Diseases and Disease Databases

http://biochem158.stanford.edu/

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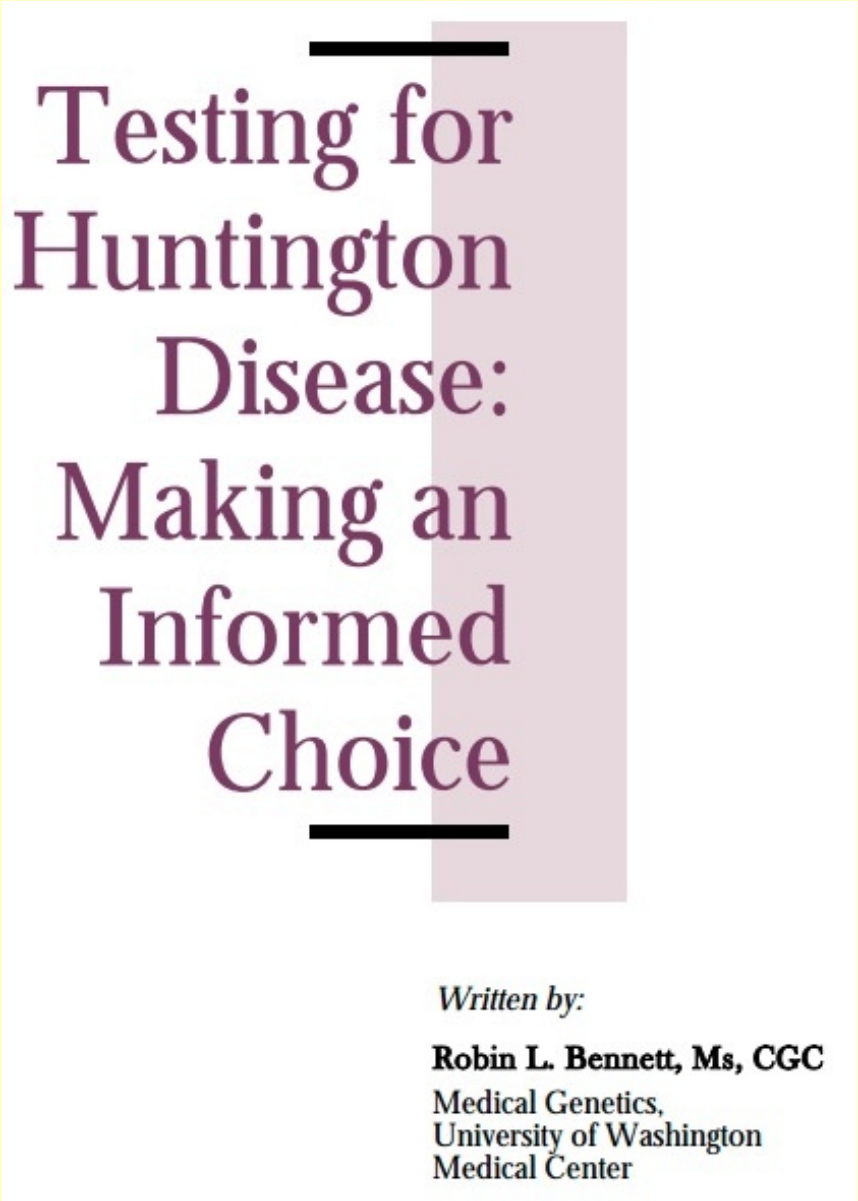
Huntington Disease

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

Scenario 1: The Inheritance

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

Huntington Testing: Making an Informed



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.

Adverse Events of Huntington's Test

- After 1 year, 15% and after 2 years 22% of those with a positive test had an adverse event.
  - Suicide, suicide attempt or suicide plan
  - Psychiatric hospitalization
  - Depression lasting > two months
  - Breakdown of important personal relations
- No incidence of increased substance abuse
- Those with a negative test result often suffered from guilt complex.

Scenario Two

- You are a physician and one of your patients, a 17 year old male has Huntington's in his family
- His grandfather died of the disease at 65 and his older uncle also acquired the disease at 50.
- His father is 40 and is symptom free so far and has specifically told you he does not want the Huntington's test himself.
- The patient comes to you asking for the genetic test to determine if he has the Huntington's gene.
- Would you test the young patient?
- What would you ask your young patient about his reaction to both a positive and a negative diagnosis prior to taking the test?

Mendelian Disease Case Presentation

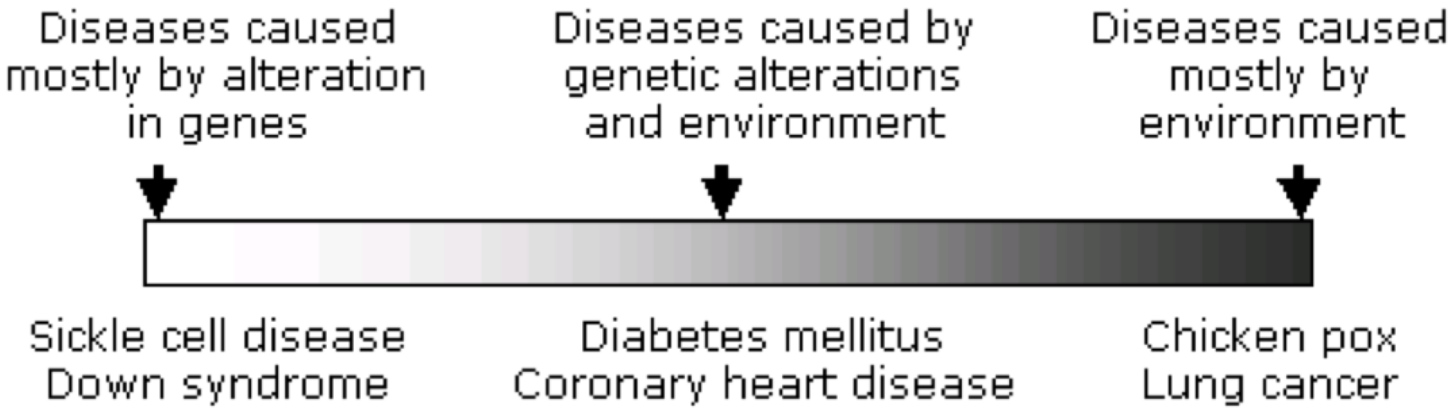
http://biochem158.stanford.edu/case-presentation.html

Please choose a single gene, Mendelian disease from one of the Disease databases (Genes and Disease, Genetics Home Reference, Gene Reviews or Online Inheritance in Man (OMIM) and prepare a written case presentation of the disease (4 pages max) of double spaced text. Figures, Tables and References need not be included in this limit, just the written text

Please Include:

1. A URL pointer to OMIM and/or Gene Reviews entry for your disease
2. A basic description of the disease and its symptoms and prevalence
3. The classical (pre-genetic) differential diagnosis of the disease
4. The classical (pre-genetic) treatment of the disease
5. A description of genetics of the disease including world and ethnic distribution of the disease gene
6. Any novel diagnostics that have resulted from knowing the genetics
7. Any novel understanding of the disease that has lead to novel therapy based on genetic knowledge.

Genetic Penetrance



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

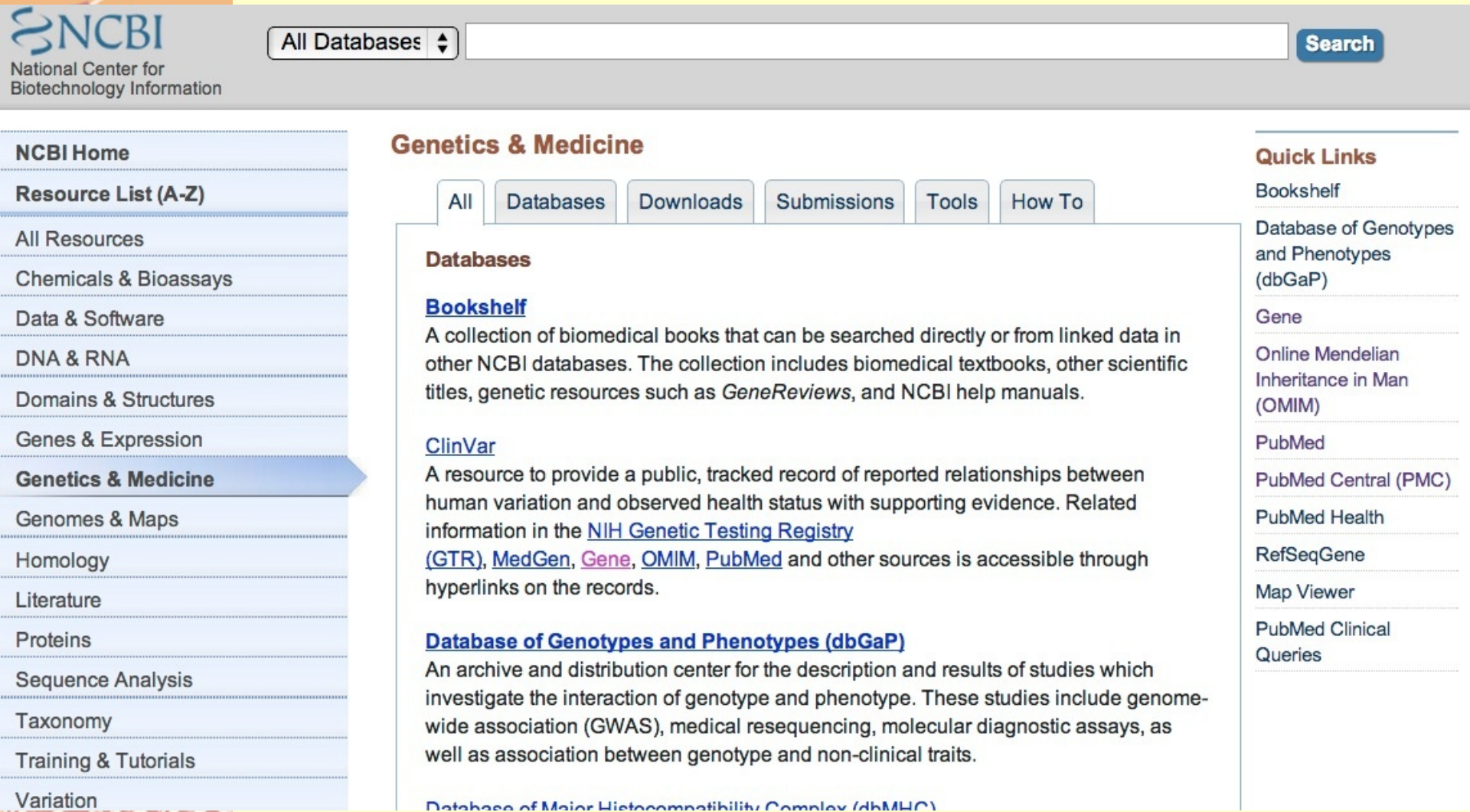
Diseases in the middle of the spectrum — including most common diseases — are **multifactorial**, and result from the interaction or additive effect of genetic and non-genetic factors.

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/



NCBI: Genetics and Medicine

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

Gene

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

GeneReviews

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

Genes and Disease

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

Genetic Testing Registry (GTR)

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

NCBI: Genetics and Medicine

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

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

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

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

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

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

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

Genes and Disease

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

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Bookshelf
U.S. National Library of Medicine
National Institutes of Health

Search This Book Limits Help

Search Clear

Bookshelf ID: NBK22183

Genes and Disease

National Center for Biotechnology Information (US)

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

Copyright notice.

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

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

Contents

Introduction to Genes and Disease

Recent activity

Turn Off Clear

- Genes and Disease — Bookshelf
- Ensemble-based methods for describing protein dynamics. — PubMed
- froda (10) — PubMed
- wells j[au] (2079) — PubMed
- wells j[au] AND froda (0) — PubMed

See more...

Genes and Disease Table of Contents

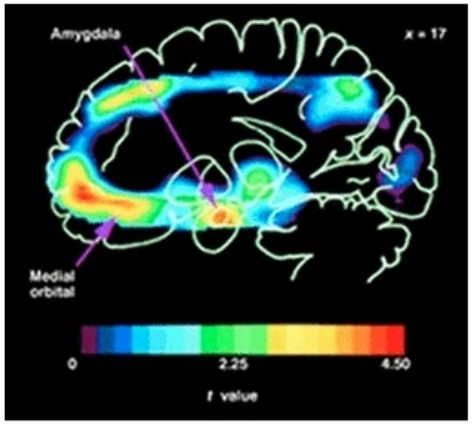
http://www.ncbi.nlm.nih.gov/books/NBK22183/

Contents

Genes and Disease: The Nervous System

http://www.ncbi.nlm.nih.gov/books/NBK22197/

The Nervous System



One of the major areas in which molecular genetics will play an important role in the future is in complex disorders like schizophrenia and depression. The figure shows areas of increased bloodflow (red hotspots) in the left amygdala and the medial orbital cortex of a person with familial, major depressive order. The molecular basis for this observation, and others like it, remain a challenge for the future. [Reproduced from Andreasen, NC (1997) Science 275, 1586-1593, with permission.]

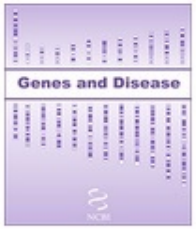
The brain and nervous system form an intricate network of electrical signals that are responsible for coordinating muscles, the senses, speech, memories, thought and emotion.

Several diseases that directly affect the nervous system have a genetic component: some are due to a mutation in a single gene, others are proving to have a more complex mode of inheritance. As our understanding of the pathogenesis of neurodegenerative disorders deepens, common themes begin to emerge: Alzheimer brain plaques and the inclusion bodies found in Parkinson disease contain at least one common component, while Huntington disease, fragile X syndrome and spinocerebellar atrophy are all 'dynamic mutation' diseases in which there is an expansion of a DNA repeat sequence. Apoptosis is emerging as one of the molecular mechanisms invoked in several neurodegenerative diseases, as are other, specific, intracellular signaling events. The biosynthesis of myelin and the regulation of cholesterol traffic also figure in Charcot-Marie-Tooth and Neimann-Pick disease, respectively.

