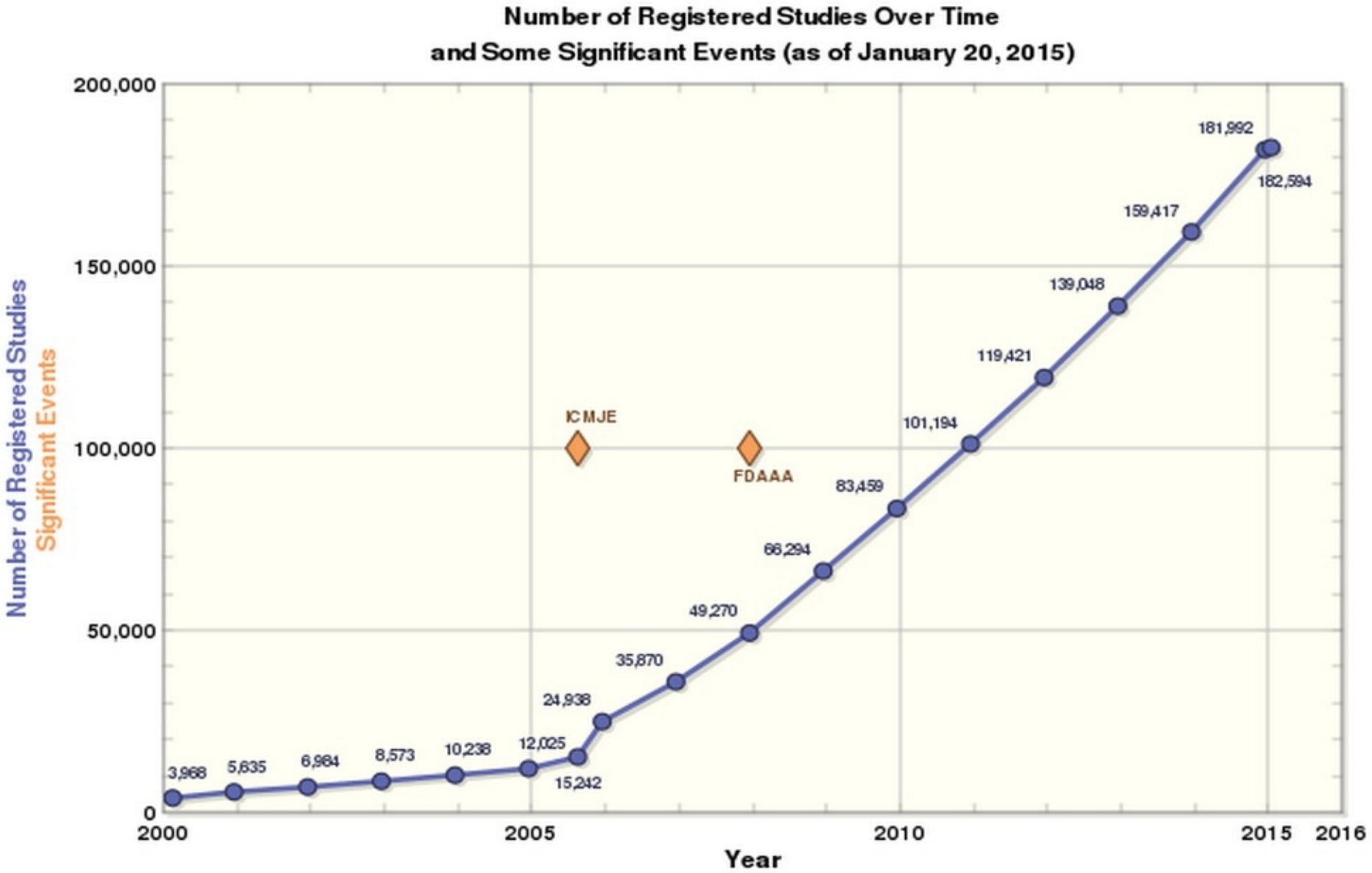
☑ Include only open studies ☐ Exclude studies with unknown status

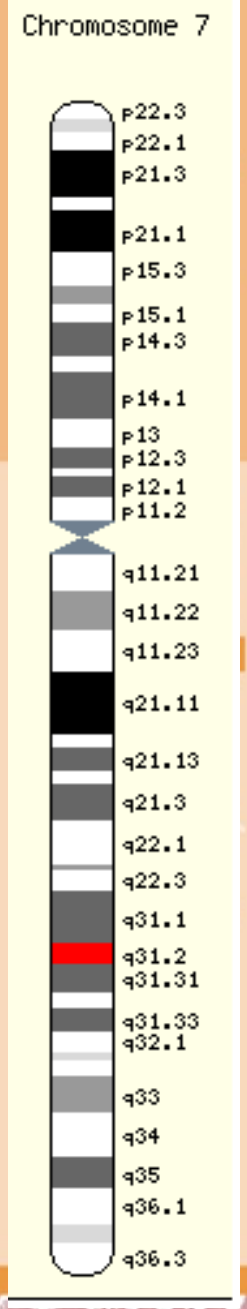
Rank	Status	Study
1	Recruiting	Study of Huntington Patients in Connection With European Huntington's Disease Network (EHDN) **Condition:** Huntington Disease **Intervention:**
2	Recruiting	REGISTRY - an Observational Study of the European Huntington's Disease Network (EHDN) **Conditions:** Huntington 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

