



DIU Génétique et Reproduction

Introduction aux Banques de données biomédicales

18 Novembre 2022
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Bases de données biomédicales

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Polymorphism

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Mutations

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Clinic and Drugs

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OMIM

Online Mendelian Inheritance in Man

<http://omim.org>



OMIM[®]

Online Mendelian Inheritance in Man[®]

An Online Catalog of Human Genes and Genetic Disorders

Updated November 3, 2017

Search OMIM for clinical features, phenotypes, genes, and more...



Advanced Search : [OMIM](#), [Clinical Synopses](#), [Gene Map](#)

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Mirror site : mirror.omim.org

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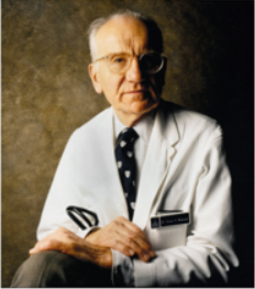
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OMIM® - Online Mendelian Inheritance in Man®

Profiles in Science
National Library of Medicine

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Victor McKusick (1921-2008) is widely considered to be the founding educator, and researcher, he established the first medical genetics program. *Mendelian Inheritance in Man*, an annually updated catalog of human genetic disorders, was conducted landmark studies of hereditary disorders in the Amish. He was involved in the early years of the Human Genome Project, and served in 1997 in recognition of his lifelong contributions he received the Lasker Award for Basic Medical Research.

NLM's Profiles in Science -- The McKusick

PERSPECTIVES IN HUMAN GENETICS

Mendelian Inheritance in Man and Its Online Version, OMIM

Victor A. McKusick

Last year marked the 40th anniversary of the publication of the first print edition of *Mendelian Inheritance in Man* (MIM).¹ This seems an appropriate juncture at which to review its origins, evolution, and present status, including and particularly those of its online version, OMIM (Online Mendelian Inheritance in Man). This is an opportunity, at the same time, to review in brief the rapid progress in an important part of medical genetics and genomics, as chronicled in MIM/OMIM over these 40 years, and to contemplate the future challenges of OMIM.

Description of MIM/OMIM

MIM^{1,2} is a comprehensive knowledgebase of human genes and genetic disorders. It contains information on genes and gene products, and on the clinical features of the disorders caused by mutations in these genes. It is useful to researchers and clinicians alike.

1963 as a resource for identifying both "old" and "new" recessive diseases in studies of the Old Order Amish.^{8,9}

In the original three catalogs corresponding to the three major modes of Mendelian inheritance, the entries were arranged alphabetically according to the preferred title of the particular phenotype, and numbering was done consecutively. By the 11th book edition in 1994,² the X-linked catalog had been joined by two other chromosome-specific catalogs: those for the Y chromosome and for the mitochondrial chromosome. Entries in the original autosomal dominant, autosomal recessive, and X-linked catalogs had been assigned unique identification numbers, beginning with 1, 2, and 3, respectively. Entries in the two new chromosome-specific catalogs were given unique numbers starting with 4 (for the Y chromosome) and 5 (for the mitochondrial).

OMIM® and Online MIM

<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC1852721/>

OMIM Content: Scope of Phenotypes

OMIM Focuses On:

- **Single-gene mendelian disease/disorders/phenotypes** (including: cystic fibrosis, sickle cell anemia, achondroplasia, phenotypic traits such as hair and eye color, susceptibility to drug reaction as in malignant hyperthermia and warfarin sensitivity, altered reaction to infection such as herpes simplex encephalitis and progression to AIDS in HIV infection, germline susceptibilities to cancer such as BRCA1 and breast/ovarian cancer, etc.)
- **Complex diseases with significant single gene contribution** (such as: complement factor H and age related macular degeneration)
- **Descriptions of recurrent deletion and duplication syndromes** (e.g., Potocki-Shaffer syndrome, and chromosome 10q26 deletion syndrome)

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



Make **your** views known
about **gene testing**!

OMIM[®]

Online Mendelian Inheritance in Man[®]

An Online Catalog of Human Genes and Genetic Disorders

Updated 28 June 2012

spinocerebellar ataxia

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examples

spinocerebellar ataxia

spinocerebellar ataxia affecting
spinocerebellar ataxia characterized
spinocerebellar ataxia combined
spinocerebellar ataxia designated
spinocerebellar ataxia due
spinocerebellar ataxia genes
spinocerebellar ataxia inherited
spinocerebellar ataxia loci
spinocerebellar ataxia locus

Require any term in your retrieval:
clinodactyly or hypertelorism
clinodactyly hypertelorism

Require all terms in your retrieval:
clinodactyly and hypertelorism
+clinodactyly +hypertelorism

Require some terms in your retrieval:
clinodactyly not hypertelorism
+clinodactyly -hypertelorism

Require phrases and terms in your retrieval:
"short stature" and clinodactyly
+"short stature" +clinodactyly



ational Human
Genome Research
Institute



18

NOTE: OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders and clinical genetics in clinical practice and in the basic 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.

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Sort by: ☒ Relevance ☐ Date updated

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map

Would you also like: ☐ "dorsal spinocerebellar" ☐ Add All

Retrieve corresponding:

Clinical synopses corresponding to the MIM search: [spinocerebellar ataxia](#).

Search: 'spinocerebellar ataxia' (Records with: clinical synopsis; Retrieve: clinical synopsis)

Results: 1 - 10 of 521 [Show top 100](#) | [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) [Next](#) [Last](#)

1 : [# 164400. SPINOCEREBELLAR ATAXIA 1; SCA1](#)

[Inheritance, Head & Neck, Muscle, soft tissues, Neurology](#)
Matching terms: ataxia, spinocerebellar

2 : [# 183090. SPINOCEREBELLAR ATAXIA 2; SCA2](#)

[Inheritance, Head & Neck, Abdomen, Genitourinary, Neurology](#)
Matching terms: ataxia, spinocerebellar

3 : [# 109150. MACHADO-JOSEPH DISEASE; SCA3](#)

[Inheritance, Head & Neck, Abdomen, Genitourinary, Muscle, soft tissues, Neurology](#)
Matching terms: ataxia, spinocerebellar

4 : [# 183086. SPINOCEREBELLAR ATAXIA 6; SCA6](#)

[Inheritance, Head & Neck, Neurologic, Miscellaneous, Muscle, soft tissues, Neurology](#)
Matching terms: ataxia, spinocerebellar

5 : [# 606658. SPINOCEREBELLAR ATAXIA 13; SCA13](#)

[Inheritance, Head & Neck, Neurologic, Miscellaneous, Muscle, soft tissues, Neurology](#)
Matching terms: ataxia, spinocerebellar

Click to open

#164400 SPINOCEREBELLAR ATAXIA 1; SCA1

CATEGORY	SUBCATEGORY	FEATURES
Inheritance	-	Autosomal dominant
Head and Neck	Eyes	Supranuclear ophthalmoplegia Nystagmus Slow saccades Hypermetria Optic atrophy
Muscle, Soft Tissue	-	Amyotrophy
Neurologic	Central Nervous System	Progressive cerebellar ataxia Hyperreflexia (early) Dorsal column and spinocerebellar tract degeneration
	Peripheral Nervous System	Sensory and motor nerve conduction abnormalities
Miscellaneous	-	Onset in third or fourth decade Genetic anticipation Paternal anticipation bias
Molecular Basis	-	Caused by expanded CAG trinucleotide repeats in the ataxin-1 gene (ATX1, 601556.0001).

[Gene Tests, Links](#)

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Clinical Synopses Results

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Sort by: ☒ Relevance ☐ Date updated

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map Display: Search History: [View](#), [Clear](#)

Retrieve corresponding:

Would you also like: ☐ "dorsal spinocerebellar" ☐ Add All

Clinical synopses corresponding to the MIM search: [spinocerebellar ataxia](#).

