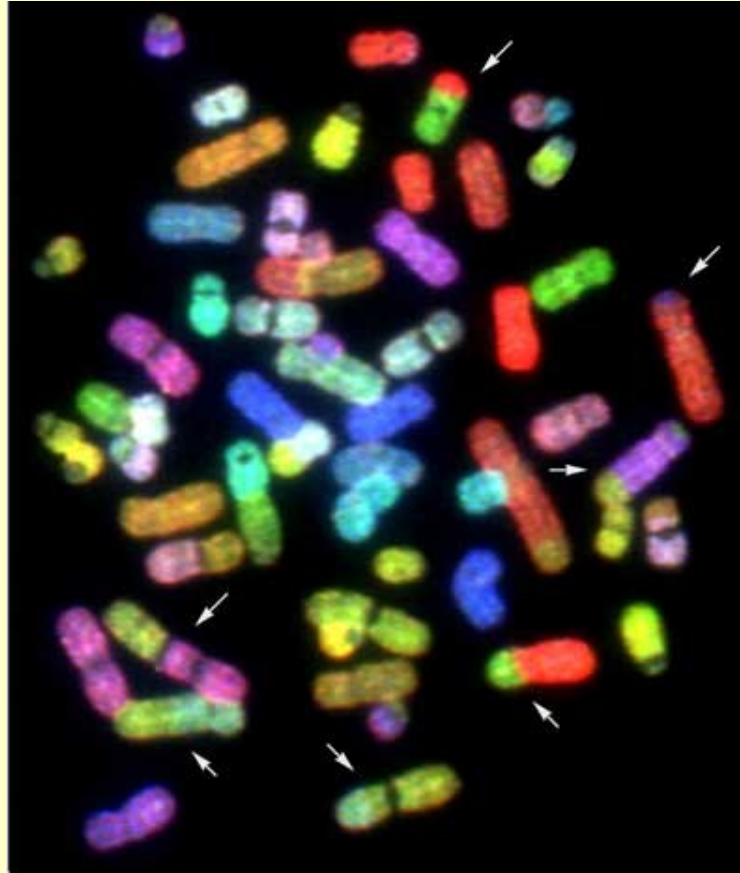


Diseases and Disease Databases

<http://biochem118.stanford.edu/>



Doug Brutlag

Departments of Biochemistry & Medicine
Stanford University School of Medicine



Portrait of a Glitch

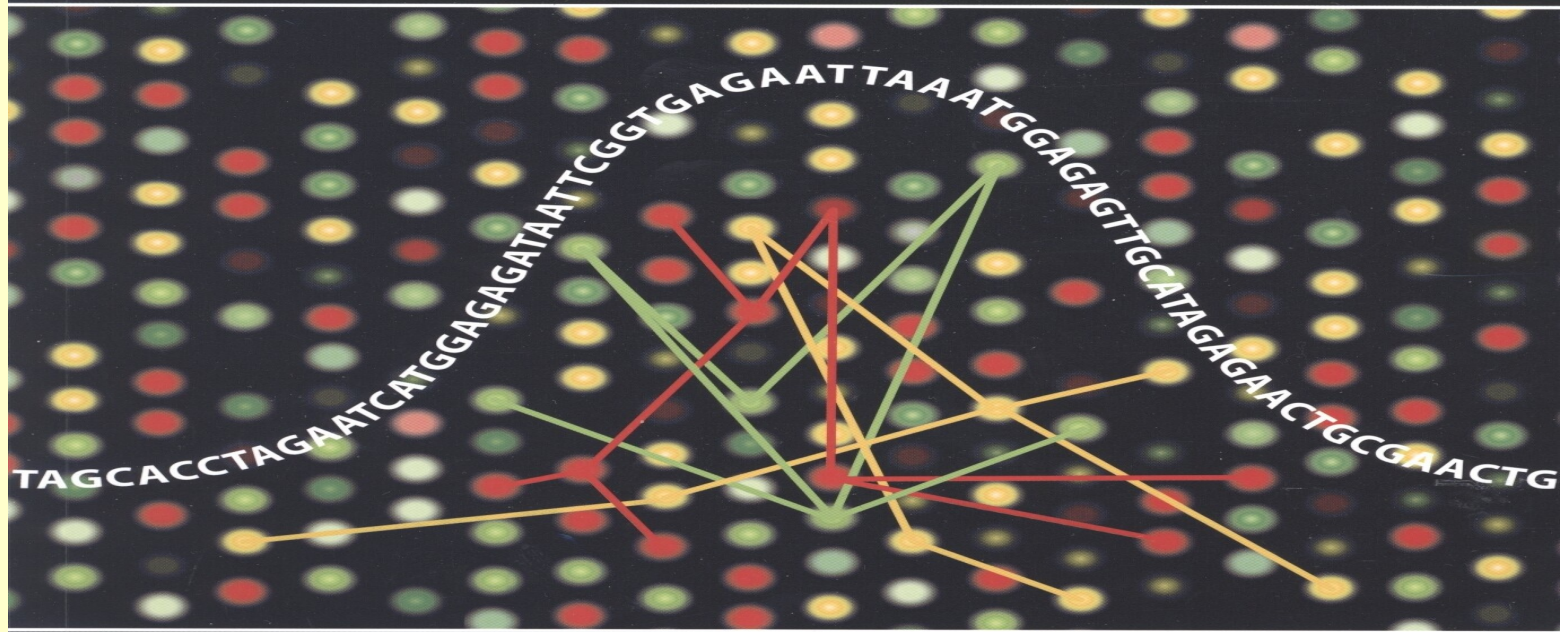
- Revere La Noue, MFA, Stanford, 2005
- What is this film about?
- What classes of glitches are mentioned?
- What do these glitches cause?
- Why did I show this film?



Greg Gibson & Spencer V. Muse
A Primer of Genome Science

A Primer of Genome Science

THIRD
EDITION



GREG GIBSON • SPENCER V. MUSE

\$65 Amazon



Doug Brutlag 2010

Huntington's Disease

- Neurodegenerative disease
 - Loss of movement control
 - Loss of cognitive skills 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

The Inheritance

- You are 19 years old.
- Your father abandoned you and your mother when you only 2 years old.
- Your father died this year at 45 years of age and left you an inheritance.
- He died from an autosomal dominant disease known as Huntington's Chorea or Huntington Disease (HD).
- Since Huntington's is autosomal dominant, 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, at approximately what age you will suffer its symptoms.
- If you test positive, there is nothing medical that can be done to prevent the disease progression.
- Would you take the genetic test or not?
- Why?