ClinicalTrials

Registered Studies in Clinical Trials

https://clinicaltrials.gov/ct2/resources/trends

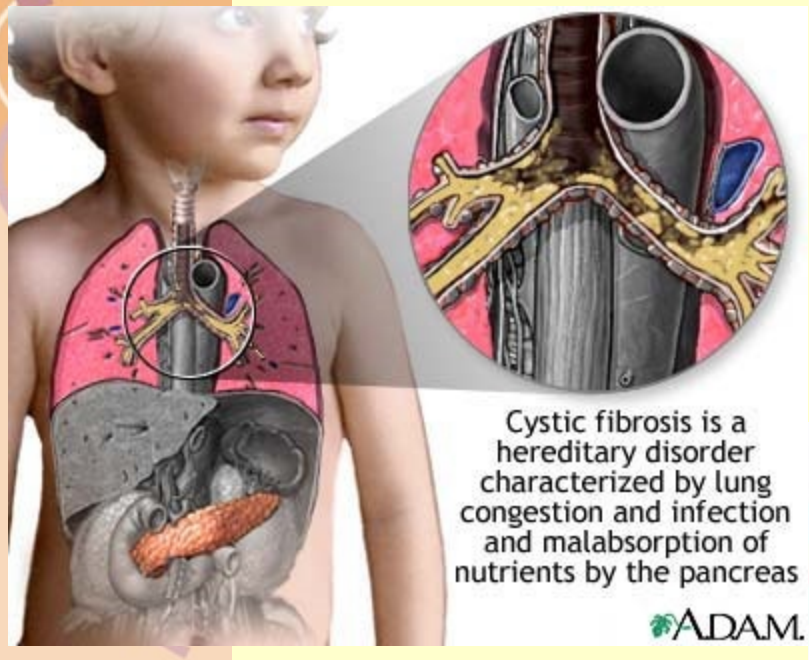


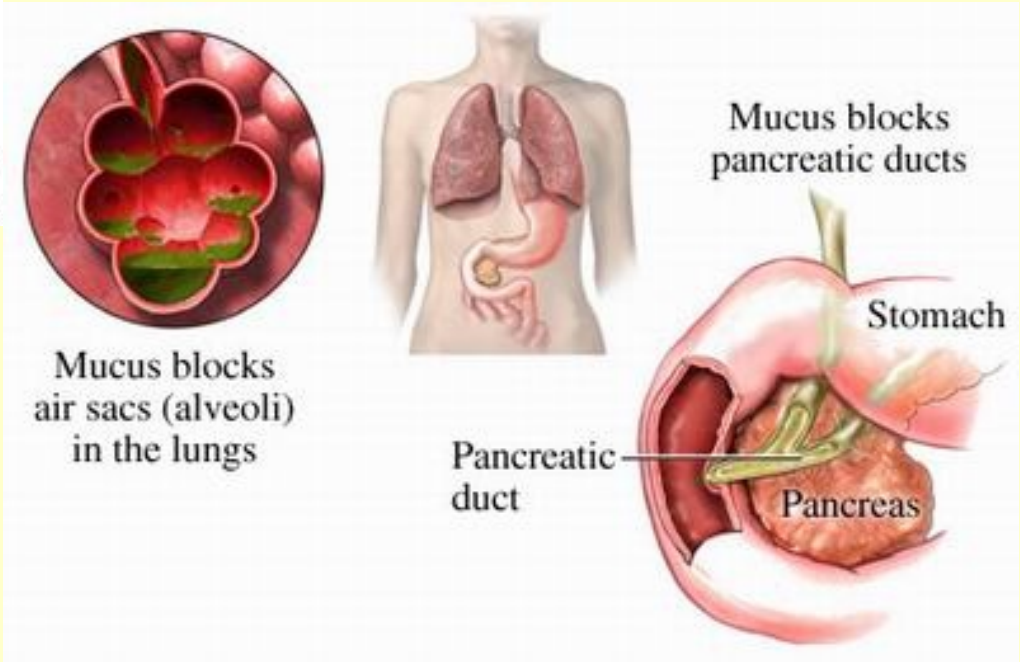


Cystic Fibrosis

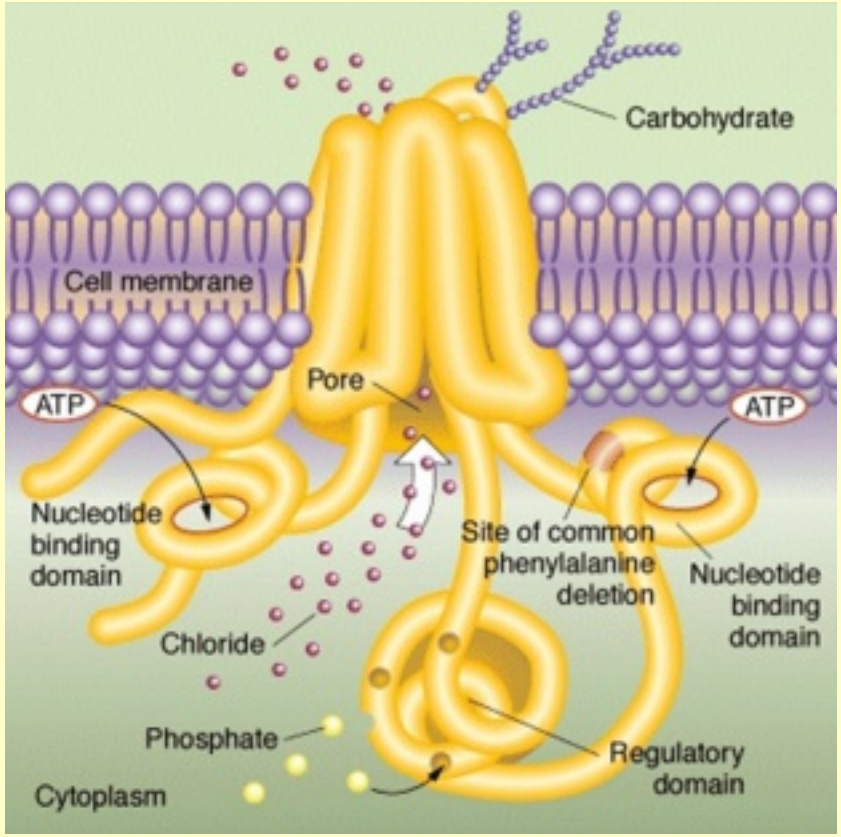
- Autosomal (chromosome 7q31.2) recessive.
- Inhibits many bodily secretions
  - Pancreatic digestive enzymes
  - Sweat glands
  - Lung mucosa in alveoli and bronchi
  - Infertility in males (>97%)
  - Cirrhosis of the liver
  - Hepatic steatosis
- Caused by mutations in the CFTR gene that encodes a chloride ion channel that pumps chloride ion and water out of cells.

Cystic Fibrosis





Cystic Fibrosis Transmembrane Conductance Regulator



CFTR is a chloride/water ion channel or pore

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

Genetic and Medical Web Sites

- National Library of Medicine and the
- National Center for Biotechnology Information
  - Genes and Diseases
  - Genetics Home Reference
  - Medline Plus
  - GeneReviews
  - Genetic Testing Registry
  - Clinical Trials

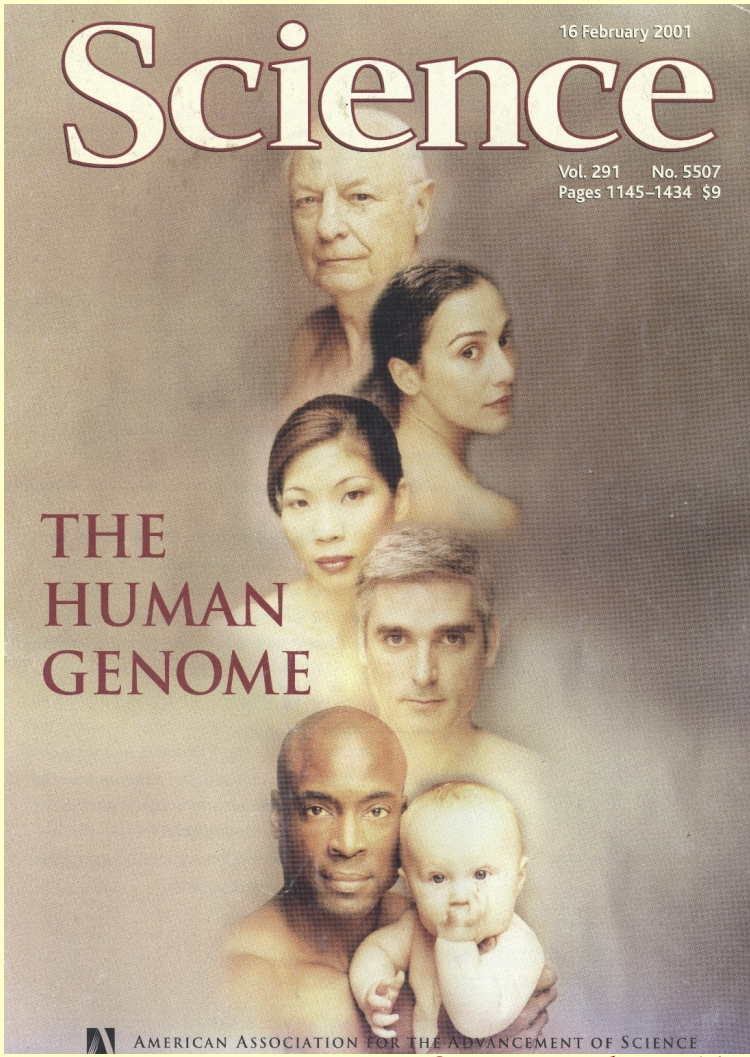
Where would you go for information on inherited diseases?