Search: 'spinocerebellar ataxia' (Records with: clinical synopsis; Retrieve: clinical synopsis)

Results: 1 - 10 of 521 | [Show top 100](#) | [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) [Next](#) [Last](#)

1: # 164400. SPINOCEREBELLAR ATAXIA 1; SCA1

[Gene Tests](#), [Links](#)

[Inheritance](#), [Head & Neck](#), [Muscle, soft tissues](#), [Neurologic](#), [Miscellaneous](#), [Molecular basis](#),

Matching terms: ataxia, spinocerebellar

2: # 183090. SPINOCEREBELLAR ATAXIA 2; SCA2

[Gene Tests](#), [Links](#)

[Inheritance](#), [Head & Neck](#), [Muscle, soft tissues](#), [Neurologic](#), [Miscellaneous](#), [Molecular basis](#),

Autosomal dominant

Matching terms: ataxia, spinocerebellar

3: # 109150. MACHADO-JOSEPH DISEASE; SCA3

[Gene Tests](#), [ICD+](#), [Links](#)

[Inheritance](#), [Head & Neck](#), [Muscle, soft tissues](#), [Neurologic](#), [Miscellaneous](#), [Molecular basis](#),

Matching terms: ataxia, spinocerebellar

4: # 183086. SPINOCEREBELLAR ATAXIA 4; SCA4

[Gene Tests](#), [Links](#)

[Inheritance](#), [Head & Neck](#), [Muscle, soft tissues](#), [Neurologic](#), [Miscellaneous](#), [Molecular basis](#),

Matching terms: ataxia, spinocerebellar

5: # 606658. SPINOCEREBELLAR ATAXIA 15; SCA15

[Gene Tests](#), [Links](#)

[Inheritance](#), [Head & Neck](#), [Neurologic](#), [Miscellaneous](#), [Molecular basis](#),

Matching terms: ataxia, spinocerebellar

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2.5 Gene Map Search Fields

1.1 Basic Search

For basic searches, simply enter the terms in the search box and click the 'Search' button:

duchenne muscular dystrophy

The search engine will search for them in any order, ranking the entries containing all the terms higher than entries containing some of the terms.

Searches are case-insensitive, so a search for 'duchenne', 'Duchenne', or 'DUCHENNE' will return the same entries.

1.2 +/- Operators

Adding a '+' (plus) operator before specific terms will ensure that these appear in the entries returned:

+duchenne +muscular dystrophy

The returned entries will contain the terms 'duchenne' and 'muscular', but may or may not contain the term 'dystrophy'.

Conversely, you can affix a '-' (minus) operator to a term if you want to exclude entries that contain it:

+muscular +dystrophy -duchenne

The returned entries will contain the terms 'muscular' and 'dystrophy', but will not contain the term 'duchenne'.

1.3 Phrase Search

Result Displays: MIM Numbers, etc.

The screenshot displays the OMIM (Online Mendelian Inheritance in Man) website. On the left, a search bar contains the text 'spinocerebellar ataxia'. Below it, a list of search results is shown. The first result is '# 164400. SPINOCEREBELLAR ATAXIA' with a cytogetic location of '6p22.3'. The second result is '# 183090. SPINOCEREBELLAR ATAXIA' with a cytogetic location of '6p21.3'. The third result is '# 109150. MACHADO-JOSEPH DISEASE' with a cytogetic location of '14q32.12'. The fourth result is '* 176980. PROTEIN KINASE C, GAMMA' with a cytogetic location of '19q13.42'. The fifth result is '* 176980. PROTEIN KINASE C, GAMMA' with a cytogetic location of '19q13.42'. The sixth result is '# 606658. SPINOCEREBELLAR ATAXIA 15; SCA15' with a cytogetic location of '15q11-q13'. A red box highlights the search bar and the first two results. A red arrow points from the first result to the second. A red box highlights the fourth result, which is marked with an asterisk. A red box highlights the fifth result, which is also marked with an asterisk. A red box highlights the sixth result, which is marked with an asterisk. A red box highlights the search bar and the first two results. A red box highlights the fourth result, which is marked with an asterisk. A red box highlights the fifth result, which is also marked with an asterisk. A red box highlights the sixth result, which is marked with an asterisk.

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Search
Linking
API Help

spinocerebellar ataxia

Advanced Search: OMIM, Clinical Synopses, OMIM
Search History: View, Clear
Would you also like: ☐ "dorsal spinocerebellar ataxia"

Search: 'spinocerebellar ataxia'
Results: 1 - 10 of 926 | Show top 100 | 1 2

1: # 164400. SPINOCEREBELLAR ATAXIA
Cytogenetic location: 6p22.3
Matching terms: ataxia, spinocerebellar

2: # 183090. SPINOCEREBELLAR ATAXIA
Cytogenetic location: 6p21.3
Matching terms: ataxia, spinocerebellar

3: # 109150. MACHADO-JOSEPH DISEASE
Cytogenetic location: 14q32.12
Matching terms: ataxia, spinocerebellar

4: * 176980. PROTEIN KINASE C, GAMMA
Cytogenetic location: 19q13.42
Matching terms: ataxia, spinocerebellar

5: * 176980. PROTEIN KINASE C, GAMMA
Cytogenetic location: 19q13.42
Matching terms: ataxia, spinocerebellar

6: # 606658. SPINOCEREBELLAR ATAXIA 15; SCA15
Cytogenetic location: 15q11-q13
Matching terms: ataxia, spinocerebellar

OMIM Frequently Asked Questions (FAQs)

1.1 What is OMIM?

1.2 What numbering system is used in the OMIM database?

1.3 What do the symbols preceding a MIM number represent?

1.4 How are mutations cataloged in OMIM?

1.2 What numbering system is used in the OMIM database?

Each OMIM entry is given a unique six-digit number as summarized below:

1----- (100000-) 2----- (200000-) Autosomal loci or phenotypes (entries created before 1994)

3----- (300000-) X-linked loci or phenotypes

4----- (400000-) Y-linked loci or phenotypes

5----- (500000-) Mitochondrial loci or phenotypes

6----- (600000-) Autosomal loci or phenotypes (entries created after May 15, 1994)

Allelic variants (mutations; see 1.4) are designated by the MIM number of the entry followed by a three-digit variant number. For example, allelic variants in the factor IX gene (300746) are designated 300746.001, 300746.002, etc.

1.3 What do the symbols preceding a MIM number represent?

An asterisk (*) before an entry number indicates a gene.

A number symbol (#) before an entry number indicates that it is a descriptive entry.

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<http://www.omim.org/help/faq#1.2>

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spinocerebellar ataxia Sort by: ☒ Relevance ☐ Date updated

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map Display: Toggle highlight
Search History: View, Clear Retrieve corresponding: [gene map](#) [clinical synopses](#)

Would you also like: ☐ "dorsal spinocerebellar" ☐ Add All

Search: 'spinocerebellar ataxia'
Results: 1 - 10 of 926 | [Show top 100](#) | 1 2 3 4 5 6 7 8 9 10 Next Last

1: # 164400. SPINOCEREBELLAR ATAXIA 1; SCA1
Cytogenetic location: 6p22.3
Matching terms: ataxia, spinocerebellar

2: # 183090. SPINOCEREBELLAR ATAXIA 2; SCA2
AMYOTROPHIC LATERAL SCLEROSIS
Cytogenetic location: 12q24.1
Matching terms: ataxia, spinocerebellar

3: # 109150. MACHADO-JOSEPH DISEASE; MJD
Cytogenetic location: 14q32.12
Matching terms: ataxia, spinocerebellar

4: # 183086. SPINOCEREBELLAR ATAXIA 6; SCA6
Cytogenetic location: 19p13.2
Matching terms: ataxia, spinocerebellar

5: * 176980. PROTEIN KINASE C, GAMMA; PRKCG
Cytogenetic location: 19q13.42, Genomic coordinates (GRCh37): 19:54,385,466 - 54,410,900
Matching terms: ataxia, spinocerebellar

6: # 606658. SPINOCEREBELLAR ATAXIA 15; SCA15

short OR ("growth retardation") OR (dwarfism)

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map Display: Toggle highlight
Search History: View, Clear

Would you also like: ☐ dwarf ☒ dwarfism ☐ Add All
☒ "growth retardation" ☐ "short stature"

Gene Tests, ICD+, Links

Gene Tests, Links

Gene Tests, Links

Gene Tests, Links

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Matching Terms

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spinocerebellar ataxia Search Sort by: ☒ Relevance ☐ Date updated

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map Display: Toggle highlight
Search History: View, Clear
Would you also like: ☐ "dorsal spinocerebellar" ☐ Add All

Retrieve corresponding: gene map clinical synopses

Search: 'spinocerebellar ataxia'
Results: 1 - 10 of 926 | Show top 100 | 1 2 3 4 5 6 7 8 9 10 Next Last

1 : **# 164400. SPINOCEREBELLAR ATAXIA 1; SCA1** Gene Tests, Links
Cytogenetic location: 6p22.3
Match 1 : **# 164400. SPINOCEREBELLAR ATAXIA 1; SCA1**
Cytogenetic location: 6p22.3
Matching terms: ataxia, spinocerebellar ← Matching terms