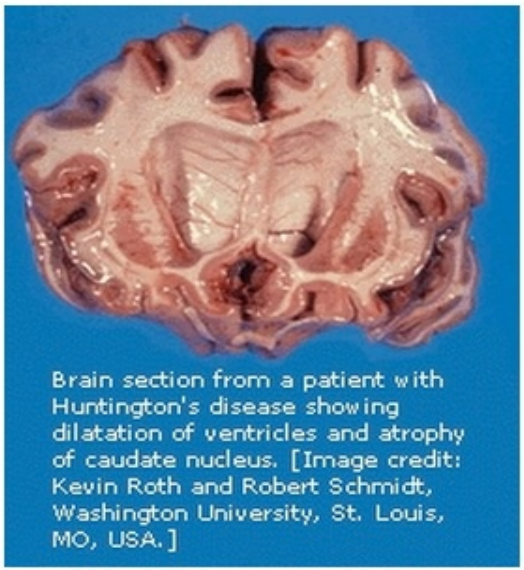
NCBI

Diseases

- Adrenoleukodystrophy
- Alzheimer disease
- Amyotrophic lateral sclerosis
- Angelman syndrome
- Ataxia telangiectasia
- Charcot-Marie-Tooth syndrome
- Cockayne syndrome
- Deafness
- Duchenne muscular dystrophy
- Epilepsy
- Essential tremor
- Fragile X syndrome
- Friedreich's ataxia
- Gaucher disease
- Huntington disease
- Lesch-Nyhan syndrome
- Maple syrup urine disease
- Menkes syndrome
- Myotonic dystrophy
- Narcolepsy
- Neurofibromatosis
- Niemann-Pick disease
- Parkinson disease
- Phenylketonuria



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.

Views

Print View

Cite this Page

PDF version of this page (265K)

Gene sequence

Genome view see gene locations

Entrez Gene collection of gene-related information

BLink related sequences in different organisms

The literature

Research articles online full text

Books online books section

OMIM catalog of human genes and disorders

GeneReviews a medical genetics resource

Websites

Huntington Disease Society of America information for patients and the public

Related information

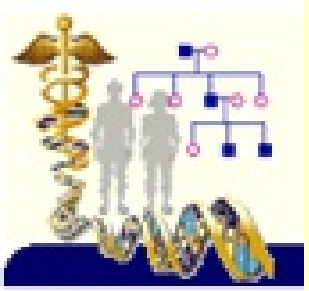
Gene

Recent Activity

Turn Off Clear

Genetics Home Reference

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



About Site Map Contact Us

Search

Genetics Home Reference

Your Guide to Understanding Genetic Conditions

A service of the U.S. National Library of Medicine®

What's New

- glycogen storage disease type 0
- cytogenetically normal acute myeloid leukemia
- congenital leptin deficiency
- More...

Newborn Screening

Detecting genetic disorders for early treatment

In the Spotlight

- Learning Activities
- What is direct-to-consumer genetic testing?
- GHR results now available from MedlinePlus Connect

Genetic Disorders A to Z
and related genes and chromosomes

Conditions
The genetics of more than 900 health conditions, diseases, and syndromes.

Genes
More than 1,100 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: January 6, 2014

Huntington Disease in Genetics Home Reference

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

Genetic Conditions >

Huntington disease

Print version

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

MedlinePlus
Health information

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

Additional NIH Resources
National Institutes of Health

Educational resources
Information pages

Patient support
For patients and families

Gene Reviews
Clinical summary

Gene Tests
DNA test labs

ClinicalTrials.gov
Research studies

PubMed
Recent literature

Online Books
Medical and science texts

OMIM
Genetic disorder catalog

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

Reviewed October 2008

What is Huntington disease?

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

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

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

How common is Huntington disease?

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

MedlinePlus

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

Huntington's in Medline Plus

MedlinePlus
Trusted Health Information for You

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

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Search MedlinePlus: Huntingtons GO

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

Refine by Type

- All Results (129)
- Health Topics (3)
- External Health Links (91)
- Drugs and Supplements (4)
- Medical Encyclopedia (13)
- Videos and Tutorials
- News (2)
- MedlinePlus Magazine (6)
- Other Resources (10)
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Refine by Keyword

All Results (129) remix

- Genetic (35)
- Brain (12)
- Research (12)
- Harvard School of Public Health (15)
- Chorea (8)
- America (8)
- Dementia (11)
- Multiple | Parkinson (7)
- Huntington Beach, CA (6)
- Nerve Diseases (5)

more

Huntington's Disease Search Help

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

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

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

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

Huntington Disease Gene Review

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

Bookshelf ID: NBK1305 PMID: 20301482

Huntington Disease

Huntington Chorea

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

Summary

Go to: Top▲

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

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

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

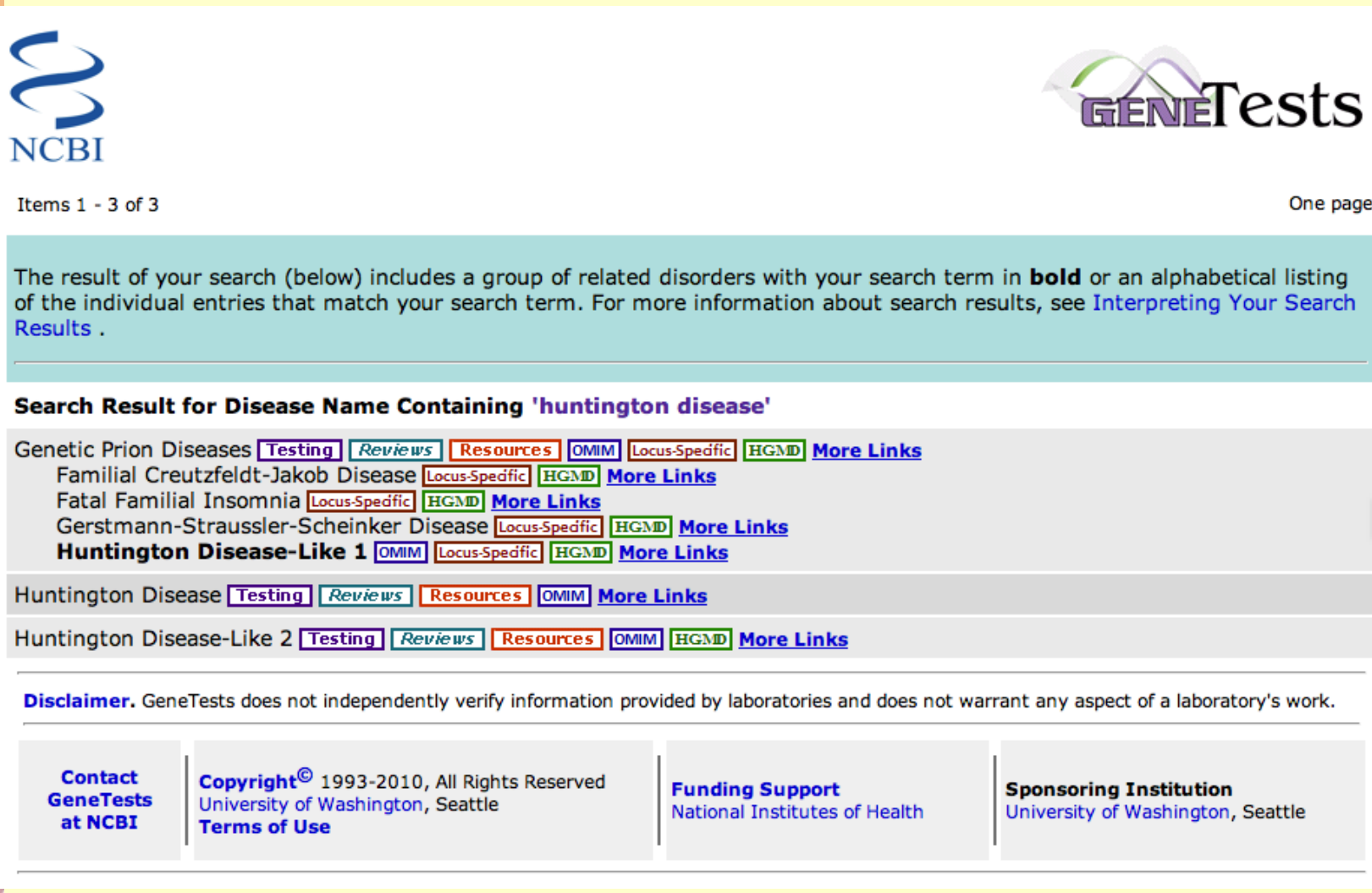
GeneReviews [Internet].
Pagon RA, Bird TC, Dolan CR, et al., editors.
Seattle (WA): University of Washington, Seattle; 1993-.
[Table of Contents Page]

GeneReviews Links

GeneTests Home Page
GeneReviews Advanced Search
About GeneTests

GeneTests & GeneReviews for Huntingtons

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



NCBI

GeneTests

Items 1 - 3 of 3 — One page

The result of your search (below) includes a group of related disorders with your search term in **bold** or an alphabetical listing of the individual entries that match your search term. For more information about search results, see Interpreting Your Search Results .

Search Result for Disease Name Containing 'huntington disease'

Genetic Prion Diseases [Testing] [Reviews] [Resources] [OMIM] [Locus-Specific] [HGMD] More Links
- Familial Creutzfeldt-Jakob Disease [Locus-Specific] [HGMD] More Links
- Fatal Familial Insomnia [Locus-Specific] [HGMD] More Links
- Gerstmann-Straussler-Scheinker Disease [Locus-Specific] [HGMD] More Links
- **Huntington Disease-Like 1** [OMIM] [Locus-Specific] [HGMD] More Links

Huntington Disease [Testing] [Reviews] [Resources] [OMIM] More Links

Huntington Disease-Like 2 [Testing] [Reviews] [Resources] [OMIM] [HGMD] More Links

Disclaimer. GeneTests does not independently verify information provided by laboratories and does not warrant any aspect of a laboratory's work.

Contact GeneTests at NCBI

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Funding Support
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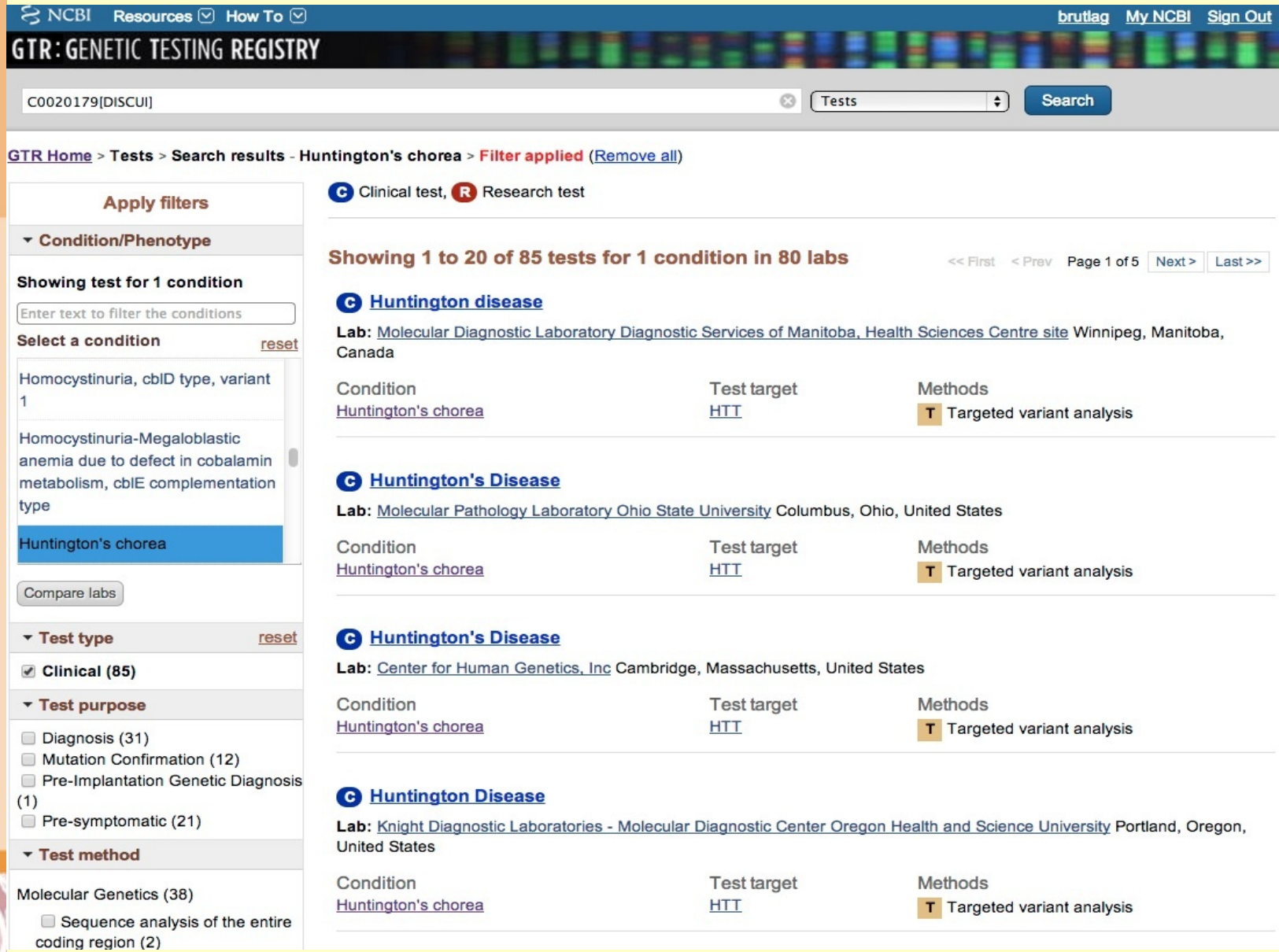
Genetic Testing Registry for Huntington

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



OMIM Home Page

http://omim.org/

Mirror sites: us-east.omim.org, europe.omim.org

OMIM®

Online Mendelian Inheritance in Man®

An Online Catalog of Human Genes and Genetic Disorders

Updated 8 January 2014

Huntington Disease | Search | Sample Searches | OMIM Tutorial

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map

McKusick-Nathans Institute of Genetic Medicine

JOHNS HOPKINS MEDICINE

National Human Genome Research Institute

NOTE: OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. While the OMIM database is open to the public, users seeking information about a personal medical or genetic condition are urged to consult with a qualified physician for diagnosis and for answers to personal questions.