- Google or Google Scholar?
- National Center for Biotechnology Information
- Genes and Disease
- Genetics Home Reference
- Medline Plus
- Gene Reviews
- Online Mendelian Inheritance in Man (OMIM)
- MedGen – human medical genetics
- PubMed Keywords or PubMed MeSh search?
- PubMed Central
- PubMed Health
- Clinical Trials Database

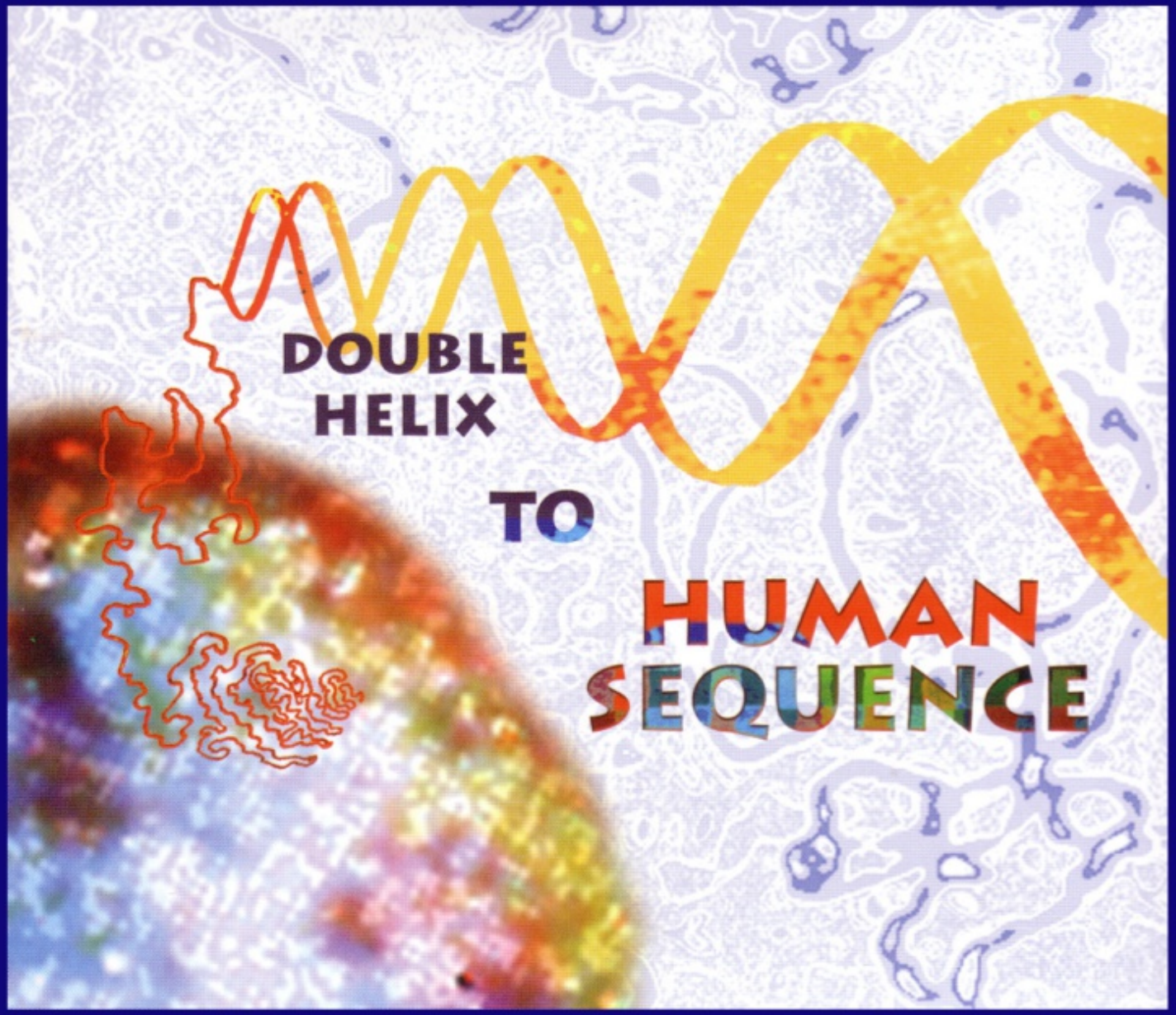
Human Genome First draft February 2001

Public Effort

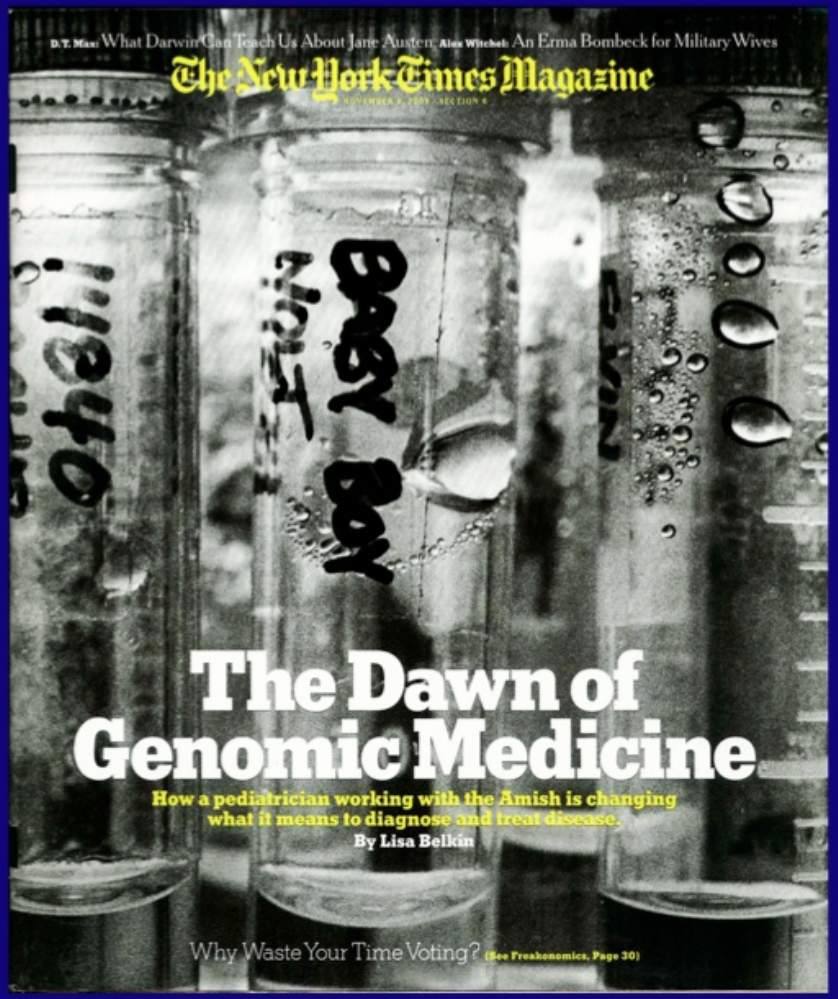
Private Effort (Celera)