2 : **# 183** Gene Tests, Links
AMYC
Cytogenetic location: 12q24.12
Matching terms: ataxia, spinocerebellar

3 : Search: 'spinocerebellar ataxia'
Results: 921 - 926 of 926 | Show top 100 | First Previous 84 85 86 87 88 89 90 91 92 **93** CD+, Links

4 : 921 : *** 614571. CHROMOSOME 5 OPEN READING FRAME 42; C5ORF42** ests, Links
Cytogenetic location: 5p13.2 Ch37: 5:37,106,329 - 37,249,529
Matching terms: ataxia ← one term

5 : *** 176980. PROTEIN KINASE C, GAMMA; PRKCG** Gene Tests, Links
Cytogenetic location: 19q13.42, Genomic coordinates (GRCh37): 19:54,385,466 - 54,410,900
Matching terms: ataxia, spinocerebellar

6 : **# 606658. SPINOCEREBELLAR ATAXIA 15; SCA15** Gene Tests, Links

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spinocerebellar ataxia Sort by: ☒ Relevance ☐ Date updated

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map, Display Toggle highlight

Search History: View, Clear

Would you also like: ☐ "dorsal spinocerebellar ataxia"

Search Result for OMIM# '164400'

Spinocerebellar Ataxia Type 1 [Testing](#) [Reviews](#) [Resources](#) [OMIM](#) [Locus-Specific](#) [HGMD](#) [More Links](#)

Search: 'spinocerebellar ataxia'

Results: 1 - 10 of 926 | [Show top 100](#) | 1 2 3 4 5 6 7 8 9 10 [Next](#) [Last](#)

1 : **# 164400. SPINOCEREBELLAR ATAXIA 1; SCA1**

Cytogenetic location: 6p22.3

Matching terms: ataxia, spinocerebellar

[Gene Tests, Links](#)

219700. CYSTIC FIBROSIS; CF

Cytogenetic locations: 7q31.2, 19q13.2

Matching terms: fibrosis, cystic

[Gene Tests, Newborn Screening, ICD+, Links](#)

External Links for #164400

Clinical Resources

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Animal Models

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Cell Lines

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- [Cellular Pathways](#)
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[Gene Tests, Links](#)

Resources

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PROFESSIONAL RESOURCES

Please choose a category below.

ACT Sheets (ACMG)

Describes the short term actions a health professional should follow in the follow-up of an infant that has screened positive.

General Resources [Links](#)

to State programs, government resources, and professional organizations.

Reports and Publications [Links](#)

and PDF files of newborn screening and genetics related papers of interest.

Newborn Screening

Newborn screening specific links.

Genetics

Genetics related links.

Birth Defects

defects related links.

 Birth

ICD+ for #219700

ICD10CM: E84.9,
ICD10CM: E84,
ICD9CM: 277.0,
SNOMEDCT: 190905008

85,466 - 54,410,900

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[Gene Tests, Links](#)

Clinical Synopsis

#164400

SPINOCEREBELLAR ATAXIA 1; SCA1

Alternative titles; symbols

SPINOCEREBELLAR ATROPHY I

OLIVOPONTOCEREBELLAR ATROPHY I; OPCA

CEREBELLOPARENCHYMAL DISORDER I; CPD

MENZEL TYPE OPCA

OLIVOPONTOCEREBELLAR ATROPHY IV; OPCA

SCHUT-HAYMAKER TYPE OPCA

Phenotype Gene Relationships

Location	Phenotype	Phenotype MIM number
6p22.3	Spinocerebellar ataxia 1	164400

Phenotypic Series

[Clinical Synopsis](#)

click

TEXT

A number sign (#) is used with this entry because (CAG)_n trinucleotide repeat in the ataxin-1 gene (4

Description

The autosomal dominant cerebellar degeneration 'spinocerebellar ataxias' (SCAs) even though 'sp

#164400

SPINOCEREBELLAR ATAXIA 1; SCA1

CATEGORY	SUBCATEGORY	FEATURES
Inheritance	-	Autosomal dominant
Head and Neck	Eyes	Supranuclear ophthalmoplegia Nystagmus Slow saccades Hypermetria Optic atrophy
Muscle, Soft Tissue	-	Amyotrophy
Neurologic	Central Nervous System	Progressive cerebellar ataxia Hyperreflexia (early) Scanning speech Dysarthria Dysmetria Hypotonia Loss of deep tendon reflexes (later) Extensor plantar response Extrapyramidal signs Corticospinal signs Chorea Bulbar palsies Dysphagia Dysdiadochokinesis Cognitive impairment, mild Olivopontocerebellar atrophy Dorsal column and spinocerebellar tract degeneration
	Peripheral Nervous System	Sensory and motor nerve conduction abnormalities
Miscellaneous	-	Onset in third or fourth decade Genetic anticipation Paternal anticipation bias
Molecular Basis	-	Caused by expanded CAG trinucleotide repeats in the ataxin-1 gene (ATX1, 601556.0001).

Table of Contents - #164400

Title

Contributors

Creation Date

Edit History

External Links:

Clinical Resources

Centers for Mendelian Genomics

Clinical Synopsis

Provides an overview of the clinical features of a phenotype

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Associated Gene/Locus

#164400

SPINOCEREBELLAR ATAXIA 1; SCA1

Alternative titles; symbols

SPINOCEREBELLAR ATROPHY I

OLIVOPONTOCEREBELLAR ATROPHY I; OPCA I; OPCA1

CEREBELLOPARENCHYMAL DISORDER I; CPD1

MENZEL TYPE OPCA

OLIVOPONTOCEREBELLAR ATROPHY IV; OPCA IV; OPCA4

SCHUT-HAYMAKER TYPE OPCA

[Table of Contents - #164400](#)

External Links:

[Clinical Resources](#)

[Animal Models](#)

[Cell Lines](#)

[Cellular Pathways](#)

[Centers for Mendelian Genomics](#)

Phenotype Gene Relationships

Location	Phenotype	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
6p22.3	Spinocerebellar ataxia 1	164400	ATXN1	601556

click

Phenotypic Series

Clinical Synopsis

TEXT

A number sign (#) is used with this entry because [spinocerebellar ataxia-1](#) is caused by an expanded (CAG)_n trinucleotide repeat in the ataxin-1 gene (ATXN1; [601556](#)).

Description

The autosomal dominant cerebellar degenerative disorders are generally referred to as '[spinocerebellar ataxias](#)' (SCAs) even though '[spinocerebellar](#)' is a hybrid term, referring to both

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OMIM Gene Entry

*601556

ATAXIN 1; ATXN1

HGNC Approved Gene Symbol: [ATXN1](#)

Cytogenetic location: [6p22.3](#) Genomic coordinates (GRCh37): [6:16,299,342 - 16,761,720](#) (from NCBI)

Gene Phenotype Relationships

Location	Phenotype	Phenotype MIM number
6p22.3	Spinocerebellar ataxia 1	164400

TEXT

Description

ATXN1 and ATXN1L ([614301](#)) are chromatin-binding factors that suppress signaling within the Notch pathway (see [190198](#)) in the absence of Notch activation ([Tong et al., 2011](#)).

Cloning

Using DNA fragments from a cosmid clone shown to contain the CAG trinucleotide repeat identified as the cause of spinocerebellar ataxia-1 (SCA1; [164400](#)) by [Orr et al. \(1993\)](#), [Banfi et al. \(1994\)](#) screened 2 human fetal brain and 1 human adult cerebellar cDNA libraries to isolate the ATXN1 transcript. [Banfi et al. \(1994\)](#) demonstrated that the ATXN1 gene encodes an 816-amino acid protein with a molecular mass of 87 kD. The (CAG)_n repeat, coding for a polyglutamine tract, lies within the coding region, and the protein is transcribed from both the wildtype and the CAG-expanded ATXN1 allele.

[Mizutani et al. \(2005\)](#) stated that the deduced 816-amino acid ATXN1 protein has an N-terminal BOAT (ATXN1L) and ataxin-1 (NBA) domain, followed by the polyglutamine tract, a self-association region (SAR), and a C-terminal ATXN1 and HBP1 (AXH) domain.