Genes and Disease

<http://www.ncbi.nlm.nih.gov:80/books/bv.fcgi?rid=gnd>



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Introduction to Genes and Disease

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. You can browse through the articles online, and you can also download a printable file (PDF) of each chapter.

From *Genes and Disease* you can delve into many online related resources with free and full access. For example, you can visit the human genome to see the location of the genes implicated in each disorder. You can also find related gene sequences in different organisms. And for the very latest information, you can search for complete research articles, and look in other books in the NCBI Bookshelf.

Currently over 80 genetic disorders have been summarized, and the content of *Genes and Disease* is continually growing. Your ideas and suggestions are welcome. You can contact us at: info@ncbi.nlm.nih.gov.



Genes & Disease

<http://www.ncbi.nlm.nih.gov:80/books/bv.fcgi?rid=gnd>

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Huntington's Disease

Genes and Disease



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NCBI » Bookshelf » Genes and Disease » Huntington disease

Huntington disease

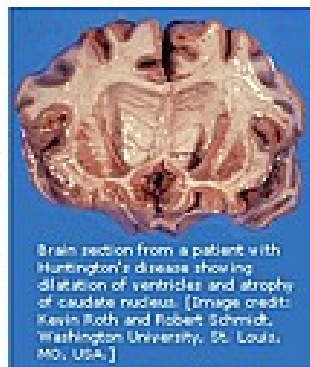
Huntington disease (HD) is an inherited, degenerative neurological disease that leads to dementia. About 30,000 Americans have HD and about 150,000 more are at risk of inheriting the disease from a parent.

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.

With the discovery of the HD gene, a new predictive test was developed that allows those at risk to find out whether or not they will develop the disease. Animal models have also been developed, and we know that mice have a gene that is similar to the human HD gene. Research on understanding the mechanism that causes the triplet repeat to increase is ongoing, since its discovery could be critical to the development of an effective treatment for this and other similar diseases.

Related diseases

[See other Diseases of the Nervous System](#)



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

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[Blink](#) related sequences in different organisms

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[OMIM](#) catalog of human genes and disorders

[GeneReviews](#) a medical genetics resource

Websites

[Huntington Disease Society of America](#) information for patients and the public

Genetics Home Reference



Genetics Home Reference

Your Guide to Understanding Genetic Conditions

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- osteopetrosis
- GRN-related frontotemporal dementia
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Detecting genetic disorders for early treatment

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Genetic Disorders A to Z and related genes and chromosomes

Genetic Conditions

The genetics of more than 550 health conditions, diseases, and syndromes.



Genes

More than 750 genes, health effects of genetic differences, and gene families.



Chromosomes

Chromosomes, mitochondrial DNA, and associated health conditions.



Concepts & Tools for understanding human genetics

Handbook

Learn about mutations, inheritance, genetic counseling, genetic testing, genomic research, and more.



Glossary

Medical and genetics definitions.



Resources

Links to other genetics information and organizations.



Genetics Home Reference provides consumer-friendly information about the effects of genetic variations on human health.

The resources on this site should not be used as a substitute for professional medical care or advice. Users seeking information about a personal genetic disease, syndrome, or condition should consult with a qualified healthcare professional. See [How can I find a genetics professional in my area?](#) in the Handbook.

Published: September 19, 2010



Doug Brutlag 2010

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Genetic Disorders A to Z and related genes and chromosomes

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Genetics Home Reference on HD



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Huntington disease

On this page: [Description](#) [Genetic changes](#) [Inheritance](#) [Treatment](#) [Additional information](#)
[Other names](#) [Glossary definitions](#)

Reviewed October 2008

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Genetic disorder catalog

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.

What genes are related to Huntington disease?