April, 2003 Completion



Genomic Medicine



Genomic Medicine

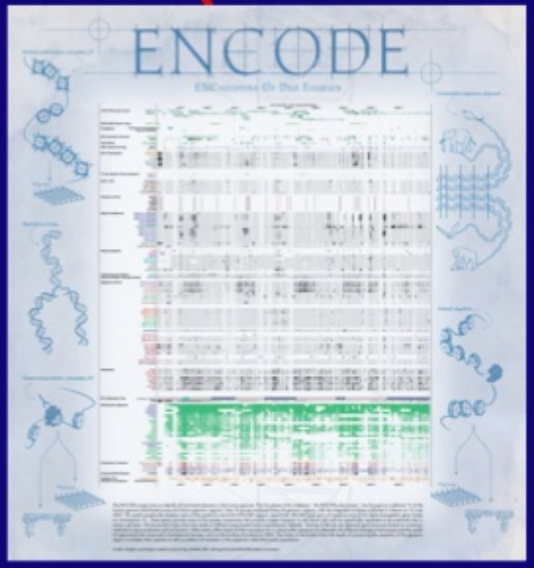
Healthcare tailored to the individual based on genomic information

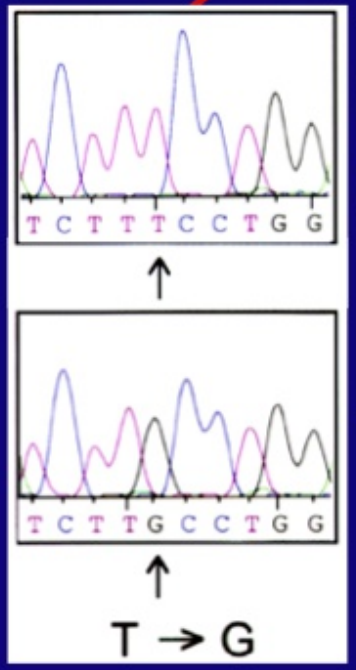
The Pathway to Genomic Medicine

Interpreting the Human Genome Sequence

Implicating Genetic Variants with Human Disease

HGP





Realization of Genomic Medicine

National Human Genome Research Institute

http://www.genome.gov/

NIH National Human Genome Research Institute

Advancing human health through genomics research

SEARCH GENOME.GOV

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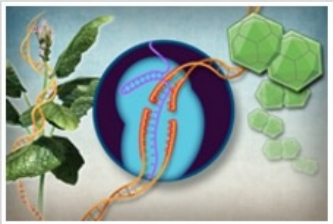
Highlights

NIH genome sequencing program targets the genomic bases of common, rare disease

The National Institutes of Health will fund a set of genome sequencing and analysis centers whose research will focus on understanding the genomic bases of common and rare human diseases. On January 14, the National Human Genome Research Institute (NHGRI), part of NIH, launched the Centers for Common Disease Genomics (CCDG), which will use genome sequencing to explore the genomic contributions to common diseases. **Read more**

Genome Advance of the Month

Gene-editing technology harnessed to protect plants from viruses



Scientists are using an exciting gene editing tool called CRISPR/Cas9 to protect plants from harmful DNA viruses. The CRISPR/Cas9 system has previously been adapted for use in many organisms, and this latest iteration develops gene-editing for use in plants. The November *Genome Advance of the Month* describes how these scientists inserted the code for an ancient bacterial immune system into a plant's genome to successfully strengthen the plant's protection against viruses. **Read more**

The Genomics Landscape

Future of ENCODE: Looking Deeper into Genome Function

For You

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

GenomeTV

Family Health History Day - Dr. Eric Green

Newsroom

Scientists create world's largest catalog of human genomic variation
September 30, 2015

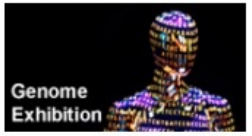
Grants to help identify variants in the genome's regulatory regions that affect disease risk
September 21, 2015

Undiagnosed Diseases Network launches online application portal
September 16, 2015

NIH grants seek best ways to combine genomic information and EHRs
September 1, 2015

All Current News Releases

Quick Links



Genome: Unlocking Life's Code

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

National Human Genome Research Institute

Advancing human health through genomics research

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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 Education Kit: Understanding the Human Genome Project
- Online Genetics Education Resources
- Smithsonian NHGRI Genome Exhibition
- Talking Glossary

Genome: Unlocking Life's Code

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.

Ongoing Programs

- **Exhibition Website**: ***Genome: Unlocking Life's Code*** [unlockinglifescode.org]
- **Traveling Exhibit** [unlockinglifescode.org]
- **Past Public Education Programs and Events**

Genome: Unlocking Life's Code

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

Ongoing Programs

- **Exhibition Website**: ***Genome: Unlocking Life's Code*** [unlockinglifescode.org]
- **Traveling Exhibit** [unlockinglifescode.org]
- **Past Public Education Programs and Events**