Expandable
TOC

Table of Contents - *601556

Title
Gene Phenotype Relationships
Text
Description
Cloning
Gene Structure
Mapping
Gene Function
Molecular Genetics
Evolution
Animal Model
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External Links:

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► [Gene Info](#)
► [Clinical Resources](#)
► [Variation](#)
► [Animal Models](#)
► [Cellular Pathways](#)

Full
entry

*602421

Gene CFTR's Table View

CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR; CFTR

Allelic Variants (Selected Examples):

Number	Phenotype	Mutation	dbSNP
.0001	CYSTIC FIBROSIS BRONCHIECTASIS WITH OR WITHOUT ELEVATED SWEAT CHLORIDE 1. MODIFIER OR INCLUDED	CFTR, PHE508DEL	[rs113993960]
		ILE507DEL	-
		GLN493TER	[rs77101217]
		ASP110HIS	[rs113993958]
		ARG117HIS	[rs78655421]
		ARG347PRO	[rs77932196]
		ALA455GLU	[rs74551128]
		IVS10, G-A, -1	[rs76713772]
		GLY542TER	[rs113993959]
		SER549ASN	-
		SER549ILE	[rs121908755]
		SER549ARG	[rs121908757], [rs121909005]
		GLY551ASP	[rs75527207]
		ARG553TER	[rs74597325]
		ALA559THR	[rs75549581]

1.4 How are mutations cataloged in OMIM?

Mutations are cataloged in OMIM in the Allelic Variants section of gene entries (see 1.2). For most genes, only selected mutations are included. Criteria for inclusion include the first mutation to be discovered, high population frequency, distinctive phenotype, historic significance, unusual mechanism of mutation, unusual pathogenetic mechanism, and distinctive inheritance (e.g., dominant with some mutations, recessive with other mutations in the same gene). Most of the allelic variants represent disease-causing mutations. A few polymorphisms are included, many of which show a positive correlation with disease.

<http://www.omim.org/help/faq#1.4>

Description

ATXN1 and ATXN1L (614301) are chromatin-binding factors that suppress signaling within the Notch pathway (see 190198) in the absence of Notch activation (Tong et al., 2011).

Cloning

Allelic Variants

Table View

References

Contributors

Creation Date

Edit History

*601556

ATAXIN 1; ATXN1

Allelic Variants (Selected Examples):

Number	Phenotype	Mutation	dbSNP
.0001	SPINOCEREBELLAR ATAXIA 1	ATXN1, (CAG) _n EXPANSION	-

Table View of Allelic Variants

External Links for Genes: Genome

*601556

ATAXIN1; ATXN1

HGNC Approved Gene Symbol: [ATXN1](#)

Cytogenetic location: [6p22.3](#) Genomic coordinates (GRCh37): [6:16,299,342 - 16,761,720](#) (from NCBI)

External Links

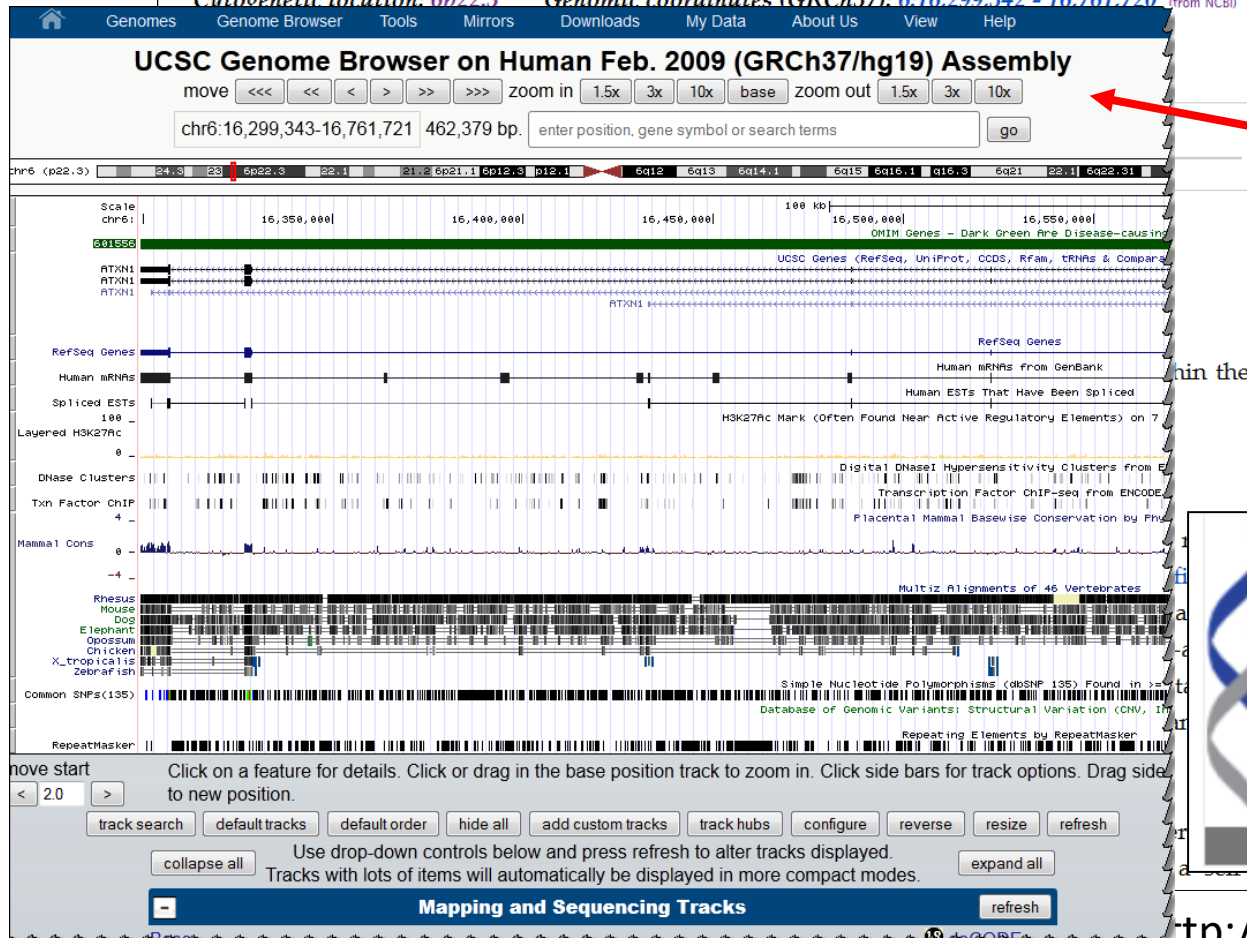


Table of Contents

External Links:

Genome

Ensembl

NCBI Map Viewer

UCSC Genome Browser

DNA

Protein

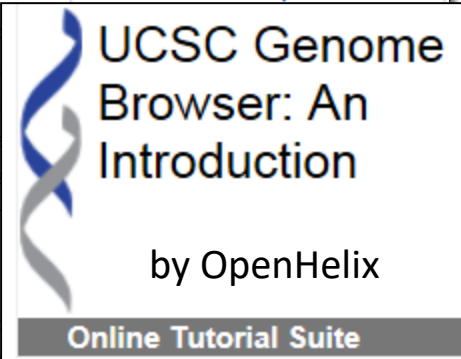
Gene Info

Clinical Resources

Variation

Animal Models

Cellular Pathways



<http://openhelix.com/catalog>

Gene Map Search

spinocerebellar ataxia

Search

Sort by: ☒ Relevance ☐ Date updated

Advanced Search: OMIM, Clinical Synopsis, **OMIM Gene Map** Display: [Toggle highlight](#)
Search History: [View](#), [Clear](#)

Search OMIM

Search

Sort by: ☒ Relevance ☐ Date updated

Advanced Search: OMIM, Clinical Synopsis, **OMIM Gene Map**
Search History: [View](#), [Clear](#)

nts - #164400

*601556

ATAXIN 1; ATXN1

HGNC Approved Gene Symbol: [ATXN1](#)

Cytogenetic location: [6p22.3](#)

Genomic coordinates (GRCh37): [6:16,299,342 -](#)

Gene Phenotype Relationships

Location	Phenotype
6p22.3	Spinocerebellar ataxia 1

Easy access to
Gene Map
search

Mirror sites: [us-east.omim.org](#), [europe.omim.org](#)



Make **your** views known
about **gene testing**!

OMIM[®]

Online Mendelian Inheritance in Man[®]

An Online Catalog of Human Genes and Genetic Disorders

Updated 5 July 2012

Search

Search

[Sample Searches](#)

Advanced Search: OMIM, Clinical Synopsis, **OMIM Gene Map**



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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.