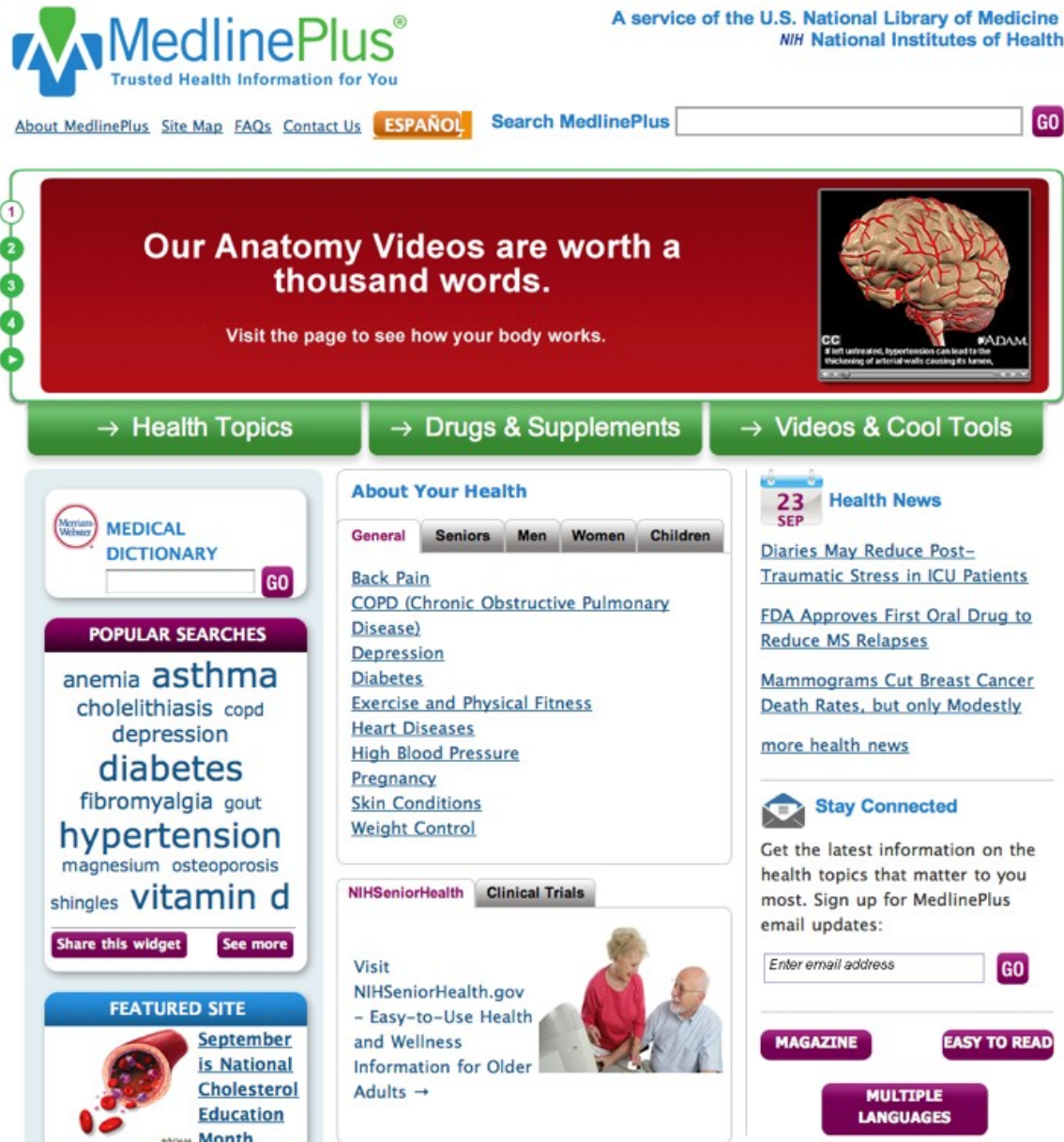

Mutations in the [HTT](#) gene cause Huntington disease. The HTT gene provides instructions for making a protein called huntingtin. Although the function of this protein is unknown, it appears to play an important role in nerve cells (neurons) in the brain.



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Medline Plus (NLM)

<http://www.nlm.nih.gov/medlineplus/medlineplus.html>



The screenshot shows the MedlinePlus website interface. At the top, the MedlinePlus logo is on the left, and the text "A service of the U.S. National Library of Medicine NIH National Institutes of Health" is on the right. Below the logo is a search bar with the text "Search MedlinePlus" and a "GO" button. A navigation menu includes links for "About MedlinePlus", "Site Map", "FAQs", "Contact Us", "ESPAÑOL", and "Search MedlinePlus".

A large red banner features the text "Our Anatomy Videos are worth a thousand words." and "Visit the page to see how your body works." To the right of the banner is an image of a brain with red vessels. Below the banner are three green buttons: "→ Health Topics", "→ Drugs & Supplements", and "→ Videos & Cool Tools".

The main content area is divided into three columns:

- Left Column:** A "MEDICAL DICTIONARY" search box with a "GO" button. Below it is a "POPULAR SEARCHES" section listing terms like anemia, asthma, cholelithiasis, copd, depression, diabetes, fibromyalgia, gout, hypertension, magnesium, osteoporosis, shingles, and vitamin d. At the bottom of this section are "Share this widget" and "See more" buttons. Below that is a "FEATURED SITE" section for "September is National Cholesterol Education Month" with an image of a pill bottle.
- Middle Column:** An "About Your Health" section with tabs for "General", "Seniors", "Men", "Women", and "Children". Under the "General" tab, there is a list of health topics: Back Pain, COPD (Chronic Obstructive Pulmonary Disease), Depression, Diabetes, Exercise and Physical Fitness, Heart Diseases, High Blood Pressure, Pregnancy, Skin Conditions, and Weight Control. Below this is a "NIHSeniorHealth Clinical Trials" section with a photo of an elderly couple and the text "Visit NIHSeniorHealth.gov - Easy-to-Use Health and Wellness Information for Older Adults →".
- Right Column:** A "Health News" section dated "23 SEP" with links to "Diaries May Reduce Post-Traumatic Stress in ICU Patients", "FDA Approves First Oral Drug to Reduce MS Relapses", and "Mammograms Cut Breast Cancer Death Rates, but only Modestly". Below this is a "Stay Connected" section with an email sign-up form and a "GO" button. At the bottom of the right column are three buttons: "MAGAZINE", "EASY TO READ", and "MULTIPLE LANGUAGES".

Huntington's in Medline Plus



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Refine by Keyword

All Results (129) remix

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- Brain (12)
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- Harvard School of Public Health (15)
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- 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

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Gene Reviews of Huntington's

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Huntington Disease

[*Huntington Chorea*]

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Rona K Graham, PhD

Department of Medical Genetics
University of British Columbia
Vancouver, BC

Michael R Hayden, MB, ChB, PhD, FRCP(C),FRSC

Department of Medical Genetics
University of British Columbia
Vancouver, BC

Initial Posting: October 23, 1998.

Last Update: July 19, 2007.

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Huntington Disease

[*Huntington Chorea*]

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University of British Columbia
Vancouver, BC

Rona K Graham, PhD

Department of Medical Genetics
University of British Columbia
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Michael R Hayden, MB, ChB, PhD, FRCP(C),FRSC

Department of Medical Genetics
University of British Columbia
Vancouver, BC

Initial Posting: October 23, 1998.

Last Update: July 19, 2007.

Summary

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.

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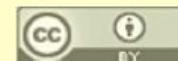
Huntington Disease | Huntington Chorea

Items 1 - 61 of 61

For this search term: [View details of clinical laboratories](#)

Laboratories offering clinical testing:

	Targeted mutation analysis	Linkage analysis
All Children's Hospital Molecular Genetics Laboratory St. Petersburg, FL O Thomas Mueller, PhD, FACMG	●	
ARUP Laboratories Molecular Genetics Laboratory Salt Lake City, UT Elaine Lyon, PhD; Rong Mao, MD; Edward R Ashwood, MD; Pinar Bayrak-Toydemir, MD, PhD	●	
Athena Diagnostics Inc Reference Lab Worcester, MA Sat Dev Batish, PhD, FACMG; Masamichi Ito, PhD, FACMG; Christine M Stanley, PhD, FACMG	●	
Baylor College of Medicine Medical Genetics Laboratories Houston, TX Sau W. Cheung, PhD; Christine M Eng, MD, FACMG; William E O'Brien, PhD; Lee-Jun Wong, PhD	●	
Boston University School of Medicine Center for Human Genetics Boston, MA Aubrey Milunsky, MD, DSc; Jeff Mark Milunsky, MD	●	
Center for Genetic Testing at Saint Francis Genetics Laboratory Tulsa, OK Frederick V Schaefer, PhD, FACMG; Nancy J Carpenter, PhD, FACMG; Michael A Kayser, DO, FACMG	●	
Centogene GmbH Institute of Molecular Diagnostics Rostock, Germany Christoph Ehlers	●	
Centre for Cellular and Molecular Biology Molecular Diagnostics Division, Genome Research Group Hyderabad, India Giriraj Ratan Chandak, MD, PhD, DNB	●	



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The GeneTests database and Web site are now hosted at NCBI.