Information to Enhance Your Visit

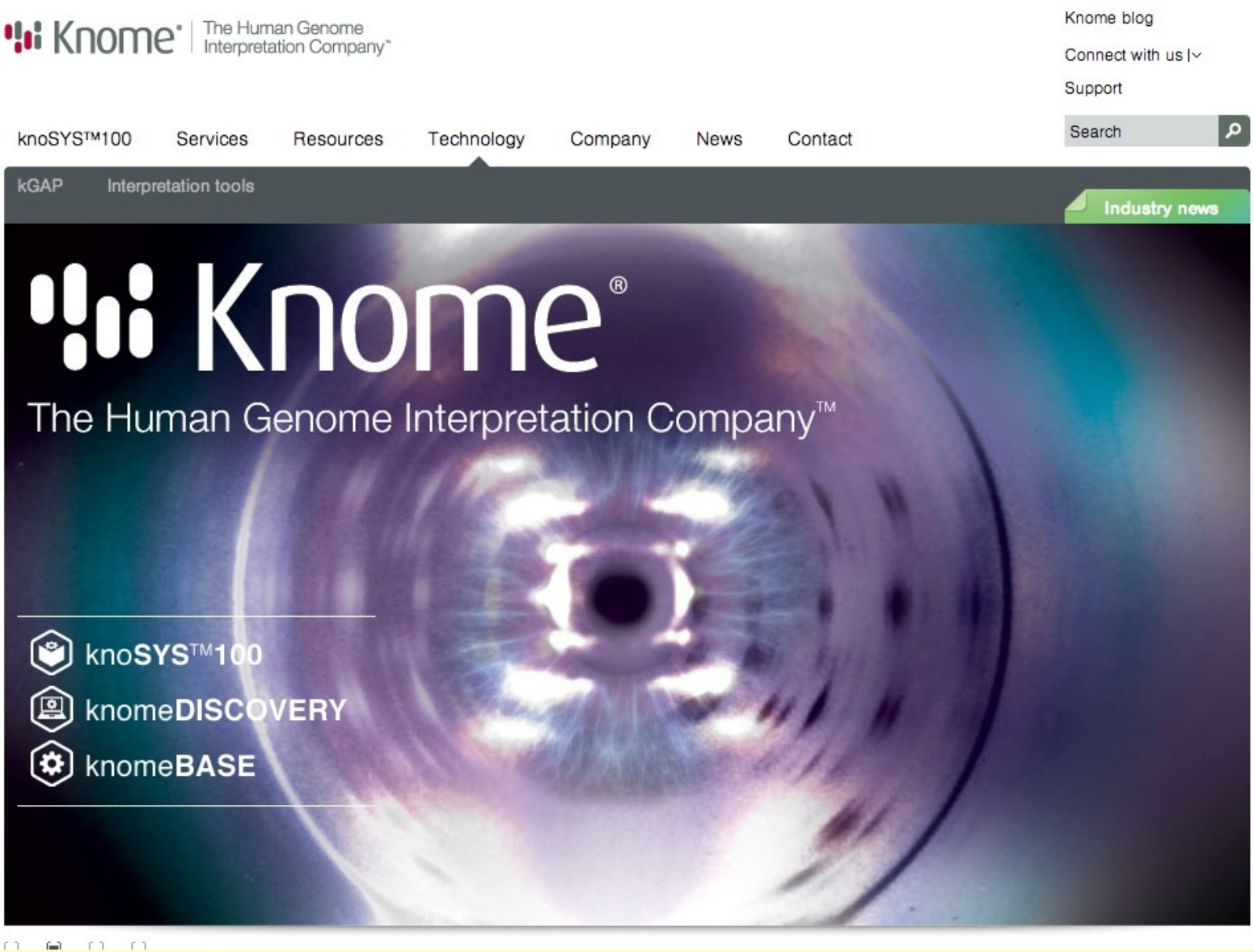
The Genomics Landscape a Decade After the Human Genome Project
Information and events about the 10th anniversary of the the Human Genome Project completion.

Genome: Unlocking Life's Code - Videos and Animations

- YouTube **Genome: Unlocking Life's Code** (2 videos)
- YouTube **History Channel ® Videos** (4 Videos)
- YouTube **Medical Mystery Videos** (3 Videos)
- YouTube **Parts of the Cell, Chromosomes and Genes** (23 animations)
- **What's a Gene?**
  Listen to some well-known scientists respond to the question, "What's a gene?"
- **The African Diaspora: Integrating Culture, Genomics and History**
  Videos from the September 12, 2013 symposium at the Smithsonian's National Museum of Natural History.

Knome

http://www.knome.com/



Complete Genomics

http://www.completegenomics.com/

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Complete Genomics is a leader in accurate whole human genomic sequencing. Using our proprietary sequencing instruments, chemistry, and software, we have sequenced more than 15,000 whole human genomes for our research customers over the past three years. Our mission is to provide the technology for sequencing one million human genomes, enabling researchers and clinicians to improve human health through the prevention, diagnosis, and treatment of genetic diseases and conditions.

BGI-Shenzhen Completes Acquisition of Complete Genomics

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Read the ground breaking Nature paper on LFR Technology