OMIM Coverage

http://www.ncbi.nlm.nih.gov/Omim/mimstats.html

January 5, 2015

OMIM Entry Statistics

Number of Entries in OMIM (Updated January 5th, 2015) :

Prefix	Autosomal	X Linked	Y Linked	Mitochondrial	Totals
* Gene description	14,027	689	48	35	14,799
+ Gene and phenotype, combined	84	2	0	2	88
# Phenotype description, molecular basis known	3,991	287	4	28	4,310
% Phenotype description or locus, molecular basis unknown	1,540	133	5	0	1,678
Other, mainly phenotypes with suspected mendelian basis	1,734	113	2	0	1,849
Totals	21,376	1,224	59	65	22,724

> 67% Genes

> 33% Phenotypes

Huntington Disease Search in OMIM

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

NCBI OMIM®

Home | About | Statistics | Downloads/API | Help | External Links | Terms of Use | Contact Us | MIMmatch | Select Language

Huntington Disease | Search | Sort by: Relevance Date updated

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map **Toggle:** search terms highlighted

Search History: View, Clear

Retrieve corresponding: gene map | clinical synopses

Search: 'Huntington Disease'

Results: 1 - 10 of 6,850 | Show top 100 | 1 2 3 4 5 6 7 8 9 10 Next Last

1 : # 143100. HUNTINGTON DISEASE; HD — ICD+, Links
Cytogenetic location: 4p16.3
Matching terms: disease, huntington

2 : * 613004. HUNTINGTIN; HTT — Gene Tests, Links
Cytogenetic location: 4p16.3 , Genomic coordinates (GRCh37): 4:3,076,407 - 3,245,686
Matching terms: disease, huntington

3 : % 604802. HUNTINGTON DISEASE-LIKE 3; HDL3 — Links
Cytogenetic location: 4p15.3 , Genomic coordinates (GRCh37): 4:11,300,000 - 21,300,000
Matching terms: disease, huntington

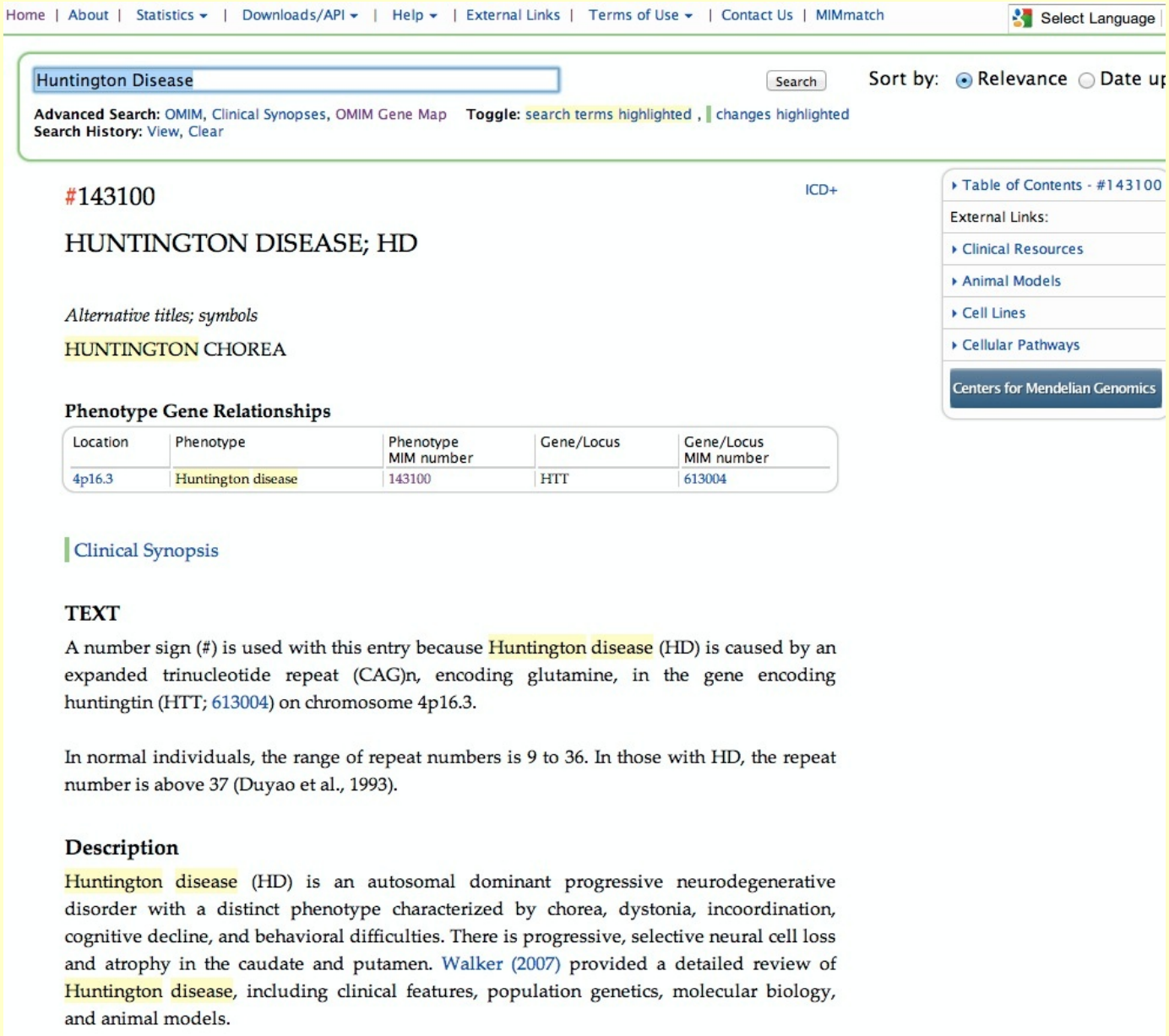
4 : # 606438. HUNTINGTON DISEASE-LIKE 2; HDL2 — Links
Cytogenetic location: 16q24.2
Matching terms: disease, huntington

5 : # 603218. HUNTINGTON DISEASE-LIKE 1; HDL1 — Links
Cytogenetic location: 20p13
Matching terms: disease, huntington

6 : # 607136. SPINOCEREBELLAR ATAXIA 17; SCA17 — Links
Cytogenetic location: 6q27
Matching terms: disease, huntington

Huntington Disease Entry in OMIM

http://www.ncbi.nlm.nih.gov/omim/143100



Home | About | Statistics | Downloads/API | Help | External Links | Terms of Use | Contact Us | MIMmatch | Select Language

Huntington Disease | Search | Sort by: Relevance | Date up

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map **Toggle:** search terms highlighted , changes highlighted
Search History: View, Clear

#143100 ICD+

HUNTINGTON DISEASE; HD

Alternative titles; symbols

HUNTINGTON CHOREA

Phenotype Gene Relationships

Location	Phenotype	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
4p16.3	Huntington disease	143100	HTT	613004

Clinical Synopsis

TEXT

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

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.

Table of Contents - #143100
External Links:
Clinical Resources
Animal Models
Cell Lines
Cellular Pathways
Centers for Mendelian Genomics

Huntingtin Protein Entry in OMIM

http://www.ncbi.nlm.nih.gov/omim/143100

- Table of Contents - *613004
- External Links:
- Genome
- DNA
- Protein
- Gene Info
- Clinical Resources
- Variation
- Animal Models
- Cellular Pathways

*613004

HUNTINGTIN; HTT

Alternative titles; symbols

IT15
HD GENE

HGNC Approved Gene Symbol: HTT

Cytogenetic location: 4p16.3 ***Genomic coordinates (GRCh37): 4:3,076,407 - 3,245,686*** (from NCBI)

Gene Phenotype Relationships

Location	Phenotype	Phenotype MIM number
4p16.3	Huntington disease	143100

TEXT

Description

The HTT gene encodes huntingtin, a ubiquitously expressed nuclear protein that binds to a number of transcription factors to regulate transcription. Abnormal expansion of a polyglutamine tract in the N terminus of huntingtin causes Huntington disease (143100), a devastating autosomal dominant neurodegenerative disease characterized by motor, psychiatric, and cognitive dysfunction (summary by Futter et al., 2009).

Entrez Gene for Huntington

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

1: HTT huntingtin [*Homo sapiens*]

GeneID: 3064 updated 04-Jan-2009

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:143100
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]

Entrez Gene Home

Table Of Contents

- Summary
- Genomic regions, transcripts...
- Genomic context
- Bibliography
- Interactions
- General gene information
- General protein information
- Reference Sequences
- Related Sequences
- Additional Links

Links Explain

- CCDS
- Genome
- GEO Profiles
- HomoloGene
- Map Viewer
- Nucleotide
- OMIM
- BioAssay
- Full text in PMC
- Probe
- Protein
- PubMed
- PubMed (OMIM)
- PubMed (GeneRIF)
- SNP
- SNP: Genotype
- ✓ SNP: GeneView
- Taxonomy
- UniSTS
- AceView
- Ensembl
- Evidence Viewer
- GeneTests for MIM: 143100
- ✓ HGMD
- HGNC
- HPRD
- HuGE Navigator
- Huntington.html

Huntington Disease Gene

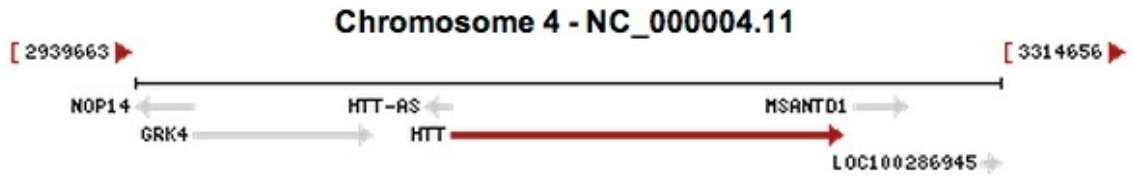
http://www.ncbi.nlm.nih.gov/gene/3064

Genomic context

Location: 4p16.3

Sequence: Chromosome: 4; NC_000004.11 (3076408..3245687)

See HTT in Epigenomics, MapViewer

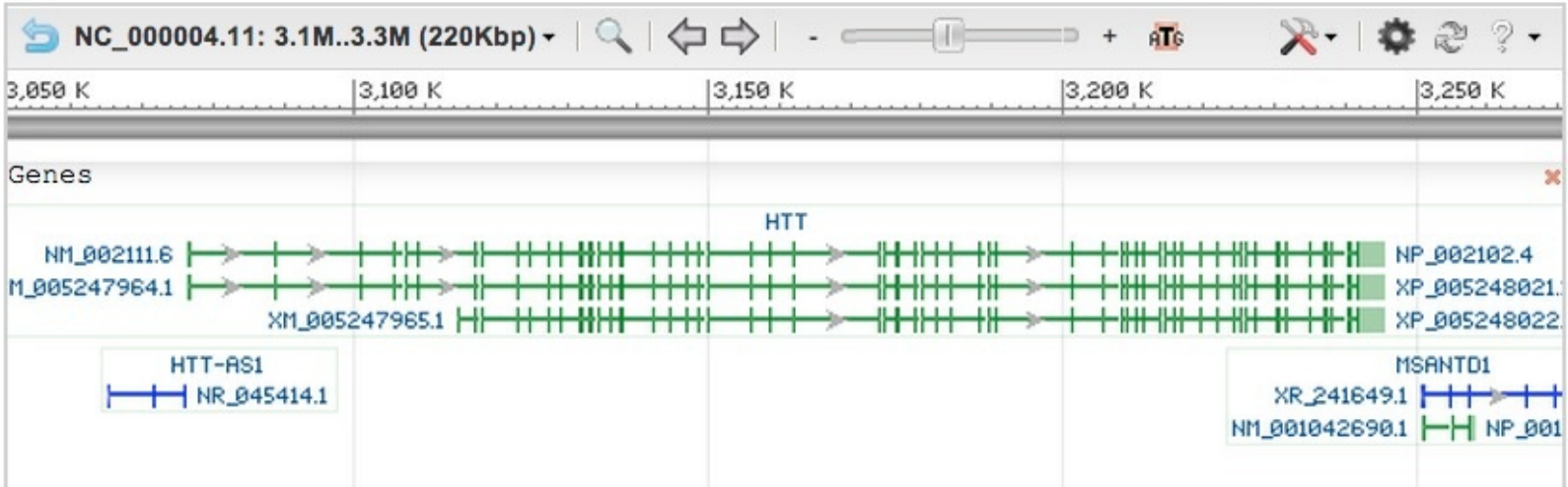


Genomic regions, transcripts, and products

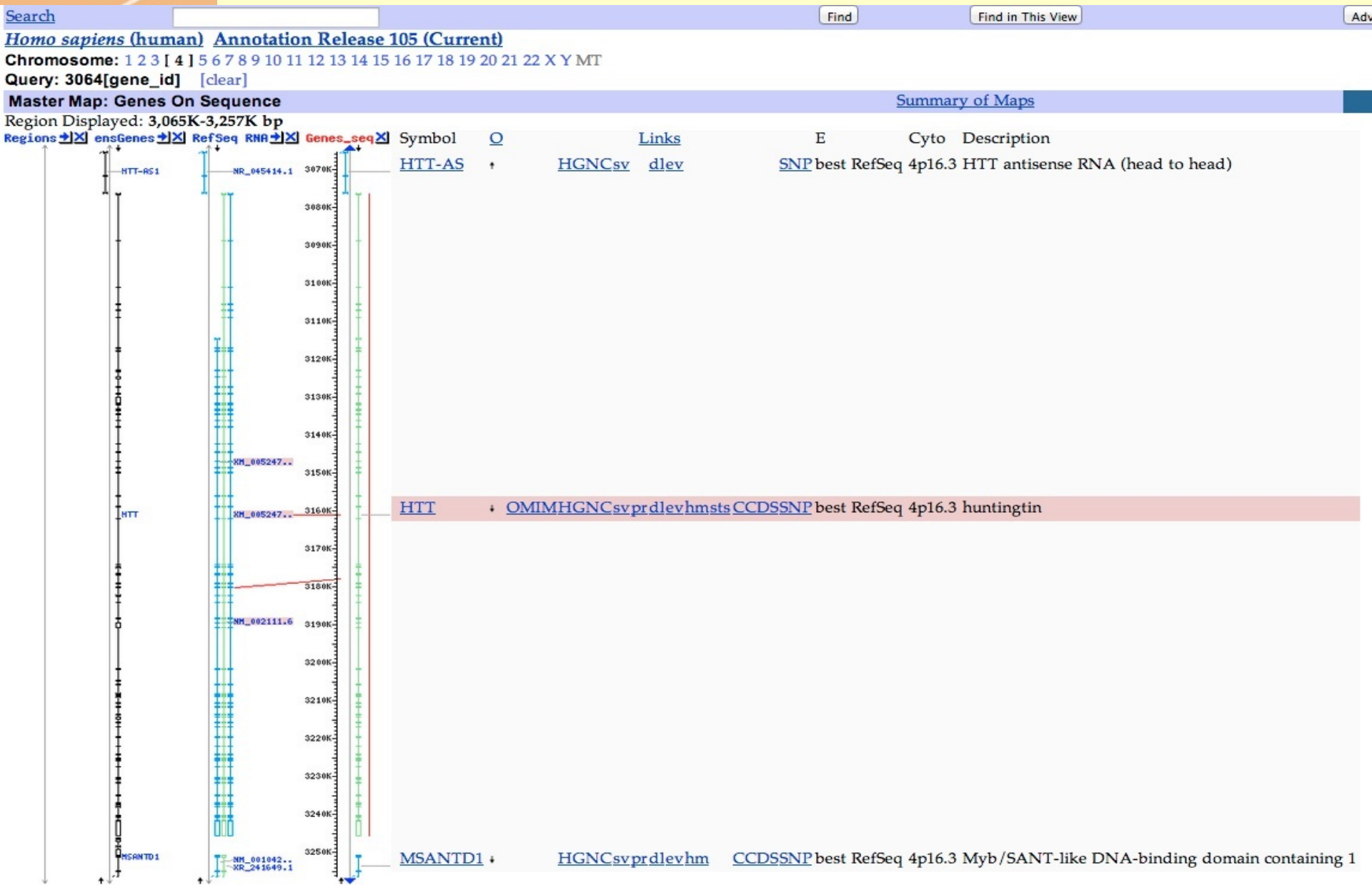
Genomic Sequence NC_000004 chromosome 4 reference GRCh37.p13 Primary Assembly