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Gene Map Search Form

Gene Map Advanced Search:

Example searches:
1p36
kinase
CFTR
etc.

3:38,411-11,129,415

Search

Entries per page: 10

Search by genomic region (or cyto location range) to get a list of all OMIM Gene/Loci in that region, for example:

'1:0-124,300,000' or '1p36-p32'

A message will be displayed indicating when a genomic region search is run.

Search by genomic location (or cyto location band) to jump to that location in the chromosome, for example:

'1:124,300,000' or '1p32'

Search by chromosome to list it, for example:

'1' or 'chr1'

All other searches will return the OMIM Gene/Loci that match that search, for example:

'disorder' or 'kinase'

Note that the genomic regions are from the GRCh37 build.

Search tips

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

Search limits

☐ Autosomal

☐ Phenotype exists

Clear

Search

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Gene Map Search Results

Gene Map

3:38,411-11,129,415

Search

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

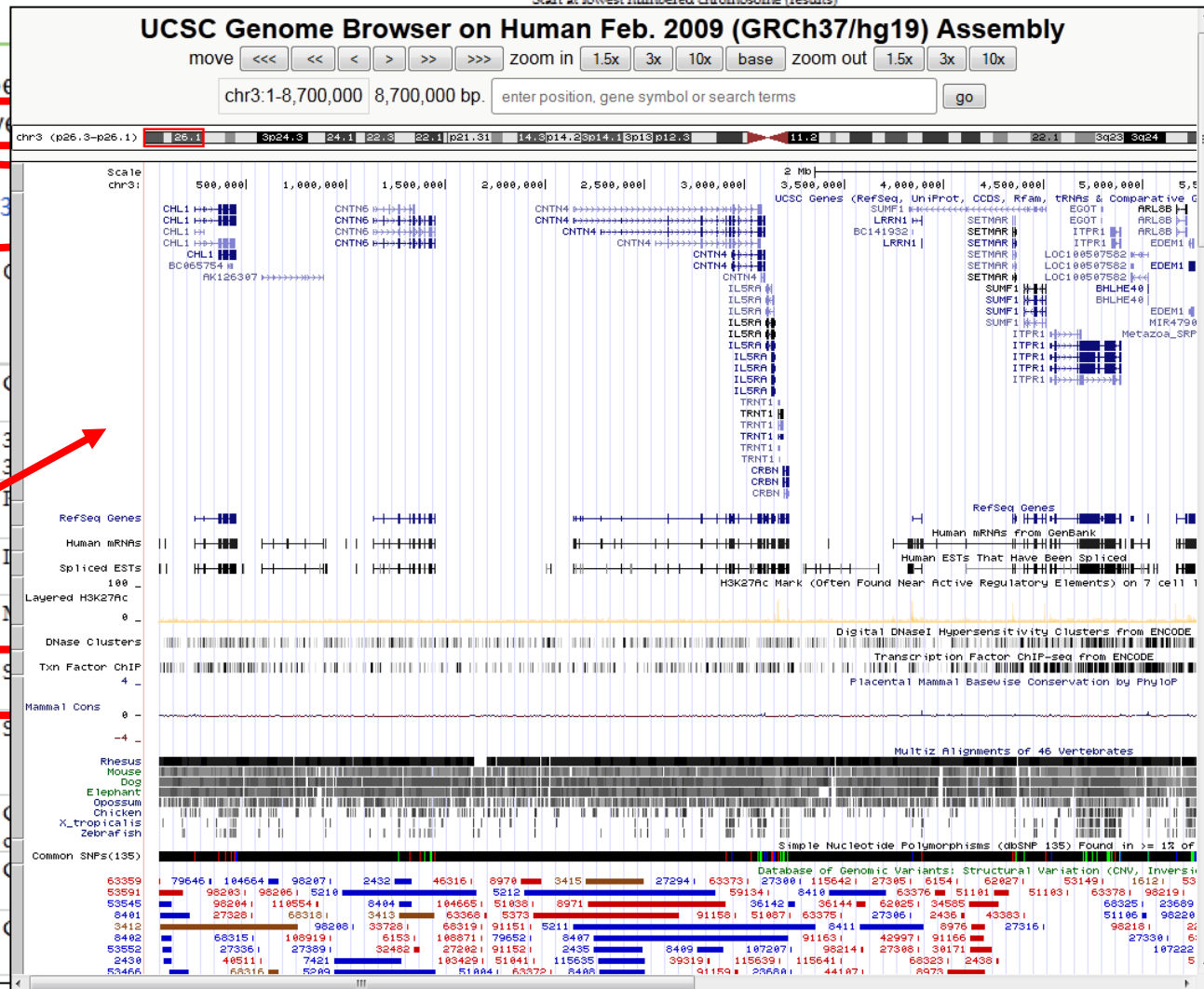
Phenotype Only Entries All Entries

The search '3:38,411-11,129,415' mapped to chr3:38,411-11,129,415 and retrieved entries that start, or end, or overlap this region.

Search: '3:38,411-11,129,415'

Results: 1 - 10 of 44 | Show all | 1 2 3

Genomic context table	Location (genomic start, cyto location) (from NCBI)	Gene/Locus
1:	3:0 3p	CRCL
2:	3:0 3pter-p25	DEL3pterp25, C3DELpterp25
3:	3:0 3p26	HPC5
4:	3:0 3p26	IBD9
5:	3:0 3p26-p24	MYMY1, MYMY
6:	3:0 3p26	SCA29
7:	3:0 3p26	STQTL5
8:	3:238,278 3p26.3	CHL1, CALL, L1CAM2
9:	3:1,134,628 3p26.3	CNTN6, NB3
10:	3:2,140,549 3p26.3-p26.2	CNTN4



OMIM Summary: Gene, Variant & Phenotype Data

#164400
SPINOCEREBELLAR ATAXIA 1; SCA1

Detailed
Entries

Alternative titles; symbols

SPINOCEREBELLAR ATROPHY I
OLIVOPONTOCEREBELLAR ATROPHY I; OPCA I; OPCA1
CEREBELLOPARENCHYMAL DISORDER I; CPD1
MENZEL TYPE OPCA
OLIVOPONTOCEREBELLAR ATROPHY IV; OPCA4
SCHUT-HAYMAKER TYPE OPCA

Phenotype Gene Relationships

Location	Phenotype
6p22.3	Spinocerebellar ataxia 1

Phenotypic Series

Clinical Synopsis

TEXT

A number sign (#) is used with this entry because the gene is located on chromosome 6 (6p22.3).

Description

The autosomal dominant cerebellar degenerative disorders are generally referred to as 'spinocerebellar ataxias,' (SCAs) even though 'spinocerebellar' is a hybrid term, referring to both clinical signs and neuroanatomical regions (Margolis, 2003). Neuropathologists have defined SCAs as cerebellar ataxias with variable involvement of the brainstem and spinal cord, and the clinical features of the disorders are caused by degeneration of the cerebellum and its afferent and efferent connections, which involve the brainstem and spinal cord (Schols et al., 2004; Taroni and DiDonato, 2004).

ADCA III was a pure form of late-onset cerebellar ataxia without additional features. SCA1, SCA2 (183090), and SCA3, or Machado-Joseph disease (109150), are considered to be forms of ADCA I. These 3 disorders are characterized at the molecular level by CAG repeat expansions on 6p24-p23, 12q24.1, and 14q32.1, respectively. SCA7 (607640), caused by a CAG repeat expansion in the ATXN7 gene (607640) on chromosome 3p13-p12, is a form of ADCA II. SCA5 (600224), SCA31 (117210), SCA6 (183086), and SCA11 (600432) are associated with phenotypes most suggestive of ADCA III. However, Schelhaas et al. (2000) noted that there is significant phenotypic overlap between different forms of SCA as well as significant phenotypic variability within each subtype.