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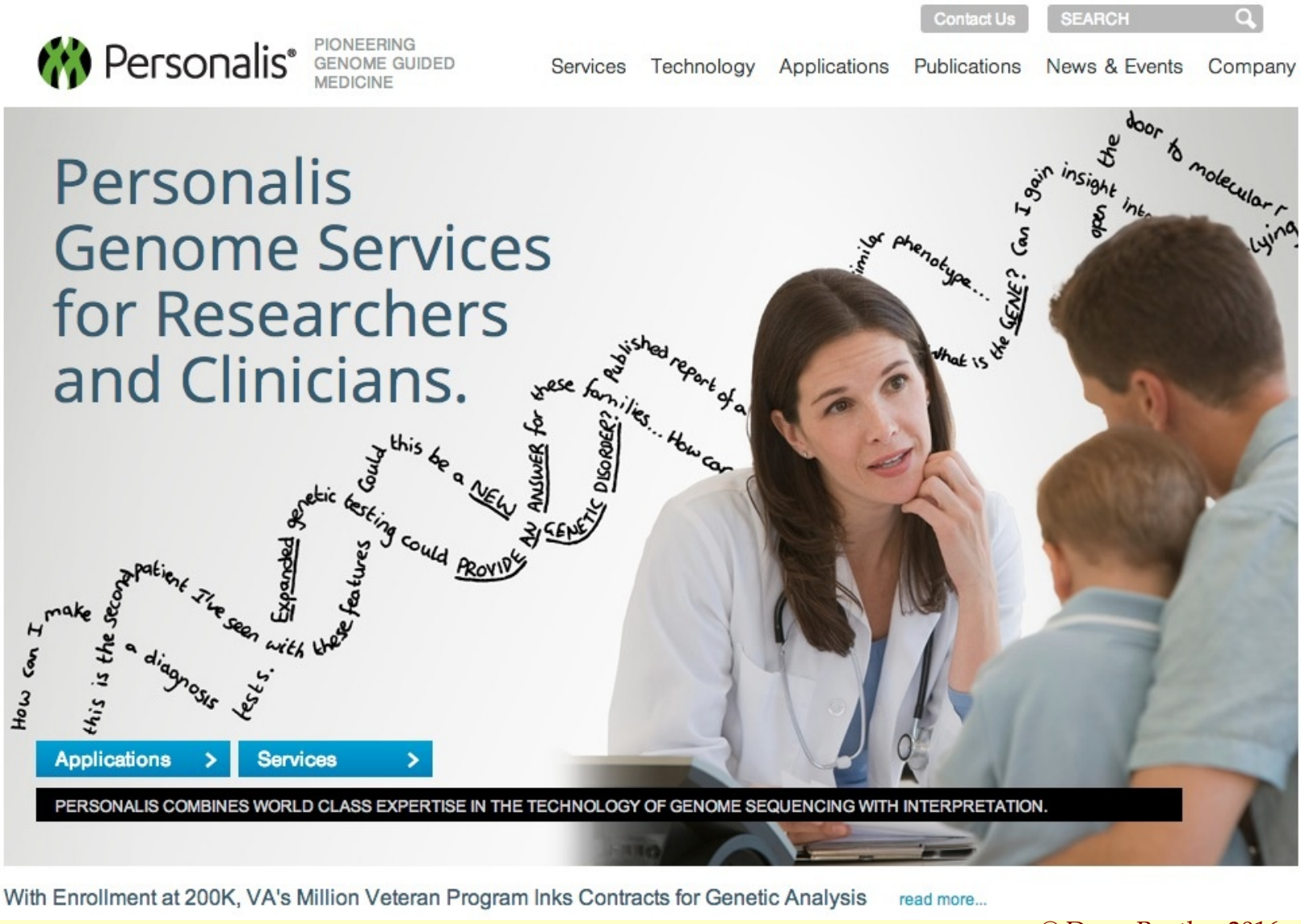
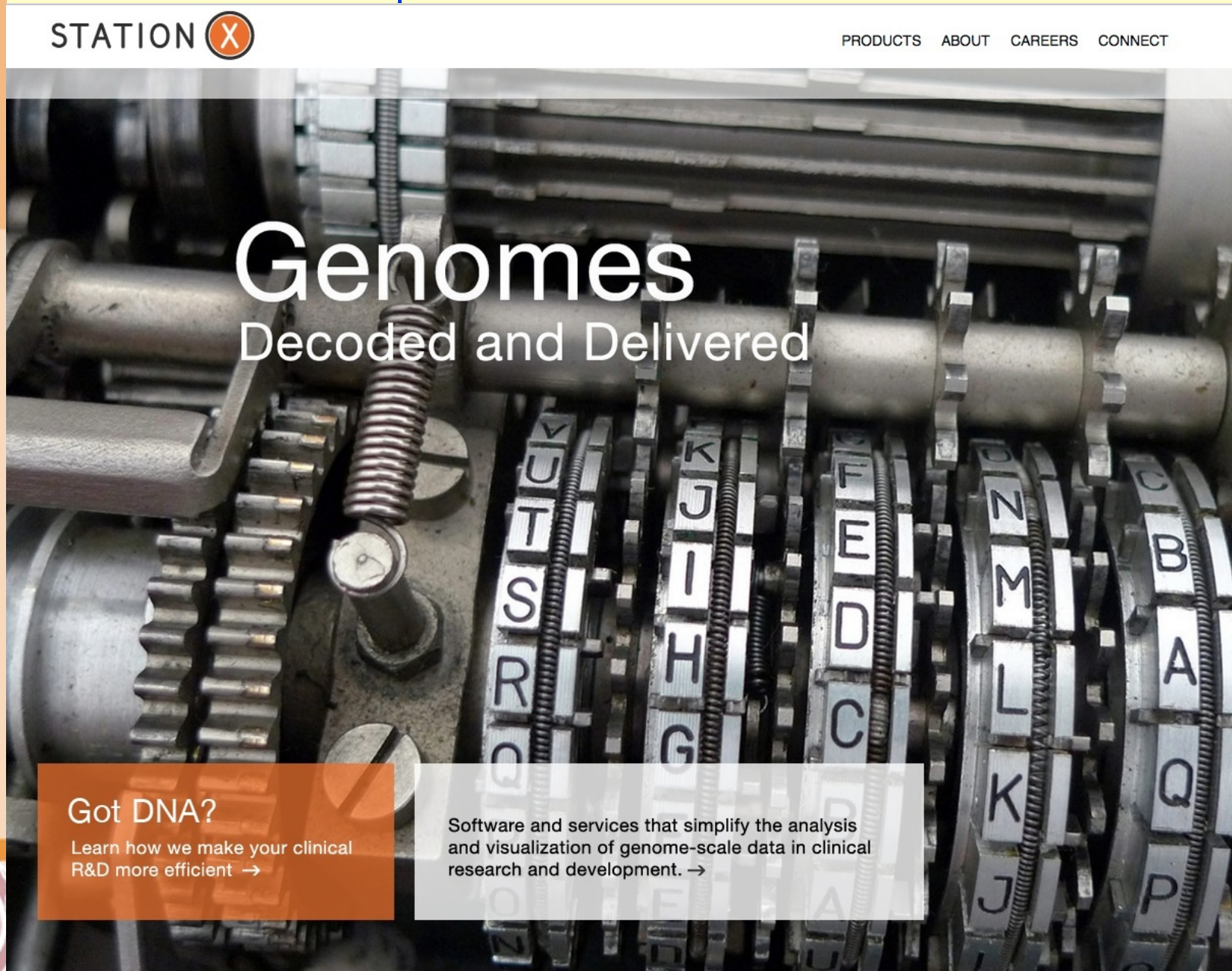


StationX

http://www.stationxinc.com/



Omicia

http://www.omicia.com/

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

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» 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

Portrait of a Glitch

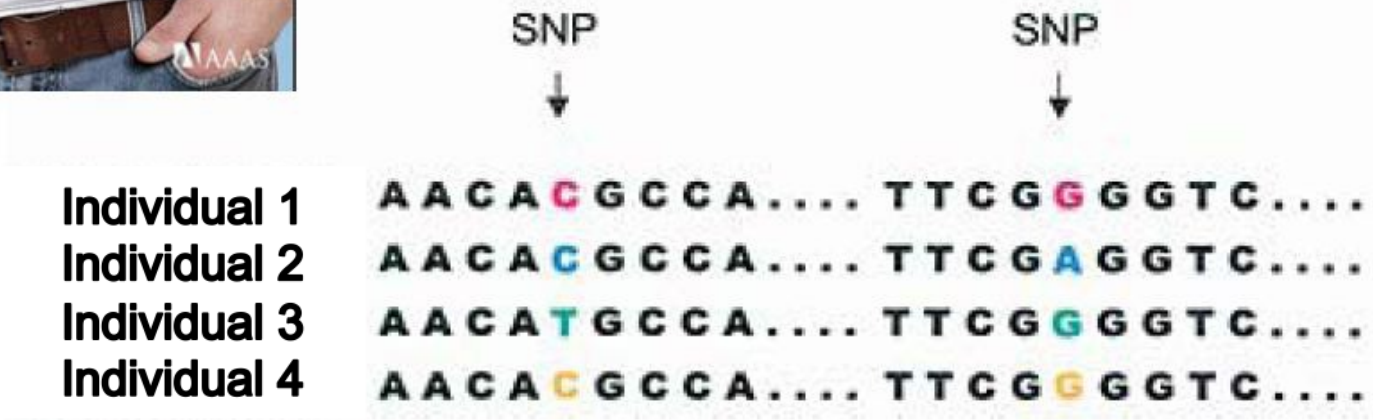
- Revere La Noue, MFA, Stanford, 2005
- What is this film about?
- What classes of glitches are mentioned?
- What do these glitches cause?
- Why did I show this film?

2007 SCIENTIFIC BREAKTHROUGH OF THE YEAR

Science Magazine, December 21, 2007

"It's all about me!"