Go to reference sequence details

Go to nucleotide Graphics FASTA GenBank



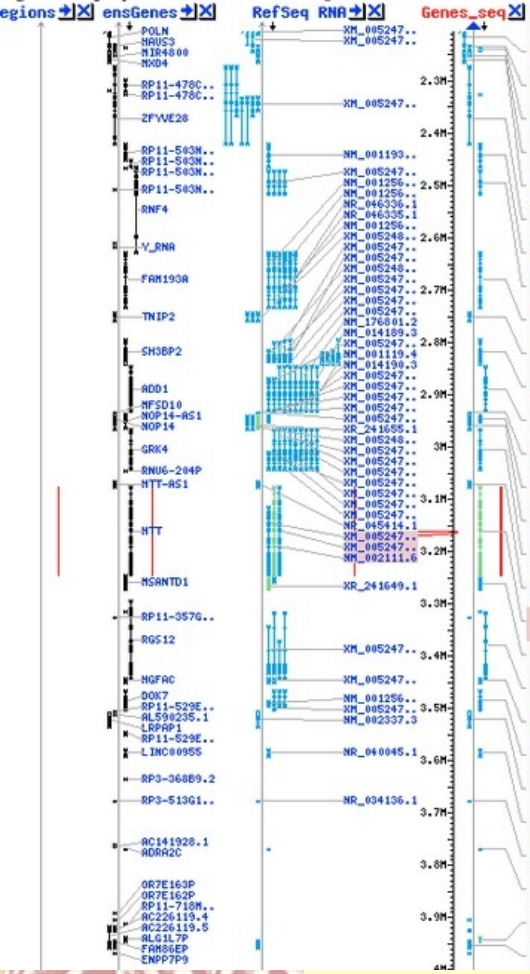
MapViewer for Huntington



MapViewer for Huntington

Master Map: Genes On Sequence — Summary of Maps — Download / View

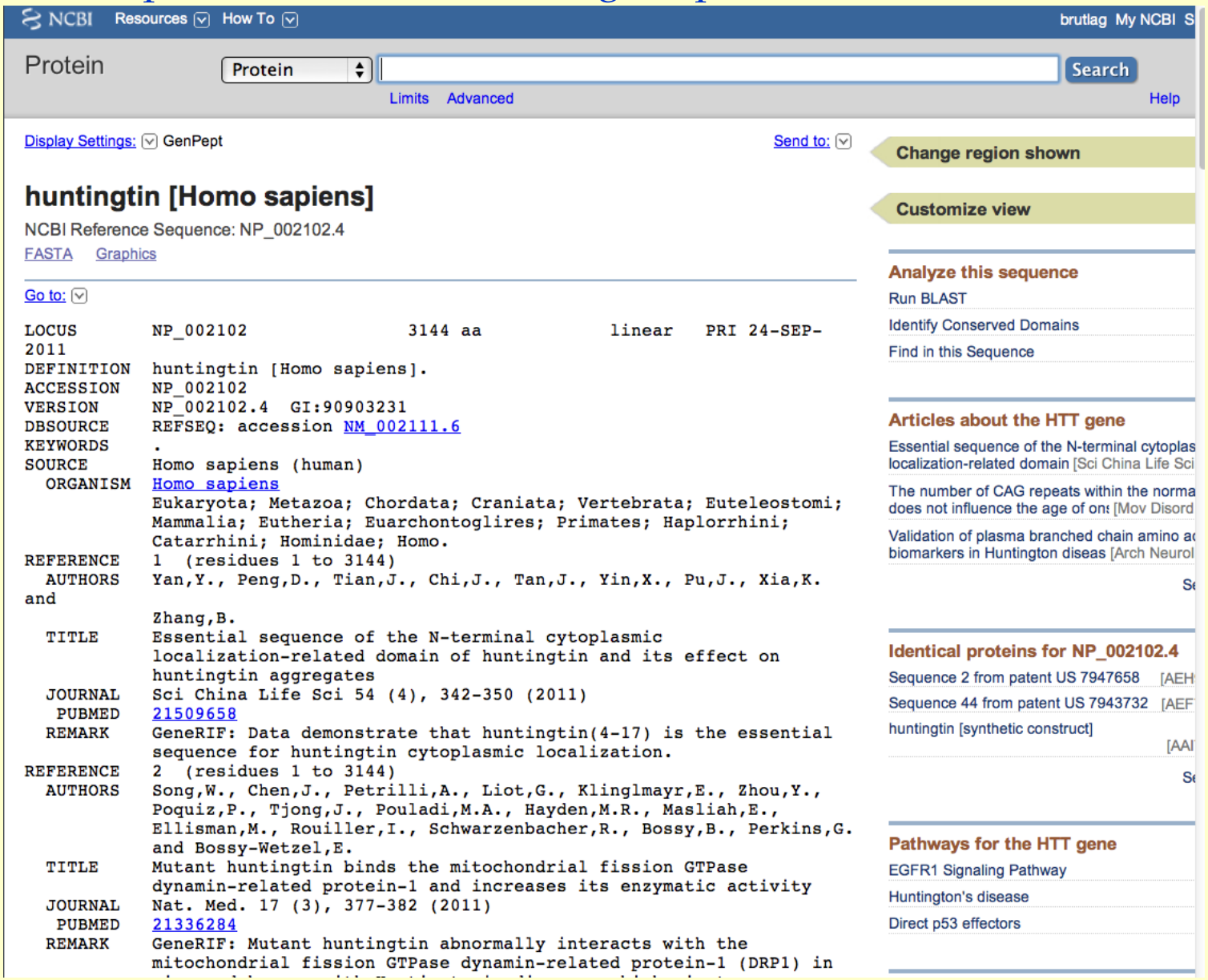
Region Displayed: **2,210K-4,120K bp**



Symbol	O	Links	E	Cyto	Description
POLN	+	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	polymerase (DNA directed) nu
HAUS3	+	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	HAUS augmin-like complex, subunit 3
COX6B1P5	-	HGNC sv dlev sts	best RefSeq	4p16.3	cytochrome c oxidase subunit VIb polypeptide 1 (ubiquitous) pseudog
MIR4800	+	HGNC sv dlev SNP	best RefSeq		microRNA 4800
MXD4	+	HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	MAX dimerization protein 4
ZFYVE28	+	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	zinc finger, FYVE domain containing 28
RP11-503N18.1	-	sv pr dlev hm SNP	best RefSeq	4p16.3	uncharacterized LOC402160
RNF4	-	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	ring finger protein 4
FAM193A	-	HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	family with sequence similarity 193, member A
TNIP2	+	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	TNFAIP3 interacting protein 2
SH3BP2	-	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	SH3-domain binding protein 2
ADD1	-	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	adducin 1 (alpha)
MFSD10	+	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	major facilitator superfamily domain containing 10
NOP14-AS1	-	HGNC sv pr dlev sts SNP	best RefSeq	4p16.3	NOP14 antisense RNA 1
NOP14	+	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	NOP14 nucleolar protein
GRK4	-	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	G protein-coupled receptor kinase 4
HTT-AS	+	HGNC sv dlev SNP	best RefSeq	4p16.3	HTT antisense RNA (head to head)
HTT	-	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	huntingtin
MSANTD1	-	HGNC sv pr dlev hm CCDS SNP	best RefSeq	4p16.3	Myb/SANT-like DNA-binding domain containing 1
RGS12	+	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	regulator of G-protein signaling 12
HGFAC	+	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16	HGF activator
DOK7	+	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	docking protein 7
LRPAP1	+	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16.3	low density lipoprotein receptor-related protein associated protein 1
LINC00955	-	HGNC sv pr dlev sts SNP	best RefSeq	4p16.3	long intergenic non-protein coding RNA 955
RP3-513G18.2	+	sv dlev sts SNP	best RefSeq		uncharacterized LOC100133461
ADRA2C	-	OMIM HGNC sv pr dlev hm sts CCDS SNP	best RefSeq	4p16	adrenoceptor alpha 2C
ALG1L7P	-	HGNC sv dlev sts	best RefSeq	4p16.3	asparagine-linked glycosylation 1-like 7, pseudogene
FAM86EP	-	HGNC sv dlev sts SNP	best RefSeq	4p16.3	family with sequence similarity 86, member A pseudogene

Huntingtin Protein

http://www.ncbi.nlm.nih.gov/protein/NP_002102.4



NCBI Resources How To brutlag My NCBI S

Protein | Protein | Search | Limits Advanced | Help

Display Settings: GenPept | Send to:

Change region shown

Customize view

huntingtin [Homo sapiens]

NCBI Reference Sequence: NP_002102.4

FASTA Graphics

Go to:

```
LOCUS       NP_002102               3144 aa            linear   PRI 24-SEP-
2011
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ACCESSION   NP_002102
VERSION     NP_002102.4  GI:90903231
DBSOURCE    REFSEQ: accession NM_002111.6
KEYWORDS    .
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
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            Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
            Catarrhini; Hominidae; Homo.
REFERENCE   1  (residues 1 to 3144)
  AUTHORS   Yan,Y., Peng,D., Tian,J., Chi,J., Tan,J., Yin,X., Pu,J., Xia,K.
and
            Zhang,B.
  TITLE     Essential sequence of the N-terminal cytoplasmic
            localization-related domain of huntingtin and its effect on
            huntingtin aggregates
  JOURNAL   Sci China Life Sci 54 (4), 342-350 (2011)
   PUBMED   21509658
  REMARK    GeneRIF: Data demonstrate that huntingtin(4-17) is the essential
            sequence for huntingtin cytoplasmic localization.
REFERENCE   2  (residues 1 to 3144)
  AUTHORS   Song,W., Chen,J., Petrilli,A., Liot,G., Klinglmayr,E., Zhou,Y.,
            Poquiz,P., Tjong,J., Pouladi,M.A., Hayden,M.R., Masliah,E.,
            Ellisman,M., Rouiller,I., Schwarzenbacher,R., Bossy,B., Perkins,G.
and Bossy-Wetzel,E.
  TITLE     Mutant huntingtin binds the mitochondrial fission GTPase
            dynamin-related protein-1 and increases its enzymatic activity
  JOURNAL   Nat. Med. 17 (3), 377-382 (2011)
   PUBMED   21336284
  REMARK    GeneRIF: Mutant huntingtin abnormally interacts with the
            mitochondrial fission GTPase dynamin-related protein-1 (DRP1) in
```

Analyze this sequence

Run BLAST

Identify Conserved Domains

Find in this Sequence

Articles about the HTT gene

Essential sequence of the N-terminal cytoplas
localization-related domain [Sci China Life Sci

The number of CAG repeats within the norma
does not influence the age of ons [Mov Disord

Validation of plasma branched chain amino ac
biomarkers in Huntington diseas [Arch Neurol

Se

Identical proteins for NP_002102.4

Sequence 2 from patent US 7947658 [AEH

Sequence 44 from patent US 7943732 [AEF

huntingtin [synthetic construct]
[AAI

Se

Pathways for the HTT gene

EGFR1 Signaling Pathway

Huntington's disease

Direct p53 effectors

Huntingtin Protein

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

Display Settings: FASTA

Send to:

RecName: Full=Huntingtin; AltName: Full=Huntington disease protein; Short=HD protein