OMIM Gene/Loci: 68 - 77 of 784 on Chromosome 6 (All Entries) | Show 100

pter Towards pter Back 3 Forward 3 Towards qter qter

Location (genomic start, cytoband location) (from NCB)	Gene/Locus	Gene/Locus name	Gene/Locus MIM number	Phenotype	Phenotype MIM number	Pheno map key	Comments	Mouse symbol (from MGI)
6:15,246,526-6p22.3	JMJ	Jumonji	601594					Jarid2
6:15,523,031-6p22.3	DTNBP1, HPS7	Dystrobrevin-binding protein 1 (dysbindin)	607145	Hermansky-Pudlak syndrom (HPS7)	614076	3		Dtnbp1

Phenotypic Series

Spinocerebellar ataxia - 164400

Location	Phenotype	Phenotype mapping	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
----------	-----------	-------------------	----------------------	------------	-----------------------

1p21-q21	Spinocerebellar ataxia 19
2p21-p13	Spinocerebellar ataxia 25
3p26	Spinocerebellar ataxia 29
3p26.1	Spinocerebellar ataxia 15
3p14.1	Spinocerebellar ataxia 7
4q34.3-q35.1	?Spinocerebellar ataxia 30
5q32	Spinocerebellar ataxia 12
6p22.3	Spinocerebellar ataxia 1
6p12.3-q16.2	Spinocerebellar ataxia 34
6q27	Spinocerebellar ataxia 17
7p21.3-p15.1	Spinocerebellar ataxia 21
7q22-q32	Spinocerebellar ataxia 18
7q32-q33	Spinocerebellar ataxia 32

#164400
SPINOCEREBELLAR ATAXIA 1; SCA1

Clinical Synopsis

CATEGORY	SUBCATEGORY	FEATURES
Inheritance	-	Autosomal dominant
Head and Neck	Eyes	Supranuclear ophthalmoplegia Nystagmus Slow saccades Hypermetria Optic atrophy
Muscle, Soft Tissue	-	Amyotrophy
Neurologic	Central Nervous System	Progressive cerebellar ataxia Hyperreflexia (early) Scanning speech Dysarthria Dysmetria Hypotonia Dorsal column and spinocerebellar tract degeneration
	Peripheral Nervous System	Sensory and motor nerve conduction abnormalities
Miscellaneous	-	Onset in third or fourth decade Genetic anticipation Paternal anticipation bias
Molecular Basis	-	Caused by expanded CAG trinucleotide repeats in the ataxin-1 gene (ATX1, 601556.0001).

Contributors: Cassandra L. Kniffin - revised : 08/14/2002
Creation Date: John F. Jackson : 6/15/1995
Edit History: kniffin : 08/14/2002

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OMIM Summary: Data, cont.

219700. CYSTIC FIBROSIS; CF

Cytogenetic locations: 7q31.2, 19q13.2

Gene Tests, Newborn Screening, ICD+, Links

Links to external sources

ICD+ for #219700

ICD10CM: E84.9,
ICD10CM: E84,
ICD9CM: 277.0,
SNOMEDCT: 190905008

Table of Allelic Variants

*602421

CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR; CFTR

Allelic Variants (Selected Examples):

Number	Phenotype	Mutation	dbSNP
.0001	CYSTIC FIBROSIS BRONCHIECTASIS WITH OR WITHOUT ELEVATED SWEAT CHLORIDE 1, MODIFIER OF, INCLUDED	CFTR, PHE508DEL	-
.0002	CYSTIC FIBROSIS	CFTR, ILE507DEL	-
.0003	CYSTIC FIBROSIS	CFTR, GLN493TER	[rs77101217]
.0004	CYSTIC FIBROSIS	CFTR, ASP110HIS	[rs113993958]
.0005	CYSTIC FIBROSIS VAS DEFERENS, CONGENITAL BILATERAL ABSENCE OF, INCLUDED	CFTR, ARG117HIS	[rs78655421]
.0006	CYSTIC FIBROSIS	CFTR, ARG347PRO	[rs77932196]
.0007	CYSTIC FIBROSIS	CFTR, ALA455GLU	[rs74551128]
.0008	CYSTIC FIBROSIS	CFTR, IVS10, G-A, -1	[rs76713772]
.0009	CYSTIC FIBROSIS	CFTR, GLY542TER	[rs113993959]
.0010	CYSTIC FIBROSIS	CFTR, SER549ASN	-
.0011	CYSTIC FIBROSIS	CFTR, SER549ILE	[rs121908755]
.0012	CYSTIC FIBROSIS	CFTR, SER549ARG	[rs121908757], [rs121909005]
.0013	CYSTIC FIBROSIS	CFTR, GLY551ASP	[rs75527207]
.0014	CYSTIC FIBROSIS	CFTR, ARG553TER	[rs74597325]
.0015	CYSTIC FIBROSIS	CFTR, ALA559THR	[rs75549581]

OMIM - Rich Source of
Gene and Phenotype
Data

Exercices sur OMIM :

Explorer le syndromes de Perrault, de l'X fragile, de Turner, de Klinefelter, de Leydig cell hypoplasia

Identifier les syndromes de hypogonadisme hypergonadotrope ou de l'insuffisance ovarienne primitive (IOP) (premature ovarian failure) ou de la diminution de réserve ovarienne DRO) (decreasing ovarian reserve)

Quels sont les phénotypes ?

Quels sont les gènes impliqués ?

Quels anomalies de caryotype ?

Quels variants ?

Pour chaque entrée :

Consulter le « Content » (à gauche) ou les « Links » (à droite)

Attention : queries in english !!

Insuffisance ovarienne précoce

 [About](#) [Statistics ▾](#) [Downloads ▾](#) [Contact Us](#) [MIMmatch](#) [Donate ▾](#) [Help ▾](#) [?](#)

[Q](#) [Options ▾](#) [View Results as: Gene Map Table](#) [Clinical Synopsis](#) [?](#)

Search: "premature ovarian failure" '
Results: 94 entries. [« First](#) | [< Previous](#) | [Next >](#) | [Last »](#)

1: [# 311360. PREMATURE OVARIAN FAILURE 1; POF1](#)
Cytogenetic location: Xq27.3
Matching terms: "premature (ovary ovarian) failure"
[► Phenotype-Gene Relationships](#) [► Phenotypic Series](#) [► ICD+](#) [► Links](#)

2: [% 609993. OSTEOSCLEROSIS WITH ICHTHYOSIS AND PREMATURE OVARIAN FAILURE](#)
Matching terms: "premature (ovary ovarian) failure"
[► ICD+](#) [► Links](#)

3: [# 300510. OVARIAN DYSGENESIS 2; ODG2](#)
[PREMATURE OVARIAN FAILURE 4, INCLUDED; POF4, INCLUDED](#)
Cytogenetic locations: Xp11.22,
Matching terms: "premature (ovary ovarian) failure"
[► Phenotype-Gene Relationships](#) [► Phenotypic Series](#) [► ICD+](#) [► Links](#)

Et suite et links ...

ORPHANET

www.orpha.net

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SC14 INSERM

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Répertoire des centres experts



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Le portail des maladies rares et des médicaments orphelins



Recherche

Outil d'aide au diagnostic

Classifications

Gènes

Handicap

Encyclopédie pour tout public

Encyclopédie pour professionnels

Orphanet Urgences

Sources/procédures

Accueil > Maladies rares > Recherche

Rechercher une maladie rare

perrault

Chercher

(*) Champ obligatoire

- ☒ Nom de maladie ☐ OMIM ☐ Nom de gène
☐ Numéro ORPHA ☐ CIM-10

Accès par phénotype

Autre(s) option(s) de recherche ▼

1 Résultat(s)

ORPHA:2855 [Syndrome de Perrault](#)

Plus d'informations

:: Perrault syndrome

ORPHA2855

Synonym(s):	XX gonadal dysgenesis-deafness syndrome
Prevalence:	<1 / 1 000 000
Inheritance:	Autosomal recessive
Age of onset:	Adolescent Childhood Adult

ICD-10:

OMIM:

UMLS:

MeSH:

MedDRA:

Q87.8

[233400](#) [[↗](#)] [614129](#) [[↗](#)] [614926](#) [[↗](#)]

[615300](#) [[↗](#)] [616138](#) [[↗](#)]

C0685838

-

-

SUMMARY

Perrault syndrome (PS) is characterized by the association of ovarian dysgenesis in females with sensorineural hearing impairment. In more recent PS reports, some authors have described neurologic abnormalities, notably progressive cerebellar ataxia and intellectual deficit.

Prevalence is unknown but 34 patients with PS (28 females and 6 males) from 15 different families have been reported so far. However, the syndrome is probably underdiagnosed: hypogonadism is not a feature in males and in the absence of an affected sister the syndrome remains undetected.

... ..