Single Nucleotide Polymorphisms (SNPs)



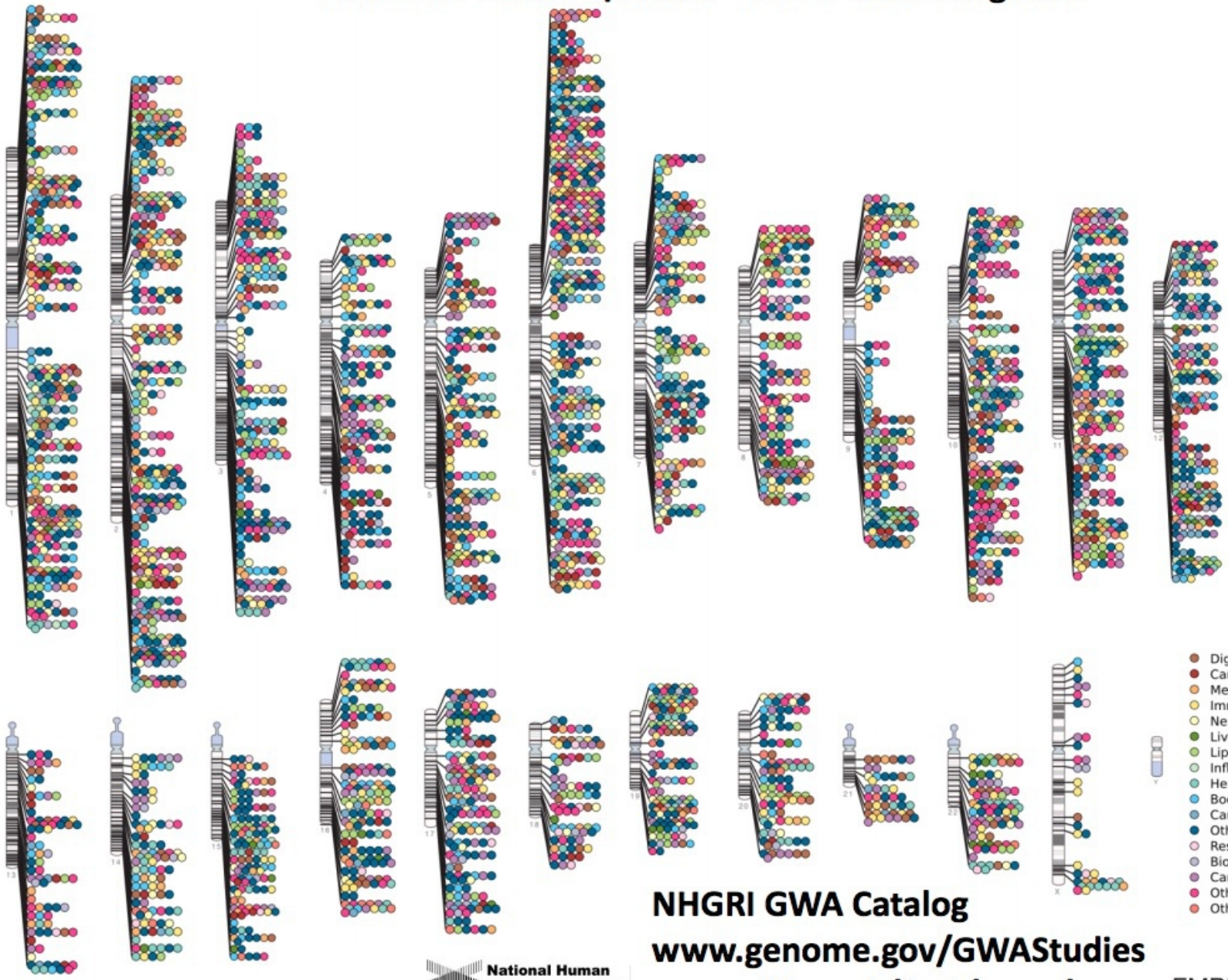
The Great Wave of GWAS Studies

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

Hokusai, K. *The Great Wave*

Published Genome-Wide Associations through 12/2013

Published GWA at p≤5X10^{-8} for 17 trait categories



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

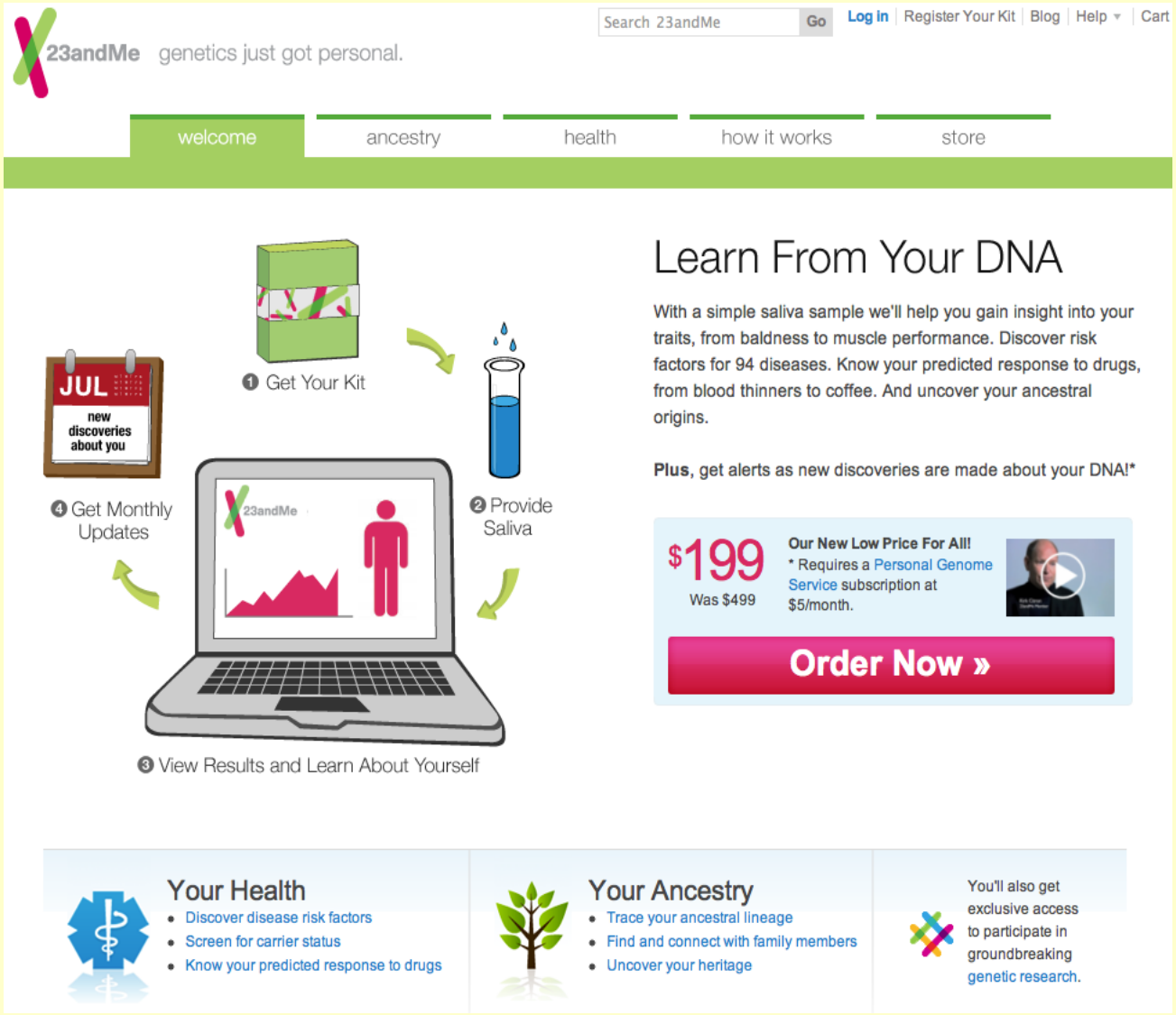
National Human Genome Research Institute

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

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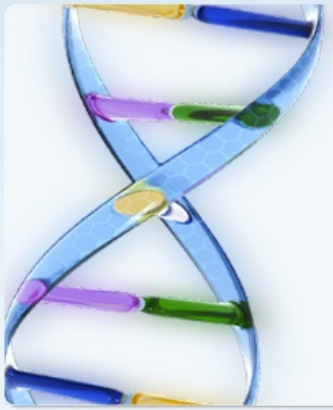
Gene-ius.
A smart way to look at your health.