UniProtKB/Swiss-Prot: P42858.2

GenPept Graphics

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disease protein; Short=HD protein
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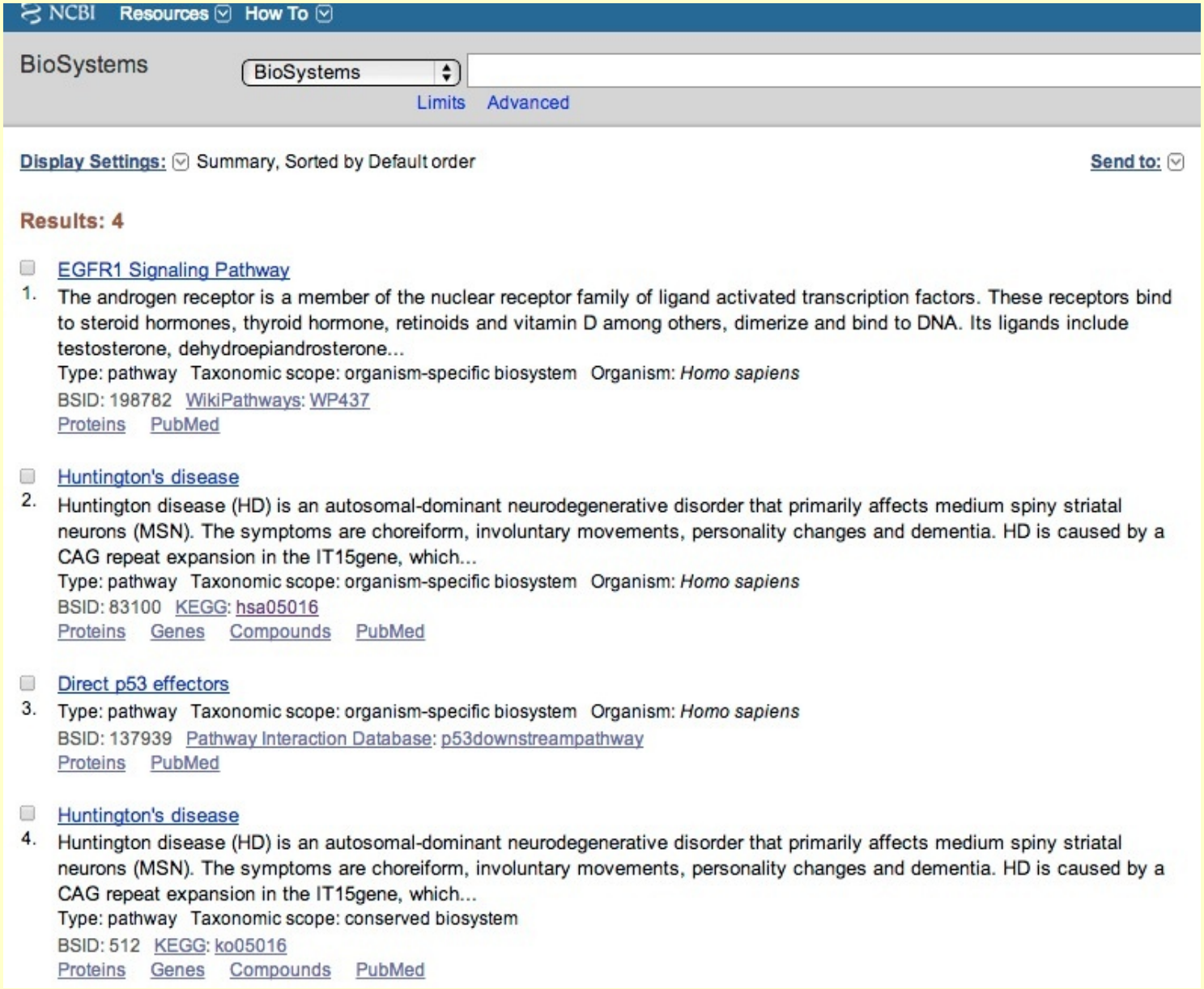
Huntington Biosystems

http://www.ncbi.nlm.nih.gov/biosystems?LinkName=gene_biosystems&from_uid=3064



Huntington Disease Biosystem

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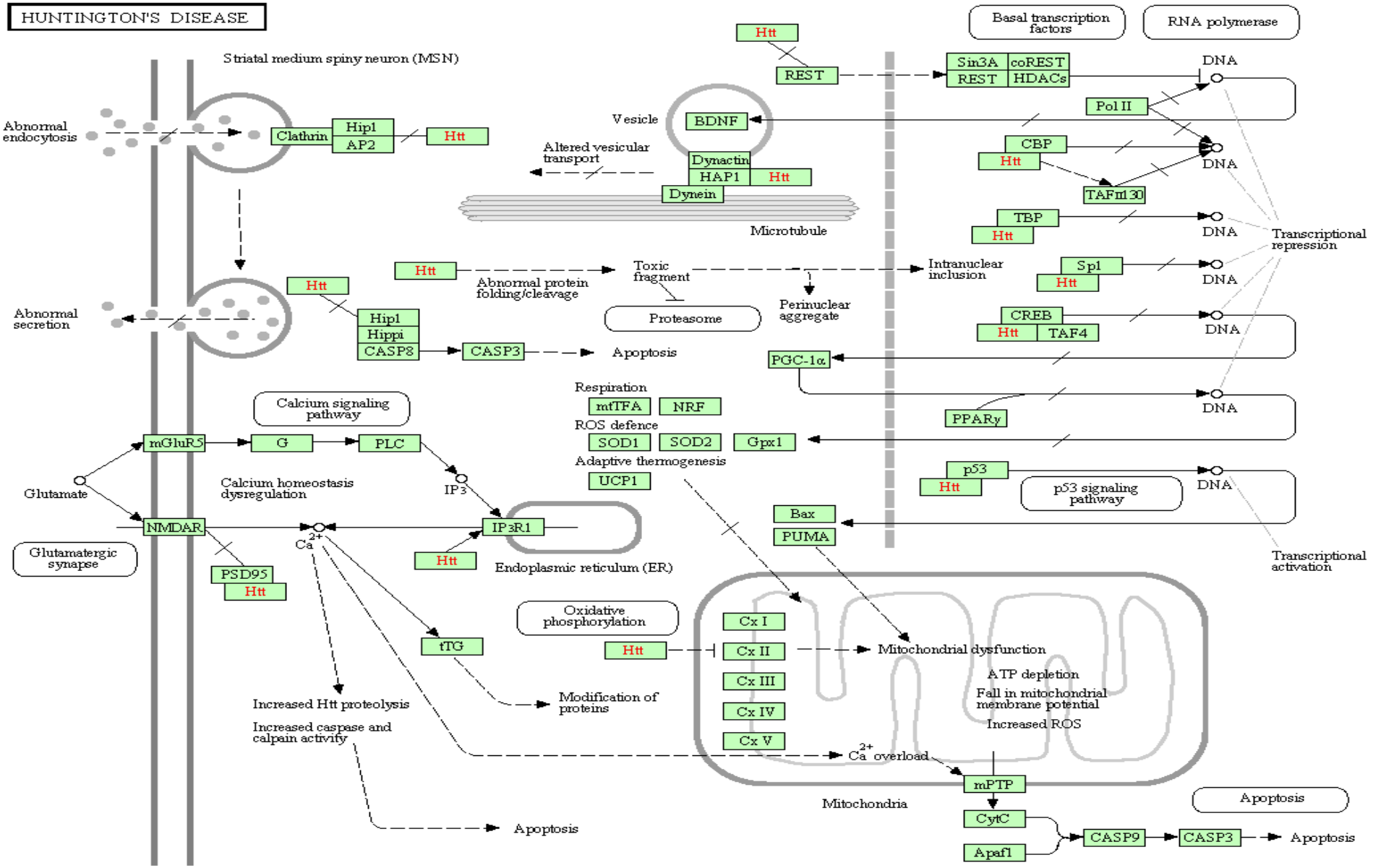
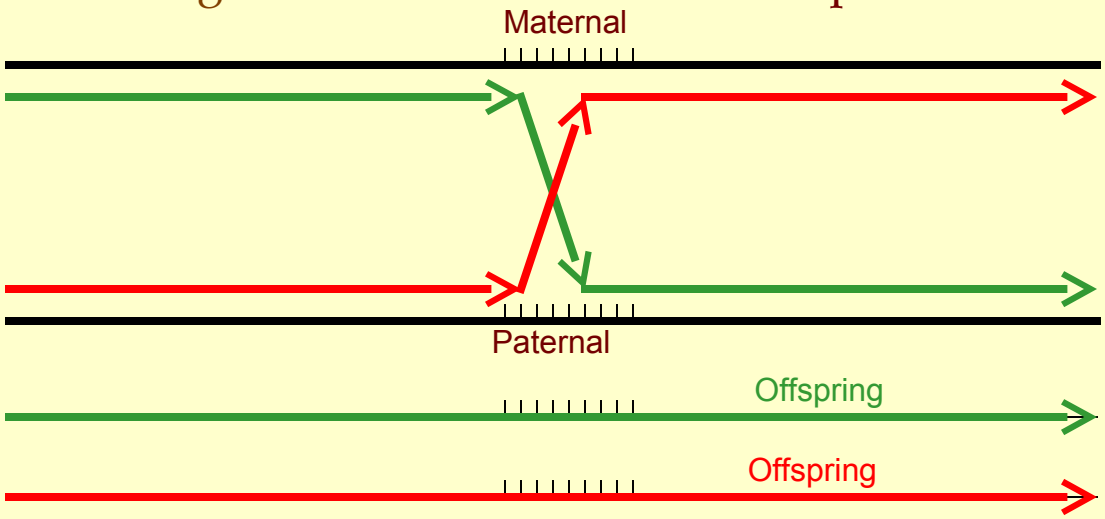
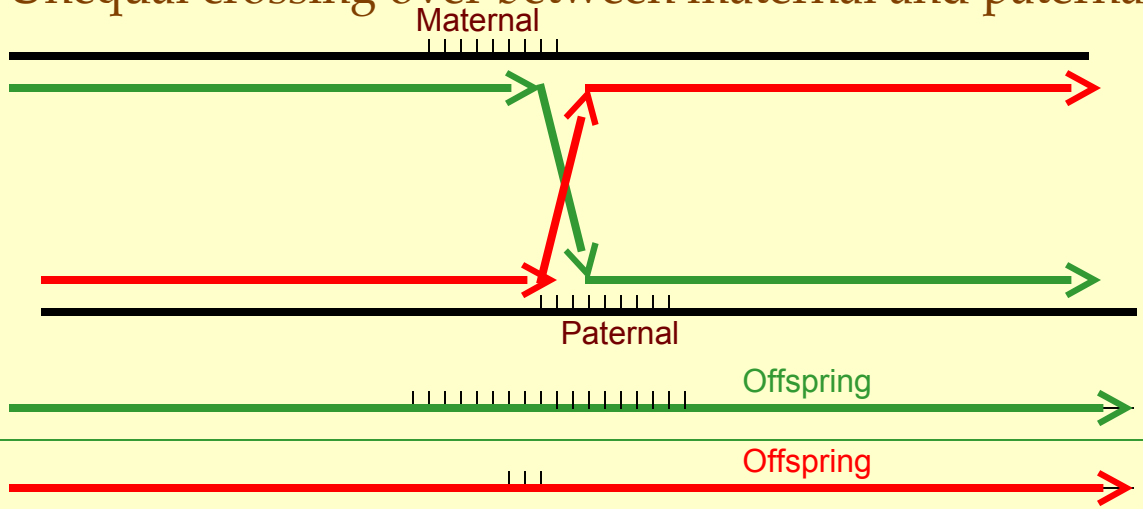