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09/29/2009

495 *GeneReviews*
1185 Clinics
604 Laboratories testing for
1802 Diseases
1533 Clinical
269 Research

Laboratory Directory Growth Chart

Administrative Use

(To update Clinic / Laboratory Directory listings)

Welcome to GeneTests

Welcome to the GeneTests Web site, a publicly funded medical genetics information resource developed for physicians, other healthcare providers, and researchers, available at no cost to all interested persons. Use of this Web site assumes acceptance of the [terms of use](#).

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Expert-authored peer-reviewed disease descriptions

Laboratory Directory

International directory of genetic testing laboratories

Clinic Directory

International directory of genetics and prenatal diagnosis clinics

Educational Materials

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- ▶ **GeneTests moved to NCBI**
- ▶ **Change in access to Genetic Tools**

New in *GeneReviews*

New Clinical Test Listings

- ▶ **16 new listings**

GeneTests is a supplement to and not a substitute for medical advice. Patients with specific questions about genetic counseling or testing should contact their healthcare provider or a genetics clinic.

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Huntington's Brain

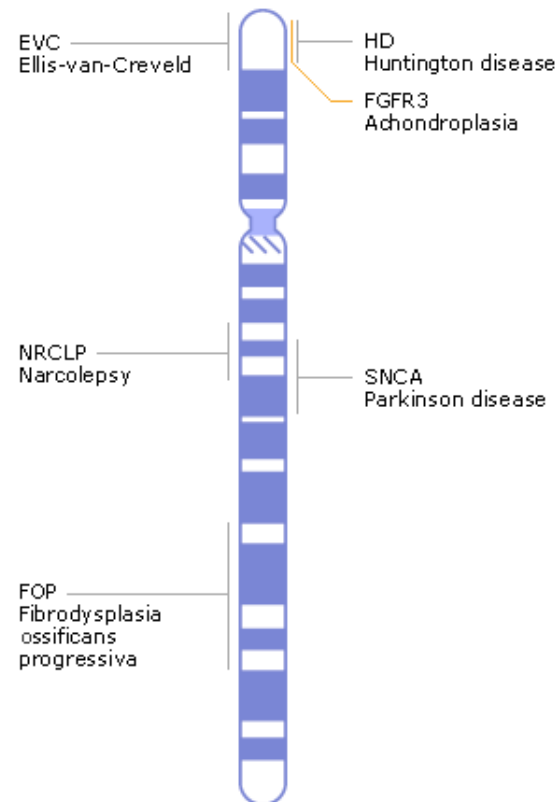


Brain section from a patient with Huntington's disease showing dilatation of ventricles and atrophy of caudate nucleus. [Image credit: Kevin Roth and Robert Schmidt, Washington University, St. Louis, MO, USA.]

Chromosome 4

Chromosome 4

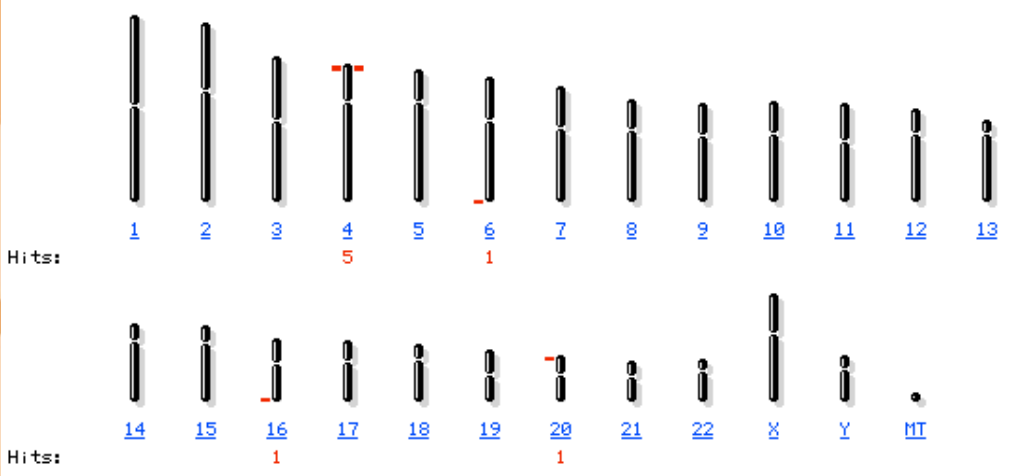
- Contains approximately 1600 genes
- Contains approximately 190 million base pairs, of which ~95% have been determined
- See the diseases associated with chromosome 4 in the [MapViewer](#)



Genome View

Homo sapiens genome view
Build 35.1 statistics

BLAST search the human genome



Search results for query "Huntington": 8 hits

Chr	Assembly	Match	Map element	Type	Maps
4	reference	all matches			
		Huntington disease-like 3	HLN2	MIM	Pheno Morbid
		Huntington disease	HD	MIM	Pheno Morbid
		huntingtin (Huntington disease)	HD	Gene	Genes_seq Genes_cyto
4	Celera	all matches			
		Huntington disease-like 3	HLN2	MIM	Pheno
		huntingtin (Huntington disease)	HD	Gene	Genes_seq
6	reference	Spinocerebellar ataxia 17, 607136; Parkinson disease, 168600...	TBP	MIM	Pheno Morbid
16	reference	Huntington disease-like 2, 606438	JPH3	MIM	Pheno Morbid
20	reference	Creutzfeldt-Jakob disease, 123400; Gerstmann-Straussler disease...	176640	MIM	Morbid Pheno

Gene Resources for Huntington

Jan 12 2009 14:16 PST

Huntington Disease Resources

- **Caring for People with Huntington's Disease**
www.kumc.edu/hospital/huntingtons/index.html
- **High Q Foundation**
350 Seventh Avenue Suite 601
New York NY 10001
Phone: 212-239-9300
Fax: 212-239-2101
Email: Please see the contacts page located at www.HighQFoundation.org/contacts
www.highqfoundation.org
- **Huntington Society of Canada**
151 Frederick Street Suite 400
Kitchener ON N2H 2M2
Canada
Phone: 800-998-7398; 519-749-7063
Fax: 519-749-8965
Email: info@hsc-ca.org
www.hsc-ca.org
- **Huntington's Disease Society of America (HDSA)**
505 Eighth Avenue
New York NY 10018
Phone: 800-345-HDSA (800-3345-4372); 212-242-1968
Fax: 212-239-3430
Email: hdsainfo@hdsa.org
www.hdsa.org
- **International Huntington Association**
Email: iha@huntington-assoc.com
www.huntington-assoc.com
- **National Library of Medicine Genetics Home Reference**
[Huntington disease](#)
- **NCBI Genes and Disease**
[Huntington disease](#)
- **Testing for Huntington Disease: Making an Informed Choice**
Booklet providing information about Huntington disease and
depts.washington.edu/neurogen/HuntingtonDis.pdf



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- Biochemical Features
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- Mapping
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- Diagnosis
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HUNTINGTON DISEASE; HD

Alternative titles; symbols

HUNTINGTON CHOREA

Gene map locus [4p16.3](#)

TEXT

A number sign (#) is used with this entry because Huntington disease (HD) is caused by an expanded trinucleotide repeat in the gene encoding huntingtin (HTT; [613004](#)) on chromosome 4p16.3.