Mean age at diagnosis is 22 years following presentation with delayed puberty in females with sensorineural deafness. Hearing defects were noted in all but one of the reported cases (mean age at diagnosis of 8 years). The hearing loss is always sensorineural and bilateral but the severity is variable (mild to profound), even in affected patients from the same family. Ovarian dysgenesis has been reported in all female cases but no gonad defects are detected in males. Amenorrhea is generally primary but secondary amenorrhea has also been reported. Delayed growth (height below the third percentile) was reported in half the documented cases. The exact frequency of the neurological abnormalities is unknown, but nine females and two males (16-37 years old) lacking neurological abnormalities have been reported. Neurological signs are progressive and generally appear later in life, however, walking delay or early frequent falls have been noted in young PS patients. Common neurological signs are ataxia, dyspraxia, limited extraocular movements, and polyneuropathy. Some cases with scoliosis have also been reported.

Mutations in the following genes have been excluded: *GJB2* (responsible for the most frequent form of isolated hearing loss), *FOXL2* (involved in premature ovarian failure) and *POLG*, *FRDA*, *AOA1* (implicated in ataxia or ophthalmoplegia). Karyotype is normal. The variability in the presence of the neurological symptoms may indicate that PS is a heterogeneous disease.

Additional information

Further information on this disease

- > Classification(s) (8)
- > Gene(s) (5)
- > Other website(s) (1)

Health care resources for this disease

- > Expert centres (391)
- > Diagnostic tests (54)

Diagnostic tests (54)

- > Patient organisations (60)
- > Orphan drug(s) (1)

Research activities on this disease

- > Research projects (54)
- > Clinical trials (0)
- > Registries/biobanks (29)
- > Networks (29)

Specialised Social Services

- > Eurordis directory [[↗](#)]

Orphanet Reports series

- > Prevalence
- > Orphan drugs in Europe

Mutations in the following genes have been excluded: *GJB2* (responsible for the most frequent form of isolated hearing loss), *FOXL2* (involved in premature ovarian failure) and *POLG*, *FRDA*, *AOA1* (implicated in ataxia or ophthalmoplegia). Karyotype is normal. The variability in the presence of the neurological symptoms may indicate that PS is a heterogeneous disease.

Diagnosis is made on the basis of the clinical signs and further investigations: CT scans revealing that the hearing loss is not associated with temporal bone malformations; hormonal tests revealing hypergonadotropic hypogonadism in females; pelvic examinations revealing absent ovaries or streak gonads, and a very hypoplastic uterus, and neurologic investigations revealing reduced nerve conduction velocities. Cerebral MRI may show a nonspecific white matter hypersignal or cerebellar atrophy.

Turner syndrome is the principle differential diagnosis (see this term).

Transmission of PS is probably autosomal recessive but no gene locus or mitochondrial mutations have been identified to date.

Treatment and follow-up should be multidisciplinary including audiologists, endocrinologists and neurologists. Hearing aids or cochlear implants may be of benefit for the hearing defect.

Life expectancy is normal. Outcome with treatment is very variable, depending on the association with other features, in particular the presence of neurologic disease.

Expert reviewer(s)

Dr Sandrine MARLIN

Last update: September 2008

Orphanet Reports series

- > Prevalence
- > Orphan drugs in Europe

Getting involved /informed

- > Read the newsletter
- > Read [OJRD](#) [↗]
- > Register your activity

Sources/procedures

- > Orphanet ICD10 coding rules [↗]
- > Scientific contributors [↗]
- > Orphanet Operating Procedures [↗]



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Informations complémentaires

Plus d'information sur cette maladie

- [Classification\(s\) \(8\)](#)
- [Gène\(s\) \(6\)](#)
- [Autre\(s\) site\(s\) Internet \(2\)](#)

Services sociaux spécialisés

- [Annuaire Eurordis](#)

Ressources médicales pour cette maladie

- [Centres experts \(391\)](#)
- [Tests diagnostiques \(61\)](#)
- [Associations \(57\)](#)
- [Médicament\(s\) orphelin\(s\) \(1\)](#)

Activités de recherche sur cette maladie

- [Projets de recherche \(47\)](#)
- [Essais cliniques \(0\)](#)
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- [Réseaux \(32\)](#)

Rechercher une maladie rare

HARS2

Chercher

(*) Champ obligatoire

☐ Nom de maladie

☐ OMIM

☒ Nom de gène

☐ Numéro ORPHA

☐ CIM-10

Accès par gène

Autre(s) option(s) de recherche ▼

1 Résultat(s)

> [histidyl-tRNA synthetase 2, mitochondrial - HARS2](#)

:: HARS2 - histidyl-tRNA synthetase 2, mitochondrial

ORPHA268052

Synonyme(s) : *HARSR, histidine tRNA ligase 2, mitochondrial (putative), HO3*

Anciens symbols et noms: *HARSL, histidyl-tRNA synthetase-like*

Type: gene with protein product

Localisation chromosomique: 5q31.3

OMIM: [600783 \[↗\]](#)

HGNC: [4817 \[↗\]](#)

UniProtKB: [P49590 \[↗\]](#)

Genatlas: [HARS2 \[↗\]](#)

Ensembl: [ENSG00000112855 \[↗\]](#)

IUPHAR-DB: -

Reactome: [P49590 \[↗\]](#)

Liste de maladies

- ▶ Mutation germinale causale dans [Syndrome de Perrault](#) ✓

✓ Vérifié

Informations complémentaires

Ressources médicales pour ce gène

- > Tests diagnostiques (4)

Activités de recherche sur ce gène

- > Projets de recherche (0)
- > Registres/bases de données (0)

Search

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Lien sur OMIM

*600783

HISTIDYL-tRNA SYNTHETASE 2; HARS2

Alternative titles; symbols

HISTIDYL-tRNA SYNTHETASE, MITOCHONDRIAL

MITOCHONDRIAL HISRS

HISTIDYL-tRNA SYNTHETASE-LIKE; HARSL

HARS-RELATED GENE; HARSR

HO3

*HGNC Approved Gene Symbol: [HARS2](#)**Cytogenetic location: [5q31.3](#) Genomic coordinates ([GRCh38](#)):**[5:140,691,425-140,699,317](#) (from NCBI)*

Table of Contents for #600783

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Gene-Phenotype Relationships

Location	Phenotype	Phenotype MIM number	Inheritance (in progress)	Phenotype mapping key
5q31.3	?Perrault syndrome 2	614926	AR	3

TEXT

Description

The HARS2 gene encodes a histidyl tRNA synthetase, a highly conserved protein that functions in mitochondria (summary by [Pierce et al., 2011](#)). 

Cloning and Expression

[O'Hanlon et al. \(1995\)](#) identified the HARS2 gene, which they called HO3, oriented head-to-head with the HARS gene ([142810](#)). The deduced 506-amino acid HO3 protein has a calculated molecular mass of 56.9 kD. HO3 shares 72% amino acid identity with HARS. Both proteins contain 3 motifs conserved among class II aminoacyl-tRNA synthetases and 2 signature regions of histidyl-tRNA synthetases. However, HARS and HARS2 have divergent N-terminal domains that

External Links

- [Genome](#)
- [DNA](#)
- [Protein](#)
- [Gene Info](#)
- [Clinical Resources](#)
- [Variation](#)
- [Animal Models](#)
- [Cellular Pathways](#)

Une entrée OMIM en * correspond à un gène

Une entrée OMIM en # correspond à un phénotype

ALLELIC VARIANTS (2 Selected Examples):

[Table View](#)[ClinVar](#)

.0001 PERRAULT SYNDROME 2 (1 family)

HARS2, LEU200VAL [[dbSNP:rs397515410](#)] [[ClinVar](#)]

In affected members of a family of European descent with Perrault syndrome-2 (PRLTS2; [614926](#)), originally reported by [Pallister and Opitz \(1979\)](#), [Pierce et al. \(2011\)](#) identified compound heterozygosity for 2 mutations in the HARS2 gene: a paternally inherited 598C-G transversion in exon 6 resulting in a leu200-to-val (L200V) substitution at a highly conserved residue, and a maternally inherited 1102G-T transversion in exon 10 resulting in a val368-to-leu (V368L; [600783.0002](#)) substitution at a highly conserved residue. The mutations were found by linkage analysis followed by candidate gene

[▶ Gene Info](#)[▶ Clinical Resources](#)[▶ Variation](#)[▶ Animal Models](#)[▶ Cellular Pathways](#)

.0002 PERRAULT SYNDROME 2 (1 family)

HARS2, VAL368LEU - [[ClinVar](#)]

For discussion of the val368-to-leu (V368L) mutation in the HARS2 gene that was found in compound heterozygous state in affected members of a family with Perrault syndrome-2 (PRLTS2; [614926](#)) by [Pierce et al. \(2011\)](#), see [600783.0001](#). 