Huntington Disease can Arise from Unequal Crossing Over During Meiosis

- Crossing over between maternal and paternal chromosomes

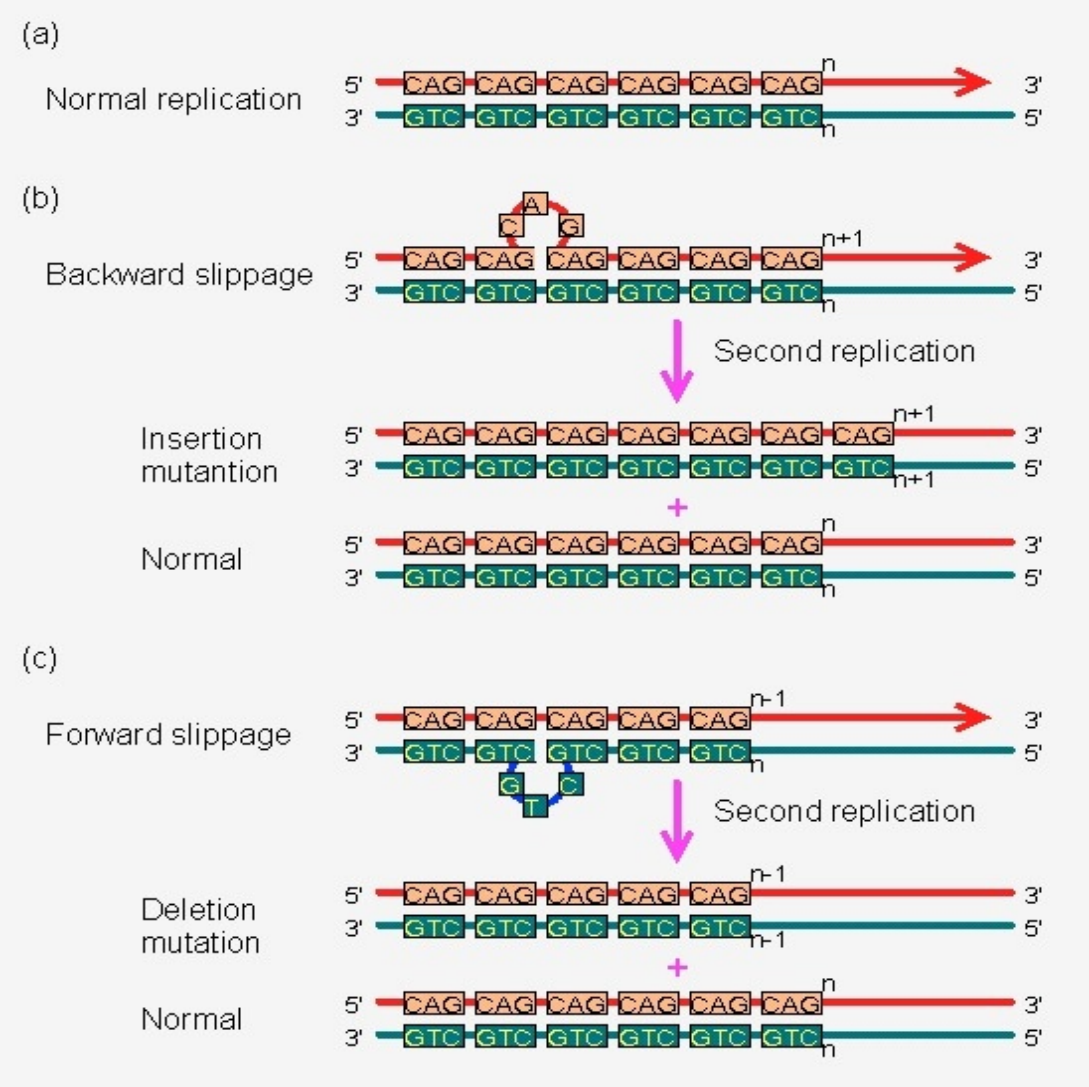


- Unequal crossing over between maternal and paternal chromosomes



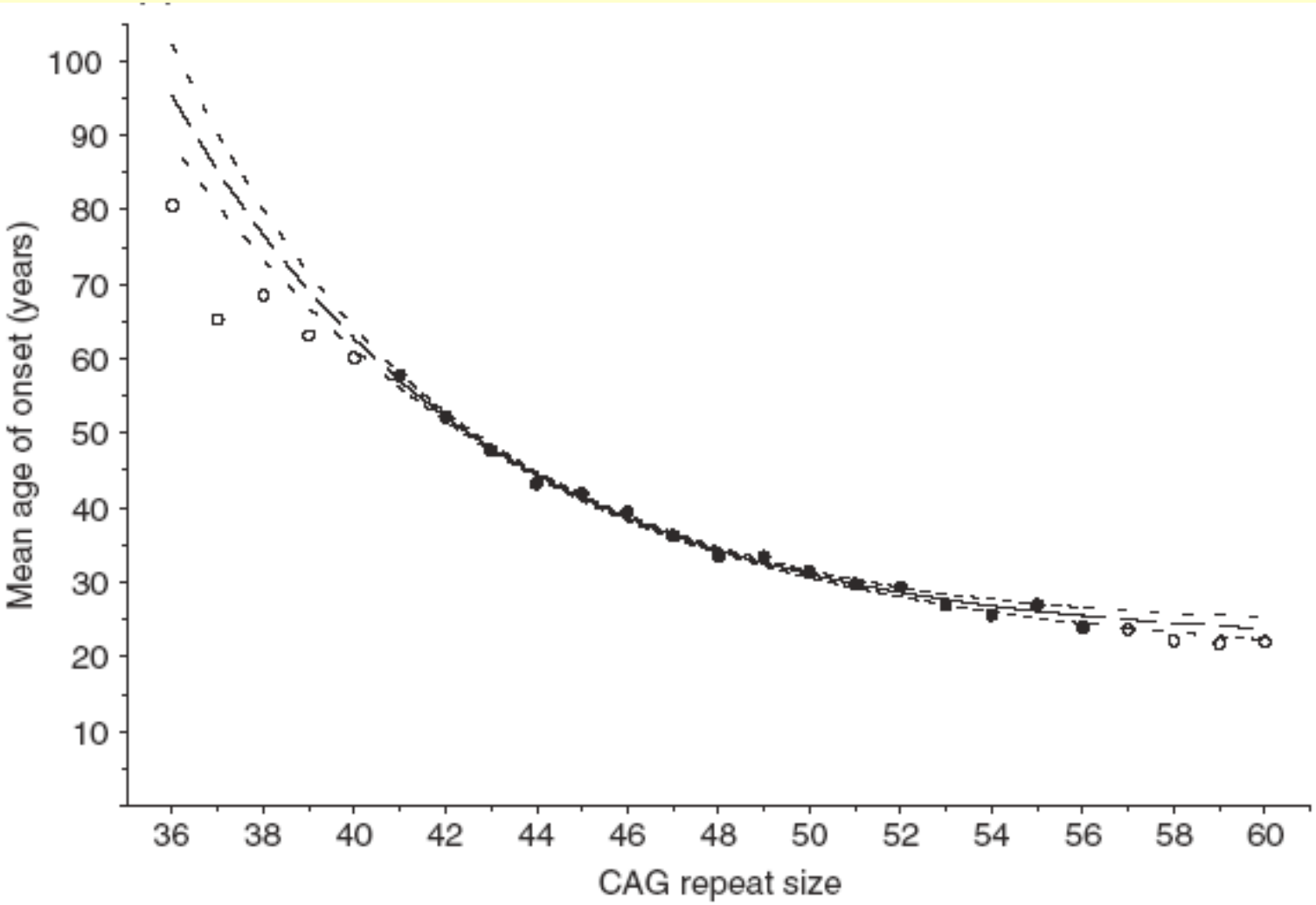
Strand Slippage during DNA Replication

http://www.web-books.com/MoBio/Free/Ch7F3.htm

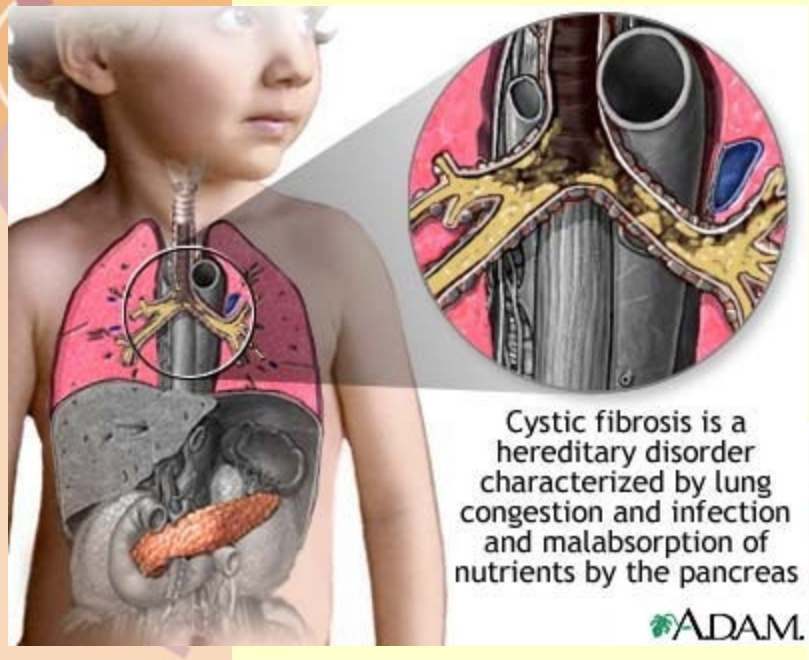


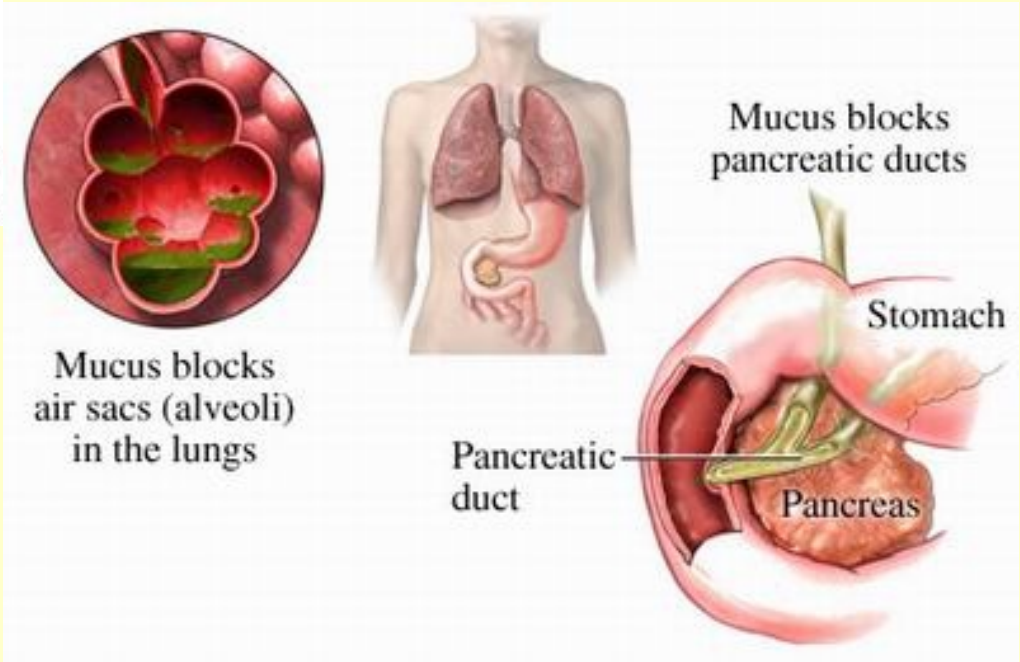
Chi and Lam (2005) Structural roles of CTG repeats in slippage expansion during DNA replication Nucleic acids Res. 33, 1604-1617.

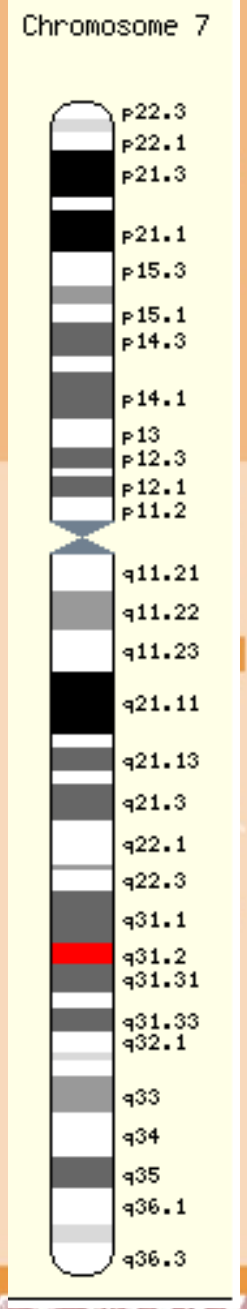
Age of Onset and Repeat Length



Cystic Fibrosis



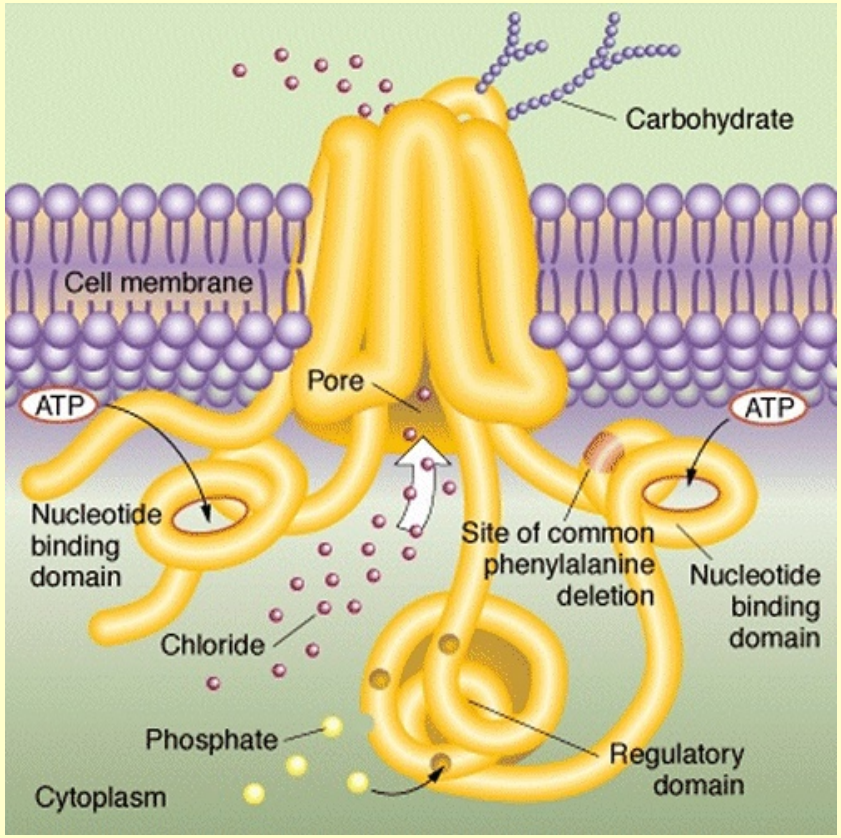




Cystic Fibrosis

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

Cystic Fibrosis Transmembrane Conductance Regulator



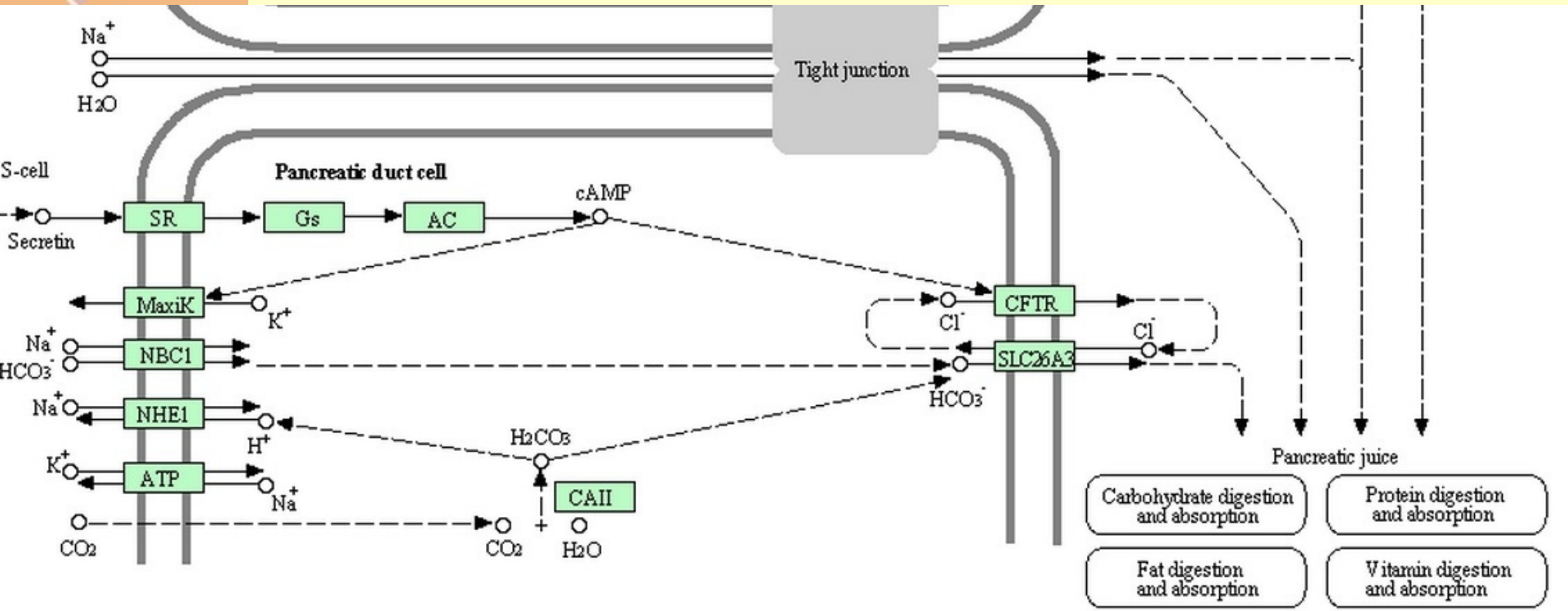
Mutations Causing Cystic Fibrosis

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

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

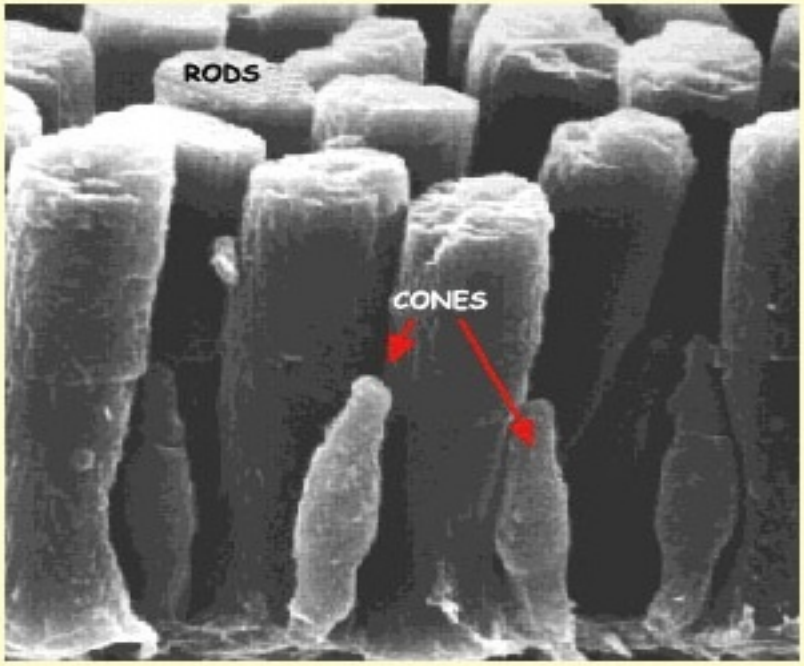
Role of CFTR in Pancreatic Secretion

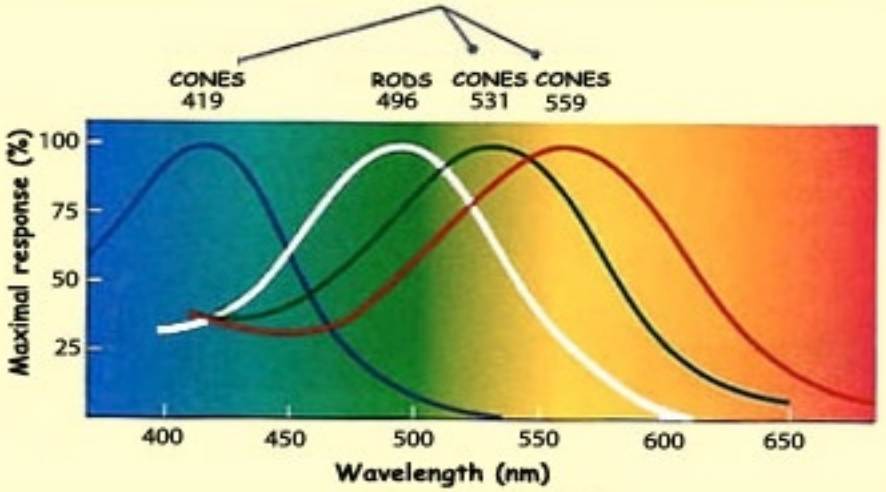
http://www.ncbi.nlm.nih.gov/biosystems/169306