DESCRIPTION

Huntington disease (HD) is an autosomal dominant progressive neurodegenerative disorder with a distinct phenotype characterized by chorea, dystonia, incoordination, cognitive decline, and behavioral difficulties. There is progressive, selective neural cell loss and atrophy in the caudate and putamen. [Walker \(2007\)](#) provided a detailed review of Huntington disease, including clinical features, population genetics, molecular biology, and animal models. 🧠

CLINICAL FEATURES

The classic signs of Huntington disease are progressive chorea, rigidity, and dementia. A characteristic atrophy of the caudate nucleus is seen radiographically. Typically, there is a prodromal phase of mild psychotic and behavioral symptoms which precedes frank chorea by up to 10 years. [Chandler et al. \(1960\)](#) observed that the age of onset was between 30 and 40 years. In a study of 196 kindreds. [Reed a](#)

- Clinical Synopsis
- Gene map

- Entrez Gene
- N Nomenclature
 - R RefSeq
 - G GenBank
 - P Protein
 - U UniGene

- LinkOut
- ...genage
 - ...HGMD
 - ...GAD
 - ...CORIELL
 - ...komp
 - ...MGI

OMIM Statistics



Johns
Hopkins
University

All Databases

PubMed

Nucleotide

Protein

Genome

Structure

PMC

Taxonomy

OMIM

OMIM Statistics for August 14, 2010

Number of Entries

	Autosomal	X-Linked	Y-Linked	Mitochondrial	Total
* Gene with known sequence	12475	611	48	35	13169
+ Gene with known sequence and phenotype	346	20	0	2	368
# Phenotype description, molecular basis known	2576	227	4	28	2835
% Mendelian phenotype or locus, molecular basis unknown	1636	136	5	0	1777
Other, mainly phenotypes with suspected mendelian basis	1853	134	2	0	1989
Total	18886	1128	59	65	20138

Entrez

OMIM

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Map

Help

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How to Link

FAQ

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Citing OMIM

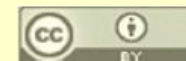
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Statistics

Update Log

Restrictions on



OMIM Links

MIM ID #143100 HUNTINGTON DISEASE; HD

[GeneTests, Links](#)

Alternative titles; symbols
HUNTINGTON CHOREA

Gene map locus: [4p16.3](#)

[Clinical Synopsis](#)

Text

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A number sign (#) is used with this entry because Huntington disease (HD) is caused by an expanded trinucleotide repeat (CAG)_n, encoding glutamine, in the gene encoding huntingtin (HTT; [613004](#)) on chromosome 4p16.3.

In normal individuals, the range of repeat numbers is 9 to 36. In those with HD, the repeat number is above 37 (Duyao et al., 1993).

Description

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Huntington disease (HD) is an autosomal dominant progressive neurodegenerative disorder with a distinct phenotype characterized by chorea, dystonia, incoordination, cognitive decline, and behavioral difficulties. There is progressive, selective neural cell loss and atrophy in the caudate and putamen. Walker (2007) provided a detailed review of Huntington disease, including clinical features, population genetics, molecular biology, and animal models.

Clinical Features

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The classic signs of Huntington disease are progressive chorea, rigidity, and dementia. A characteristic atrophy of the caudate nucleus is seen radiographically. Typically, there is a prodromal phase of mild psychotic and behavioral symptoms which precedes frank chorea by up to 10 years. Chandler et al. (1960) observed that the age of onset was between 30 and 40 years. In a study of 196 kindreds, Reed and Neel (1959) found only 8 in which both parents of a single patient with Huntington chorea were 60 years of age or older and normal. The clinical features developed progressively with severe increase in choreic movements and dementia. The disease terminated in death on average 17 years after manifestation of the

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[Books](#)
[Free in PMC](#)





Symbol Report: **HTT**



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Giving unique and meaningful names to every human gene

 [Quick Gene Search](#)

Core Data		Database Links			
Approved Symbol +	HTT	RefSeq IDs +			
Approved Name +	huntingtin	NM_002111	GenBank	EMBL	DDBJ UCSC
HGNC ID +	HGNC:4851	Accession Numbers +			
Status +	Approved	L12392	GenBank	EMBL	DDBJ UCSC
Chromosome +	4p16.3	Mouse Genome Database ID +			
Previous Symbols +	HD	MGI:96067	MGD ID		
Previous Names +	"huntingtin (Huntington disease)"	Rat Genome Database ID (mapped data supplied by RGD) +			
Aliases +	IT15	RGD:68337	RGD ID		
Name Aliases +		Entrez Gene ID +			
Locus Type +	gene with protein product	3064	Gene	Map Viewer	
		CCDS IDs +			
Gene Symbol Links		CCDS43206.1	CCDS ID		
GENATLAS GeneCards GeneClinics / GeneTests GoPubMed		Pubmed IDs +			
HCOP	H-InvDB	8458085	PMID	CiteXplore	
Treefam	wikigenes	Ensembl ID (mapped data supplied by Ensembl) +			
		ENSG00000197386	Ensembl GeneView	UCSC	
Specialist Database Links		OMIM ID (mapped data supplied by NCBI) +			
COSMIC Orphanet:16190		613004	OMIM		
		UCSC ID (mapped data supplied by UCSC) +			
		uc010icr.1	UCSC Index		
		UniProt ID (mapped data supplied by UniProt) +			
		P42858	UniProt	UCSC	

1: HTT huntingtin [*Homo sapiens*]

GeneID: 3064

updated 16-Sep-2009

Summary

Entrez Gene Home

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 Probe
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 SNP: Genotype
 SNP: GeneView
 Taxonomy
 UniSTS
 AceView
 Ensembl
 Evidence Viewer
 GeneTests for MIM: 143100
 HGNC
 HPRD

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

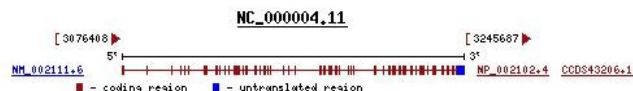
Organism *Homo sapiens*

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]