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

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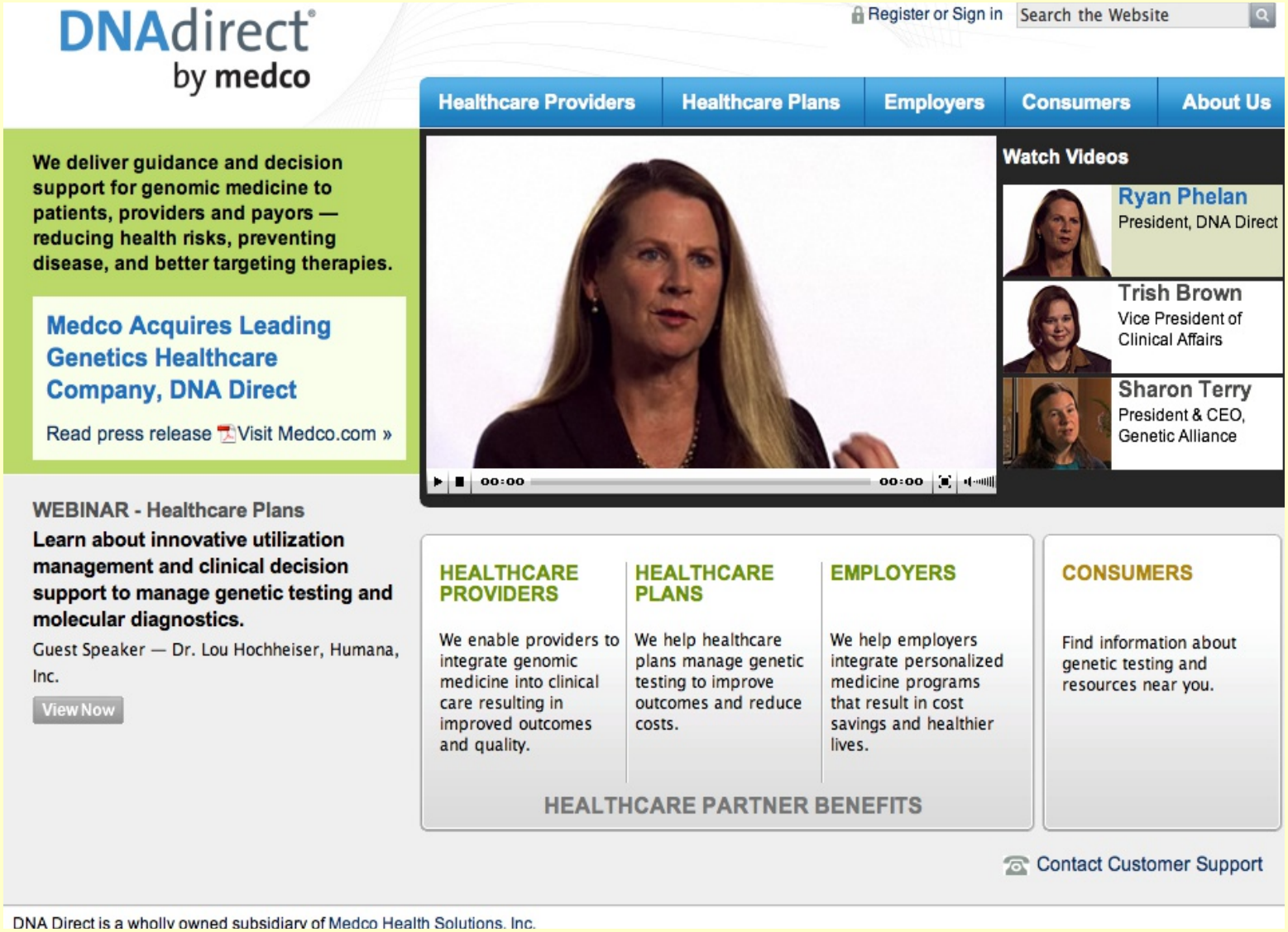
DNAdirect

DNAdirect: Clinical Genetic Testing

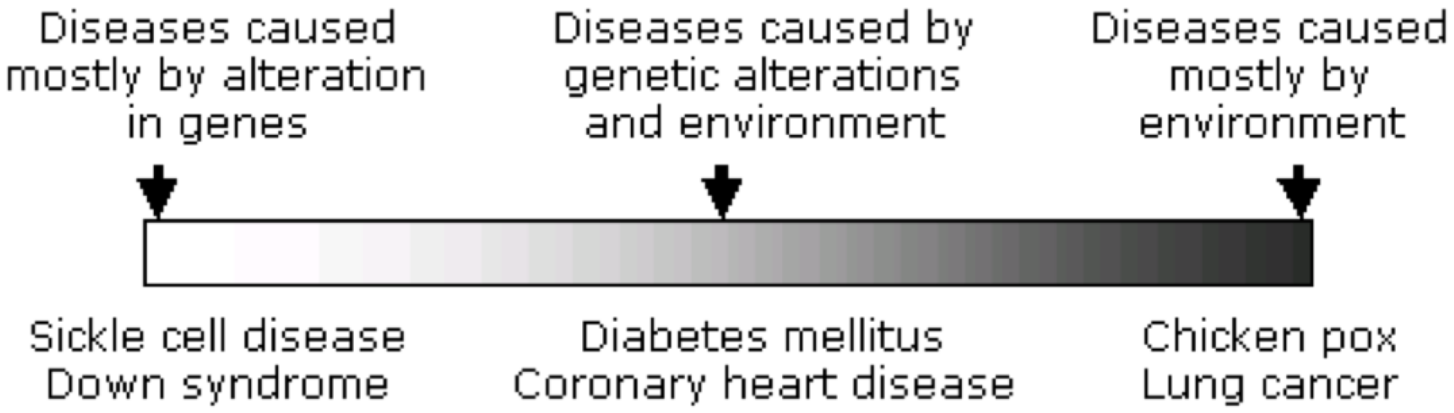


http://www.dnadirect.com/web/

INFORMED Medical Decisions

http://informeddna.com/

Genetic Penetrance



Genetic diseases, at the left of the spectrum, are categorized as **single gene** or **chromosomal** disorders, depending on the specific genetic cause.

Diseases in the middle of the spectrum — including most common diseases — are **multifactorial**, and result from the interaction or additive effect of genetic and non-genetic factors.