Cone Cells in Retina Permit Color Vision

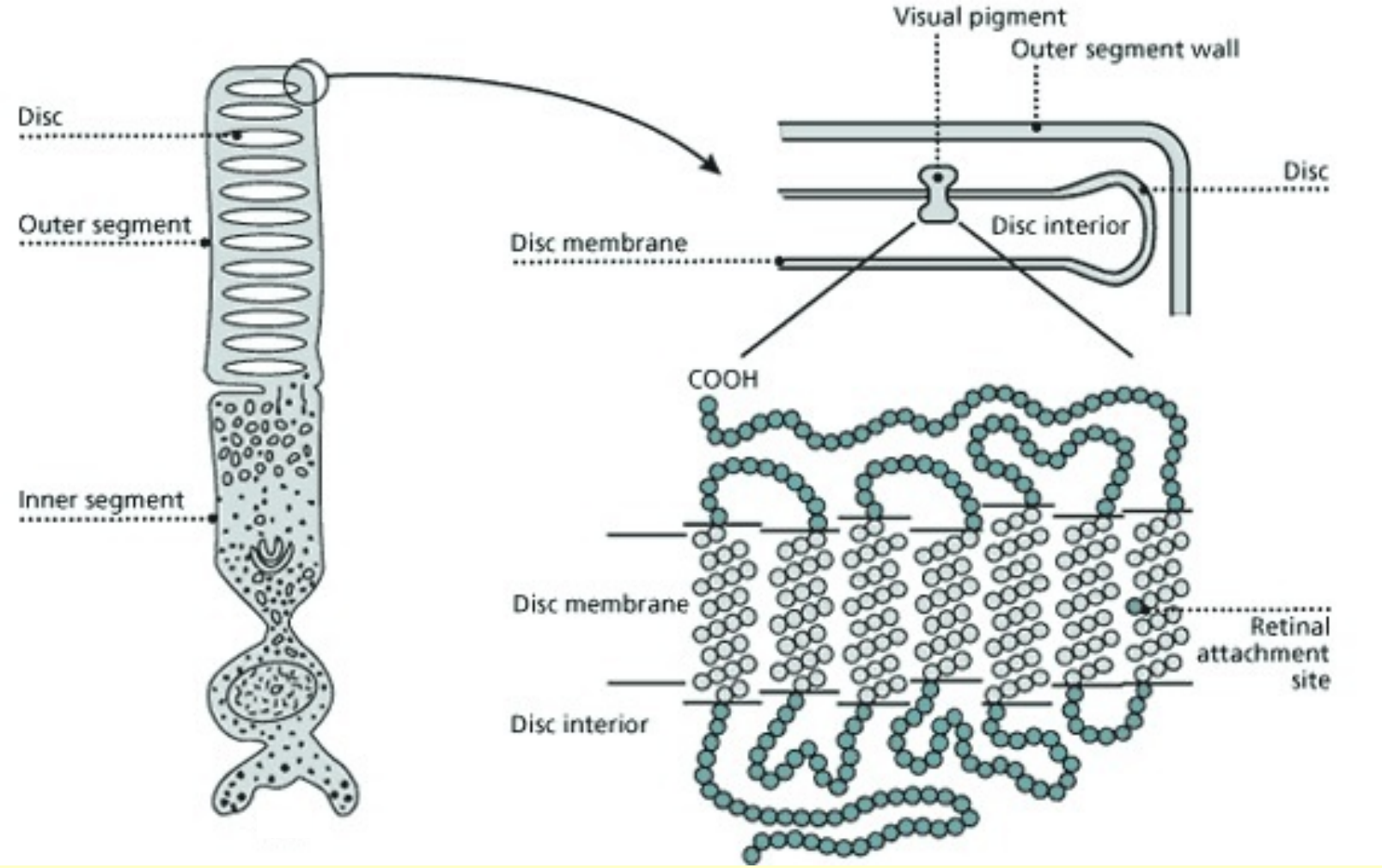
http://en.wikipedia.org/wiki/Opsin





Opsins and Colorblindness

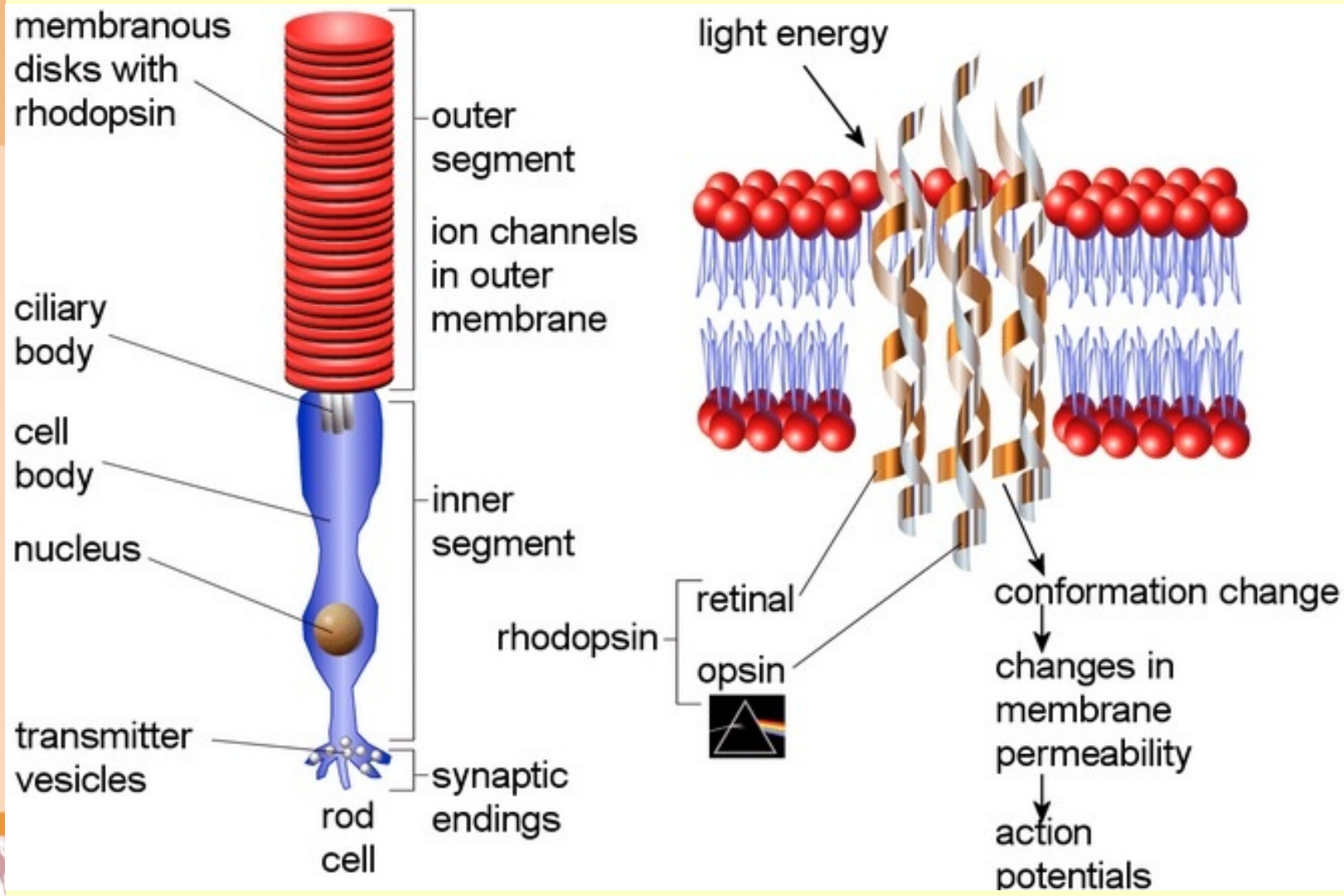
http://en.wikipedia.org/wiki/Opsin



Opsins are the visual pigments in the rod and cone cells

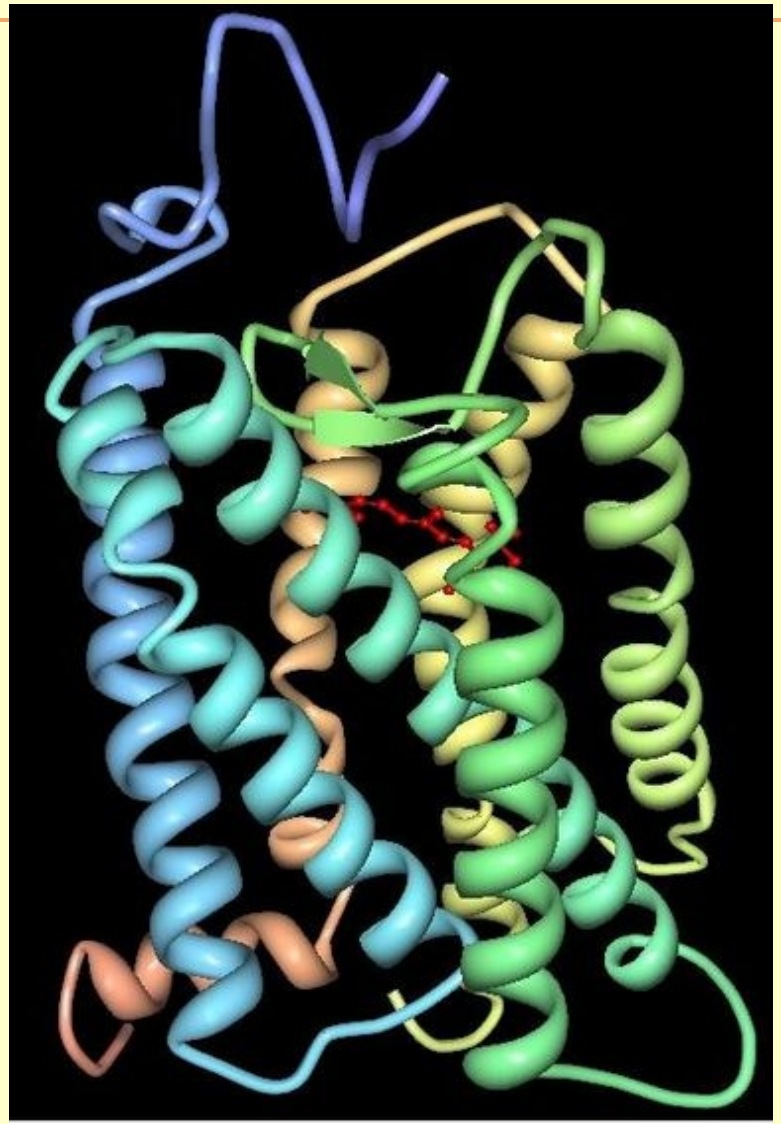
Rhodopsin and Colorblindnes

http://justinpamute.files.wordpress.com/2010/06/rhodopsin1.gifs



Opsins and Colorblindness

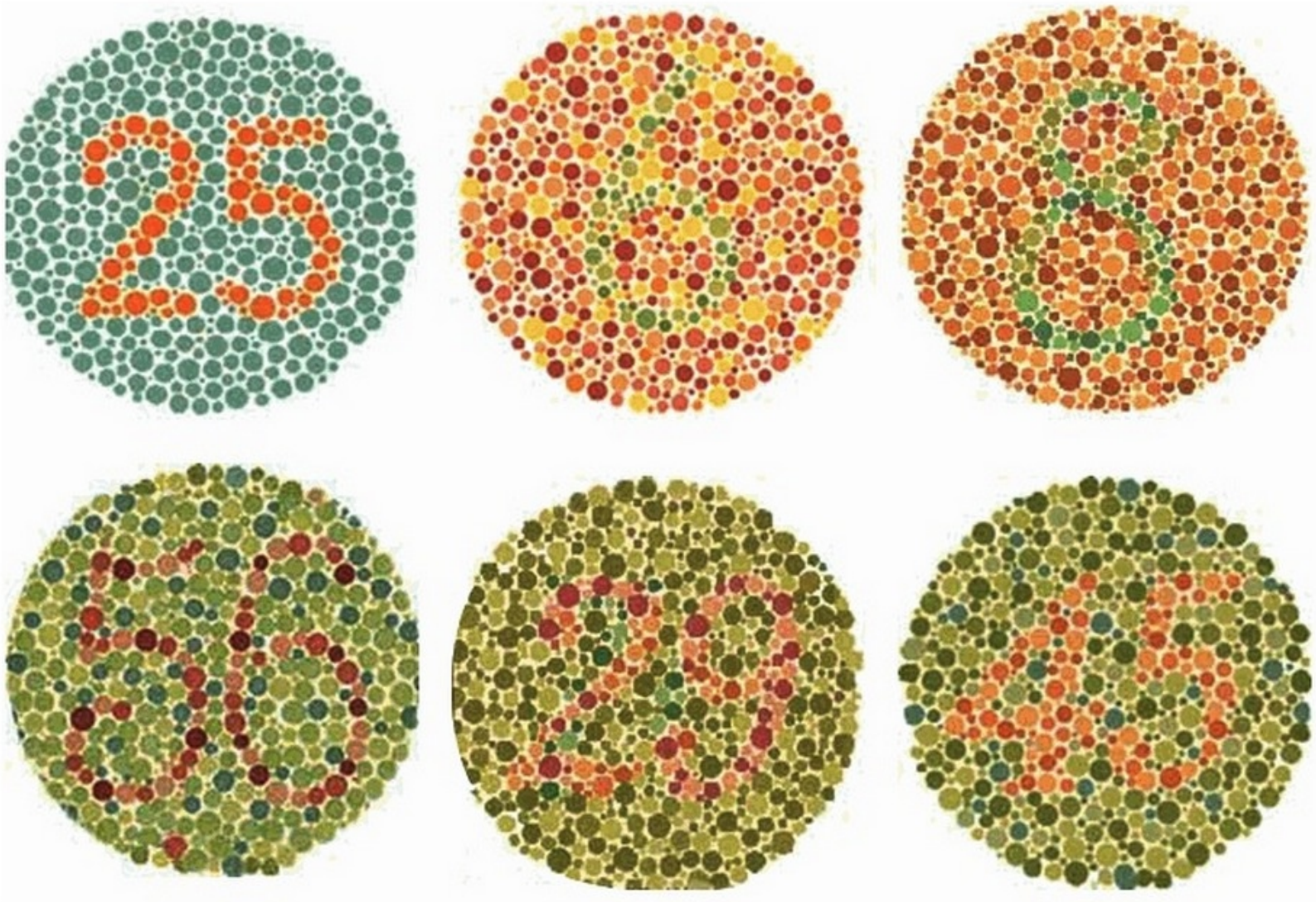
http://en.wikipedia.org/wiki/Opsin



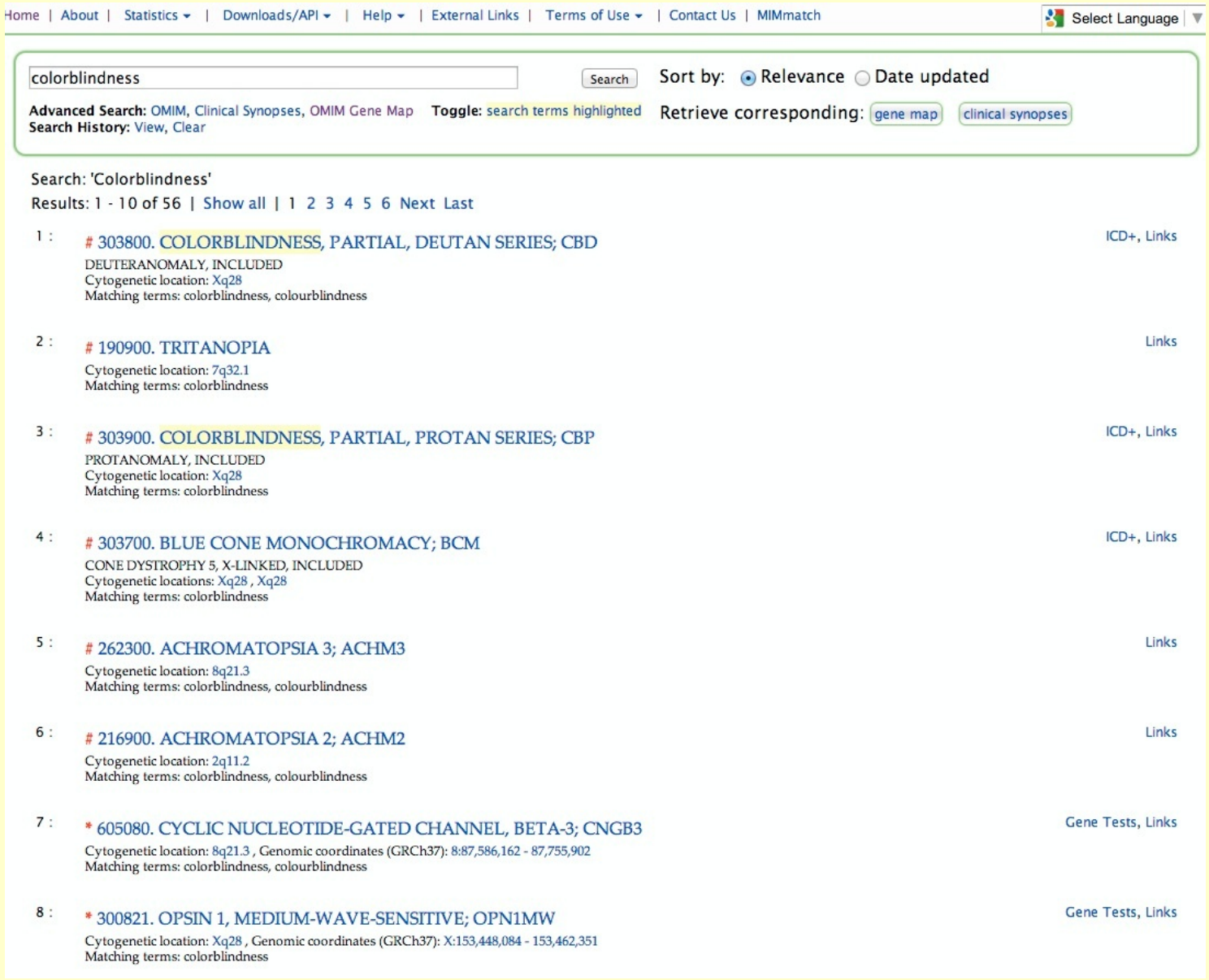
Rhodopsin (7TM, GPCR) and 11-cis retinal

Diagnosis of Colorblindness

http://www.ncbi.nlm.nih.gov/books/NBK1301/



Colorblindness in OMIM



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colorblindness Search Sort by: Relevance Date updated

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map Toggle: search terms highlighted
Search History: View, Clear

Retrieve corresponding: gene map clinical synopses

Search: 'Colorblindness'

Results: 1 - 10 of 56 | Show all | 1 2 3 4 5 6 Next Last

1 : # 303800. COLORBLINDNESS, PARTIAL, DEUTAN SERIES; CBD — ICD+, Links
DEUTERANOMALY, INCLUDED
Cytogenetic location: Xq28
Matching terms: colorblindness, colourblindness

2 : # 190900. TRITANOPIA — Links
Cytogenetic location: 7q32.1
Matching terms: colorblindness

3 : # 303900. COLORBLINDNESS, PARTIAL, PROTAN SERIES; CBP — ICD+, Links
PROTANOMALY, INCLUDED
Cytogenetic location: Xq28
Matching terms: colorblindness

4 : # 303700. BLUE CONE MONOCHROMACY; BCM — ICD+, Links
CONE DYSTROPHY 5, X-LINKED, INCLUDED
Cytogenetic locations: Xq28 , Xq28
Matching terms: colorblindness

5 : # 262300. ACHROMATOPSIA 3; ACHM3 — Links
Cytogenetic location: 8q21.3
Matching terms: colorblindness, colourblindness

6 : # 216900. ACHROMATOPSIA 2; ACHM2 — Links
Cytogenetic location: 2q11.2
Matching terms: colorblindness, colourblindness

7 : * 605080. CYCLIC NUCLEOTIDE-GATED CHANNEL, BETA-3; CNGB3 — Gene Tests, Links
Cytogenetic location: 8q21.3 , Genomic coordinates (GRCh37): 8:87,586,162 - 87,755,902
Matching terms: colorblindness, colourblindness

8 : * 300821. OPSIN 1, MEDIUM-WAVE-SENSITIVE; OPN1MW — Gene Tests, Links
Cytogenetic location: Xq28 , Genomic coordinates (GRCh37): X:153,448,084 - 153,462,351
Matching terms: colorblindness

Colorblindness in OMIM

http://omim.org/entry/303800

#303800

COLORBLINDNESS, PARTIAL, DEUTAN SERIES; CBD

Alternative titles; symbols

DEUTAN COLORBLINDNESS; DCB
DEUTERANOPIA
GREEN COLORBLINDNESS

Other entities represented in this entry:

DEUTERANOMALY, INCLUDED

Phenotype Gene Relationships

Location	Phenotype	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
Xq28	Colorblindness, deutan	303800	OPN1MW	300821

Clinical Synopsis

TEXT

A number sign (#) is used with this entry because deutan colorblindness is caused by mutation in the OPN1MW gene (300821), which encodes green cone pigment.

Description

Normal color vision in humans is trichromatic, being based on 3 classes of cone that are maximally sensitive to light at approximately 420 nm (blue cones; 613522), 530 nm (green cones; 300821), and 560 nm (red cones; 300822). Comparison by neural circuits of light absorption by the 3 classes of cone photoreceptors allows perception of red, yellow, green, and blue colors individually or in various combinations. Dichromatic color vision is severely defective color vision based on the use of only 2 types of photoreceptors, blue plus green (protanopia; see 303900) or blue plus red (deuteranopia). Anomalous trichromacy is trichromatic color vision based on a blue, green, and an anomalous red-like

Opsin1 Gene in OMIM

http://omim.org/entry/300821

*300821

OPSIN 1, MEDIUM-WAVE-SENSITIVE; OPN1MW

Alternative titles; symbols

GREEN CONE PIGMENT; GCP

***HGNC Approved Gene Symbol:** OPN1MW*

***Cytogenetic location:** Xq28* ***Genomic coordinates (GRCh37):** X:153,448,084 - 153,462,351* (from NCBI)

Table of Contents - *300821
External Links:
Genome
DNA
Protein
Gene Info
BioGPS
Ensembl
NCBI Gene
GeneCards
KEGG
PharmGKB
UCSC
Clinical Resources
Variation
Animal Models
Cellular Pathways

Gene Phenotype Relationships

Location	Phenotype	Phenotype MIM number
Xq28	Blue cone monochromacy	303700
	Colorblindness, deutan	303800

TEXT

Description