Genomic regions, transcripts, and products

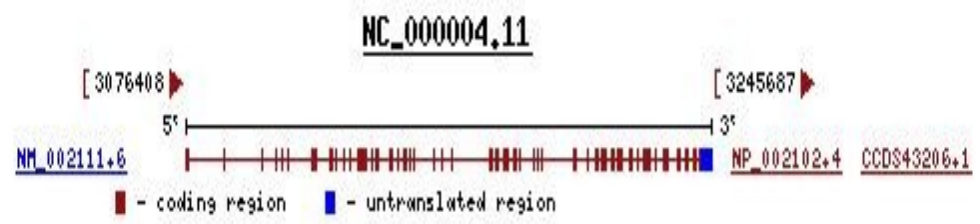
Go to [reference sequence details](#)[Try our new Sequence Viewer](#)

Genomic regions, transcripts, and products



Go to [reference sequence details](#)

[Try our new Sequence Viewer](#)

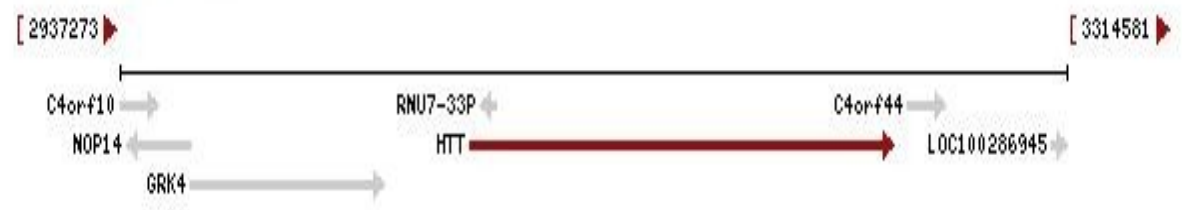


Genomic context



chromosome: 4; Location: 4p16.3

[See HTT in MapViewer](#)



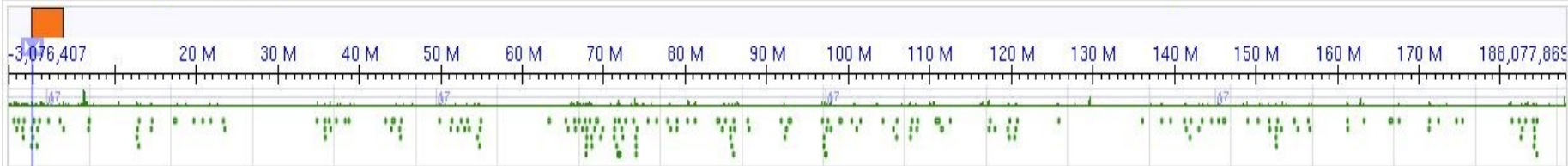
NCBI Reference Sequence: NC_000004.11

Homo sapiens chromosome 4, GRCh37 primary reference assembly

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NC_000004.11 (191154276 bases) (Sequence origin: 3076407)

Sequence Set Origin Views & Tools Markers Find gene...



-77313 : 290409 (367722 bases shown, positive strand)

Sequence Flip Strands Tools Markers Default Options



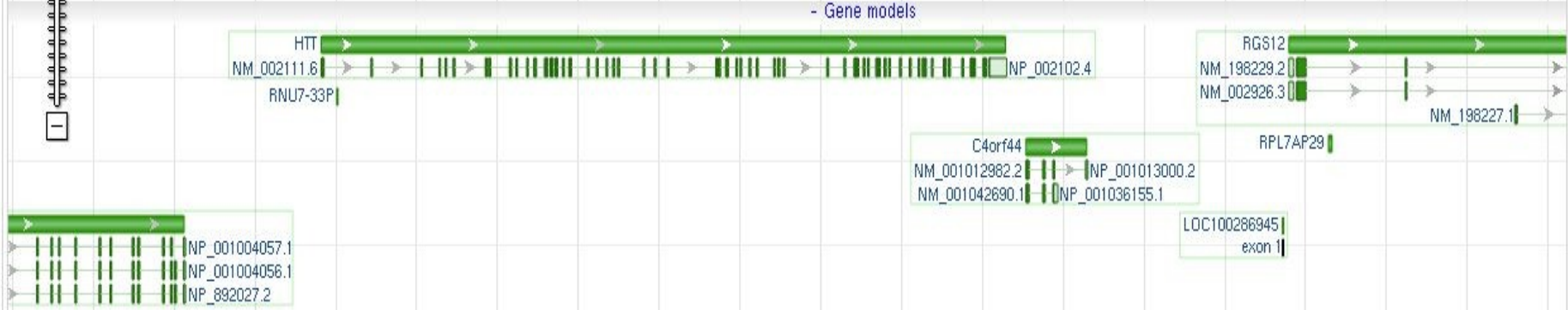
reference assembly - NC_000004.11: Homo sapiens chromosome 4, GRCh37 primary reference assembly

Variations, density (seq-feats)

Component map, other segments: 1

NT_006051.18

Gene models



MapView of HTT Gene

Homo sapiens (human) Build 37.1 (Current)

[BLAST The Human Genome](#)

Chromosome: [1](#) [2](#) [3](#) [[4](#)] [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) [11](#) [12](#) [13](#) [14](#) [15](#) [16](#) [17](#) [18](#) [19](#) [20](#) [21](#) [22](#) [X](#) [Y](#) [MT](#)