The medium-wave-sensitive opsin-1 gene (OPN1MW) encodes green cone pigment, 1 of 3 light-sensitive pigments that mediate human color vision. The green-sensitive and the red-sensitive (OPN1LW; 300822) opsins comprise a family of repeated genes on the X chromosome. Whereas there is a single red pigment gene, green pigment genes vary in number among persons with normal color vision. The red pigment gene and the multiple green pigment genes are arranged in a head-to-tail tandem array. The maximal sensitivity of green cones is 530 nm (Nathans et al., (1986, 1986)).

A master switch for the genes of this locus, called the locus control region (LCR; 300824), is located between 3.1 kb and 3.7 kb 5-prime of the gene array and has been shown to be essential for expression of both the red and green pigment genes as well as cone-specific expression of the genes and their segregated expression in separate cones (summary by Deeb, 2005).

Cloning

Opsin1MW Gene Entry

http://www.ncbi.nlm.nih.gov/gene/2652

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Display Settings: Full Report Send to:

OPN1MW opsin 1 (cone pigments), medium-wave-sensitive [*Homo sapiens*]

Gene ID: 2652, updated on 27-Sep-2011

Summary

Official Symbol	OPN1MW provided by HGNC
Official Full Name	opsin 1 (cone pigments), medium-wave-sensitive provided by HGNC
Primary source	HGNC:4206
See related	Ensembl:ENSG00000147380; HPRD:02365; MIM:300821
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	CBD; GCP; GOP; CBBM; COD5; OPN1MW1; OPN1MW2; MGC176615; MGC177321; MGC198468; MGC198469
Summary	This gene encodes for a light absorbing visual pigment of the opsin gene family. The encoded protein is called green cone photopigment or medium-wavelength sensitive opsin. Opsins are G-protein coupled receptors with seven transmembrane domains, an N-terminal extracellular domain, and a C-terminal cytoplasmic domain. The long-wavelength opsin gene and multiple copies of the medium-wavelength opsin gene are tandemly arrayed on the X chromosome and frequent unequal recombination and gene conversion may occur between these sequences. X chromosomes may have fusions of the medium- and long-wavelength opsin genes or may have more than one copy of these genes. Defects in this gene are the cause of deutanopic colorblindness. [provided by RefSeq, Mar 2009]

Genomic context

Location : Xq28
Sequence : Chromosome: X; NC_000023.10 (153448085..153462352)

See OPN1MW in MapViewer

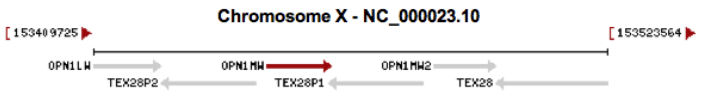

Table of contents

- Summary
- Genomic context
- Genomic regions, transcripts, and products
- Bibliography
- Phenotypes
- General gene info
- General protein info
- Reference sequences
- Related sequences
- Additional links

Links

- Order cDNA clone
- BioAssay, by Gene target
- BioAssays, Gene target, Active
- BioProjects
- BioSystems
- Books
- CCDS
- Conserved Domains
- dbVar
- Full text in PMC
- Genome
- GEO Profiles
- HomoloGene
- Map Viewer
- Nucleotide
- OMIM
- Probe
- Protein
- PubChem Compound
- PubChem Substance

Color Vision Post Gene Therapy

http://www.neitzvision.com/content/genetherapy.html#daltonvideo

Genetic and Medical Web Sites

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

Mendelian Disease Case Presentation

http://biochem158.stanford.edu/case-presentation.html

Please choose a single gene, Mendelian disease from one of the Disease databases (Genes and Disease, Genetics Home Reference, Gene Reviews or Online Inheritance in Man (OMIM) and prepare a written case presentation of the disease (4 pages max) of double spaced text. Figures, Tables and References need not be included in this limit, just the written text

Please Include:

1. A URL pointer to OMIM and/or Gene Reviews entry for your disease
2. A basic description of the disease and its symptoms and prevalence
3. The classical (pre-genetic) differential diagnosis of the disease
4. The classical (pre-genetic) treatment of the disease
5. A description of genetics of the disease including world and ethnic distribution of the disease gene
6. Any novel diagnostics that have resulted from knowing the genetics
7. Any novel understanding of the disease that has lead to novel therapy based on genetic knowledge.

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?

Centers for Mendelian Genomics

http://mendelian.org/