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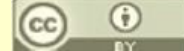
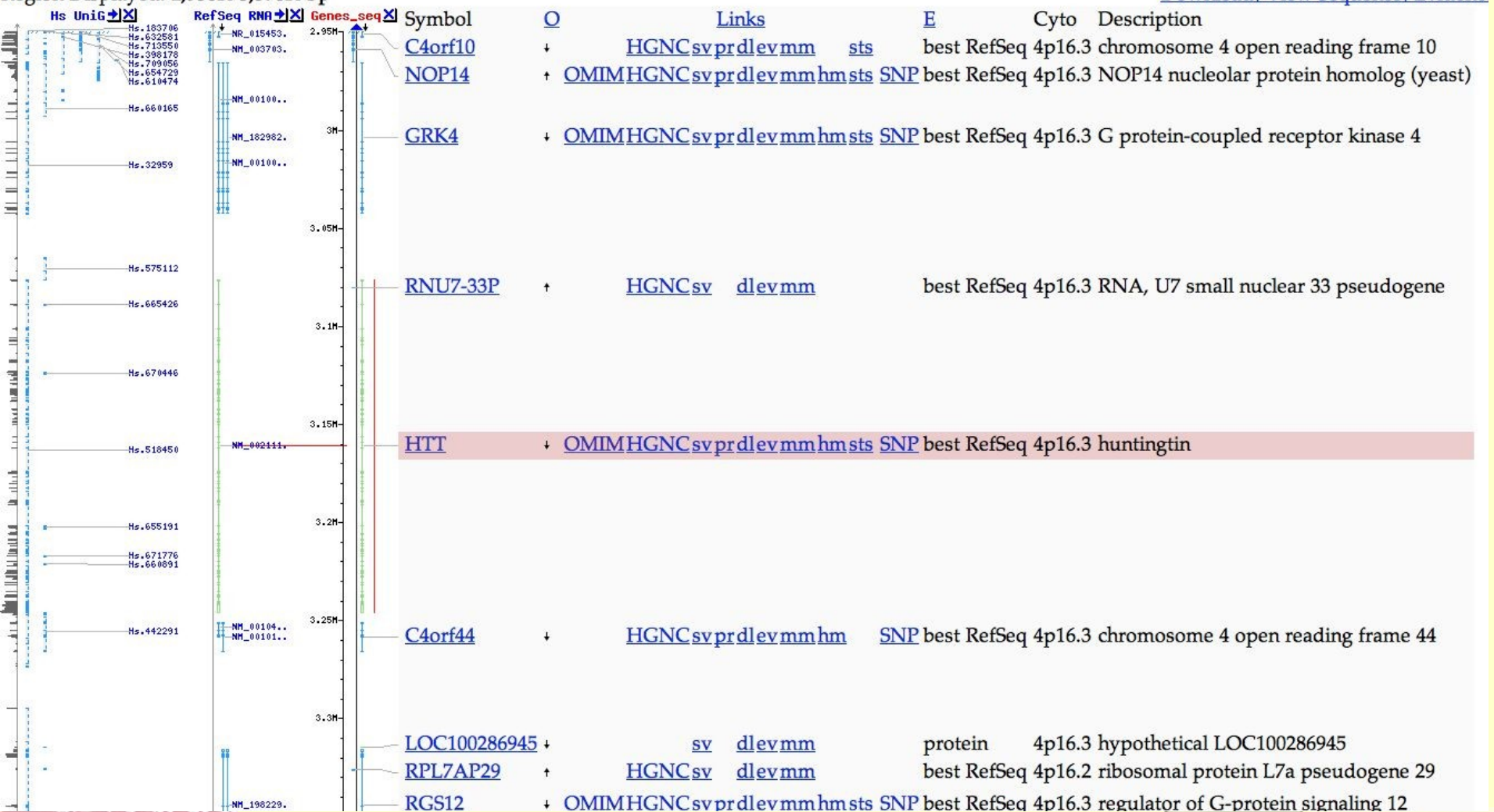
Master Map: Genes On Sequence

[Summary of Maps](#)

[Maps & Options](#)

Region Displayed: 2,950K-3,370K bp

[Download/View Sequence/Evidence](#)



Huntingtin Protein

NCBI Reference Sequence: NP_002102.4

huntingtin [Homo sapiens]

[Comment](#) [Features](#) [Sequence](#)

LOCUS NP_002102 3144 aa linear PRI 18-SEP-2009
 DEFINITION huntingtin [Homo sapiens].
 ACCESSION NP_002102
 VERSION NP_002102.4 GI:90903231
 DBSOURCE REFSEQ: accession [NM_002111.6](#)
 KEYWORDS .
 SOURCE Homo sapiens (human)
 ORGANISM [Homo sapiens](#)
 Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
 Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
 Catarrhini; Hominidae; Homo.
 REFERENCE 1 (residues 1 to 3144)
 AUTHORS Harper,S.Q.
 TITLE Progress and challenges in RNA interference therapy for Huntington
 disease
 JOURNAL Arch. Neurol. 66 (8), 933-938 (2009)
 PUBMED [19667213](#)
 REMARK GeneRIF: Reducing mutant huntingtin expression may offer a
 treatment for Huntington disease. RNA interference has emerged as a
 powerful method to silence dominant disease genes.
 Review article
 REFERENCE 2 (residues 1 to 3144)
 AUTHORS Morfini,G.A., You,Y.M., Pollema,S.L., Kaminska,A., Liu,K.,
 Yoshioka,K., Bjorkblom,B., Coffey,E.T., Bagnato,C., Han,D.,
 Huang,C.F., Banker,G., Pigino,G. and Brady,S.T.
 TITLE Pathogenic huntingtin inhibits fast axonal transport by activating
 JNK3 and phosphorylating kinesin
 JOURNAL Nat. Neurosci. 12 (7), 864-871 (2009)
 PUBMED [19525941](#)
 REMARK GeneRIF: data identify JNK3 as a critical mediator of pathogenic
 Htt (polyQ-Htt)toxicity and provide a molecular basis for
 polyQ-Htt-induced inhibition of fast axonal transport

Hu

huntingtin [Homo sapiens]

(TA)

```

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PGSPYHRLLTCLRNVHKVTTTC

```



SNP Viewer for Huntington

http://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?locusId=3064

SNP linked to Gene [HTT](#)([geneID:3064](#)) Via Contig Annotation

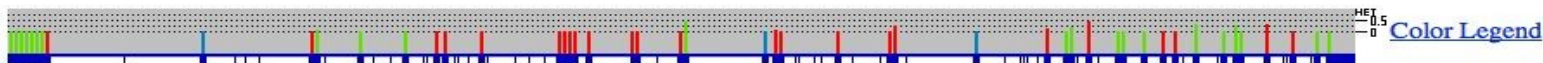
Send rs# on all gene models to Batch Query Download all rs# to file. Genotype

Gene Model (mRNA alignment) information from genome sequence

Total gene model (contig mRNA transcript):				3		
mrna	transcript	protein	mrna orientation	Contig	Contig Label	List SNP
NM_002111.6	plus strand	NP_002102.4	forward	NT_006081.18	reference	<- currently shown
NM_002111.6	plus strand	NP_002102.4	forward	NW_921918.1	Celera	View snp on GeneModel
NM_002111.6	minus strand	NP_002102.4	reverse	NW_001838896.2	HuRef	View snp on GeneModel

Include clinically associated in gene region cSNP has frequency double hit refresh

gene model (contig mRNA transcript):	Contig Label	Contig	mrna	protein	mrna orientation	transcript	snp count
reference	NT_006081.18	NM_002111.6	NP_002102.4		forward	plus strand	49, coding



Region	Contig position	mRNA pos	dbSNP rs# cluster id	Heterozygosity	Validation	3D	Clinically Associated	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos	PubMed
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								contig reference	-		1	10	
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								contig reference	-		3	17	
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								contig reference	G	Gln [Q]	3	35	
	1582315	256	rs473915	N.D.				synonymous	G	Gln [Q]	3	37	
								contig reference	A	Gln [Q]	3	37	
	1582345	286	rs9993367	N.D.				synonymous	T	Pro [P]	3	47	
								contig reference	G	Pro [P]	3	47	

Predictive Testing for Huntington's: Adverse Psychological Events

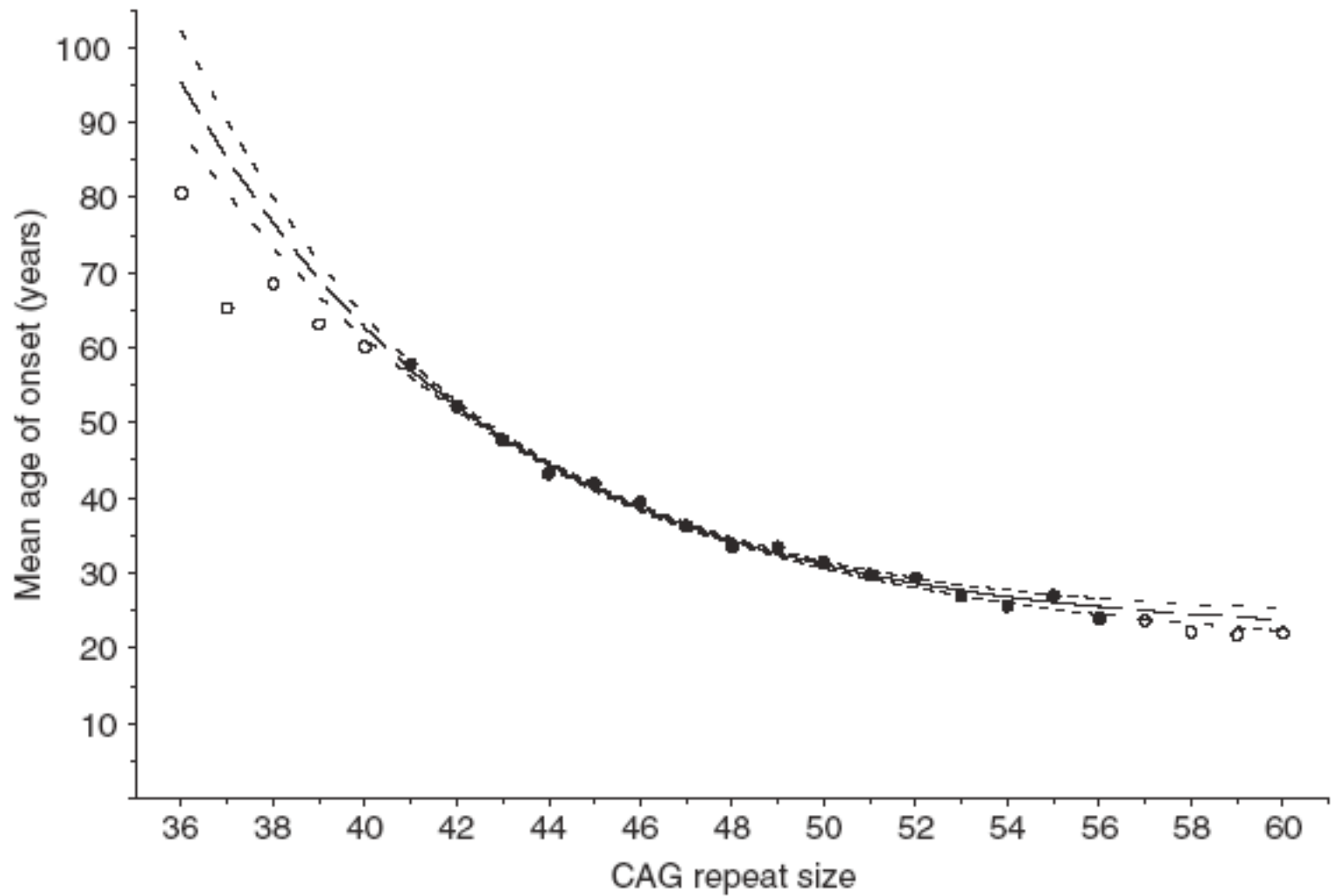
Adverse psychological events occurring in the first year after predictive testing for Huntington's disease. The Canadian Collaborative Study Predictive Testing.

Lawson K, Wiggins S, Green T, Adam S, Bloch M, Hayden MR.

Department of Medical Genetics, University of British Columbia, Vancouver, Canada.

A total of 135 participants in the Canadian predictive testing programme for HD were followed for at least one year in one of four study groups: increased risk ($n = 37$), decreased risk ($n = 58$), uninformative ($n = 17$), or not tested ($n = 23$). Clinical criteria for an adverse event were a suicide attempt or formulation of a suicide attempt plan, psychiatric hospitalisation, depression lasting longer than two months, a marked increase in substance abuse, and the breakdown of important relationships. Quantitative criteria, as measured by changes on the General Severity Index of the Symptom Checklist 90-R and the Beck Depression Inventory, were also used to identify people who had adverse events. Twenty of the 135 participants (14.8%) had an adverse event. There were no significant differences between those with or without an adverse event with respect to age, sex, marital status, education, psychiatric history, general psychiatric distress, or social supports at baseline. However, evidence for depression was associated with an increased frequency of adverse events ($p < 0.04$). The adverse events were similar and seen with equivalent frequency in those receiving an increased risk or decreased risk and persons at risk who did not receive a modification of risk. However, a significant difference was found in the timing of adverse events for the increased and decreased risk groups ($p < 0.0002$). In the increased risk group all of the adverse events occurred within 10 days after results whereas, in the decreased risk group, all of the adverse events occurred six months or later after reviewing test results. These results suggest that people entering into predictive testing with some evidence of clinical depression warrant special vigilance and also suggest that counselling and support should be available for all participants in predictive testing irrespective of the direction of test results.

Age of Onset and Repeat Length



Case Presentations

- Choose an inherited disease of interest
 - Send disease name in email to brutlag@stanford.edu
- Case Presentation
 - Describe disease and classical symptoms and diagnosis
 - Describe classical treatments if any
 - Describe molecular genetics
 - Mendelian, familial, complex, predisposition?
 - Penetrance
 - Does genetics lead to better diagnostics?
 - Does genetics lead to better therapies?

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 - <http://biochem118.stanford.edu/>
- File Repository
 - <http://wherever.stanford.edu/biochem118/>