**OMIM apporte plus
d'informations sur les variants**

REFERENCES

1. Bonnefond, L., Fender, A., Rudinger-Thirion, J., Giege, R., Florentz,

Rechercher un centre expert

perrault

Chercher

(*) Champ obligatoire

- ☒ Consultation spécialisée
- ☐ Conseil Génétique
- ☐ Les deux types de consultation

- ☒ Adultes
- ☐ Enfants
- ☐ Tout

France

☒ Centre officiellement désigné

[Autre\(s\) option\(s\) de recherche](#) ▼

 **FRANCE**
ILE-DE-FRANCE
PARIS

[Centre national de ressources Robert Laplane](#)

Centre de Ressources Expérimental Robert Laplane

Plus
d'informations



 **FRANCE**
AUVERGNE-
RHONE-ALPES
CLERMONT-
FERRAND

[Centre de référence des anomalies du développement et syndromes malformatifs du Sud-Est - Site constitutif](#)

CHU de Clermont-Ferrand - Hôpital d'Estaing

Plus
d'informations



UCSC Browser et Orphanet

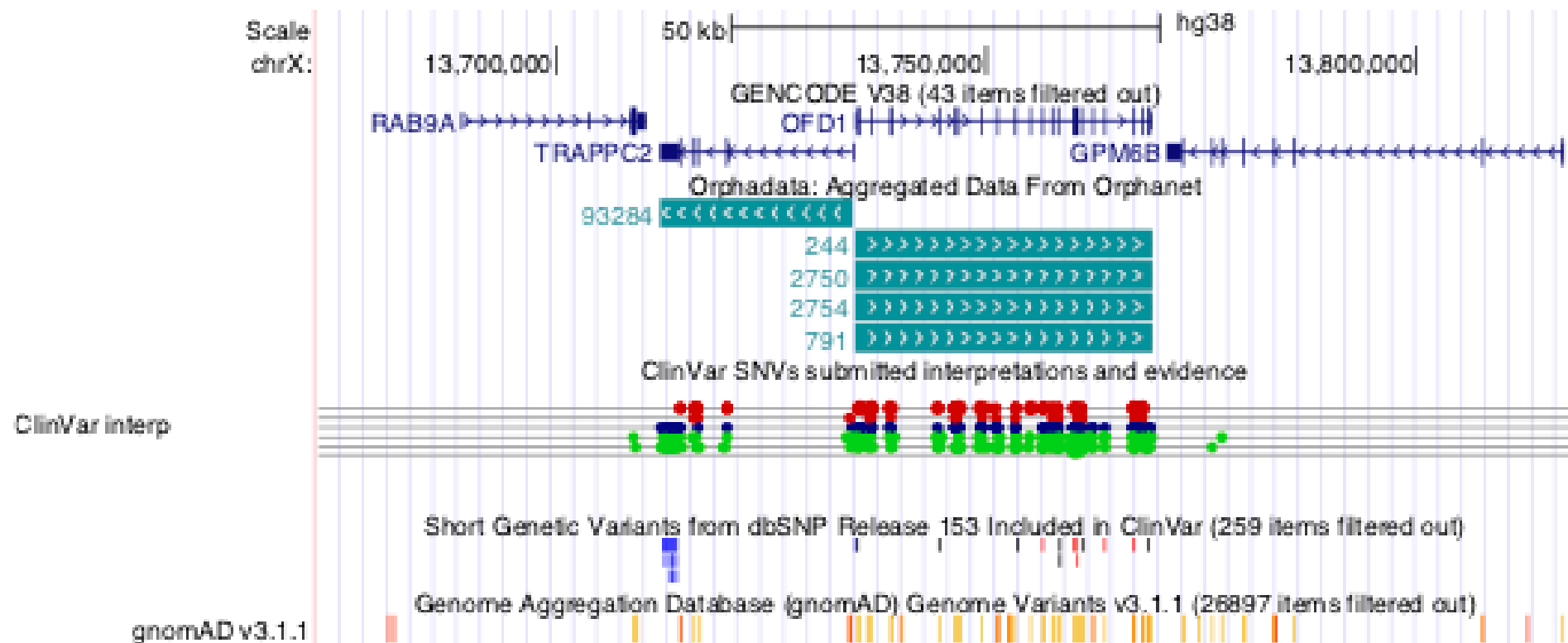
Hello Genome Browser enthusiasts,
We are happy to share another clinical resource for genetic disease analysis, the [Orphadata track](#) from the [Orphanet consortium](#).

This track shows nearly 8000 gene-disease associations on hg19 and hg38. This data track includes information on human disorders and epidemiological information including Human Phenotype Ontology (HPO) disorder name, association type, modes of inheritance, age of first symptoms, and population disease prevalence (if available). For ease of use, we added mouseover information and a variety of filters. See our full announcement here:

<https://genome.ucsc.edu/goldenPath/newsarch.html#111821>

See this track along with a few other popular clinical tracks here:

<https://genome.ucsc.edu/s/dschmelt/Orphadata.Orphanet.hg38>



Free access data from Orphanet
orph^{an}et

orphadata

Home

About Orphadata

Catalogue of products (Academia)

Catalogue of products (Industry)

How are the data produced?

About Orphanet

Sunday, 2 October, 2016

Welcome to Orphadata

The mission of Orphadata is to provide the scientific community with a comprehensive, high-quality and freely-accessible dataset related to rare diseases and orphan drugs, in a reusable format.

For more information on Xml format files, see the [user's guide](#).

See ["How the data are produced"](#)

—

METHODOLOGY

Orphanet provides a comprehensive inventory of rare diseases in Europe, published biannually as a list. Rare diseases registered in Orphanet are defined according to two scopes:

- Every entity is defined by its clinical homogeneity, regardless of its etiology or the number of causing genes identified;
- The rarity is defined according to the European legislation defining a prevalence threshold of not more than 5 affected persons per 10'000 (Regulation (EC) N°141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products, http://ec.europa.eu/health/files/eudralex/vol-1/reg_2000_141/reg_2000_141_en.pdf).

Data collection

As new scientific knowledge arises, the Orphanet rare diseases inventory is updated through the regular addition/update of diseases via two non-exclusive sources: documented sources and/or expert advice.

The scientific knowledge is monitored through:

- A bi-monthly analysis of a defined set of international peer-reviewed scientific journals covering the diversity of medical specialities represented in Orphanet;
- A monthly Medline search algorithm: (nosology[Title] OR classification[Title] OR nomenclature[Title] OR terminology[Title]) AND (rare disease* OR syndrome* OR disorder*);

About Orphanet

Access Orphanet[→]

Contact

FAQ

Freely accessible datasets

Disorders, cross referenced with other nomenclatures

Orphanet classifications

Phenotypes associated with rare disorders **new!**

Disorders with their associated genes

Linearisation of disorders

Orphanet Rare Disease Ontology

Freely-accessible dataset

Orphanet
Rare
Diseases
Ontology
(ORDO)

Disorders,
cross
referenced with
other
nomenclatures

Orphanet
Classifications

SPARQL
ENDPOINT
(beta)

Epidemiological
data

Phenotypes
associated
with rare
disorders

Disorders
with their
associated
genes

Linearisation of
disorders

The dataset is a partial extraction of the data stored in Orphanet, which is also accessible at www.orpha.net for consultation purposes only.

Exercices sur Orphanet : www.orpha.net

Explorer le syndromes de Perrault, de l'X fragile, de Turner, de Klinefelter,
de Leydig cell hypoplasia

Identifier les syndromes de hypogonadisme hypergonadotrope
ou de l'insuffisance ovarienne primitive
ou de la diminution de la réserve ovarienne

Quels sont les phénotypes ?

Quels sont les gènes impliqués ?

Quels anomalies de caryotype ?

A-t-on les mêmes renseignements que sur OMIM ?

Qu apporte de plus ORPHANET ?

Quels centres experts de références en région Bretagne ?

(manque la limitation à la localisation Distance de domicile !!)

Rechercher une maladie rare

Insuffisance ovarienne précoce *

(*) Champ obligatoire

Chercher

☒ Nom de maladie

7 Résultat(s)

ORPHA:243 (Pathologie) [Dysgénésie gonadique 46,XX](#)

Plus d'informations

Mots-clés: Insuffisance ovarienne précoce

ORPHA:95709 (Groupe de pathologies) [Insuffisance ovarienne précoce acquise](#)

Plus d'informations

ORPHA:485382 (Groupe de pathologies) [Insuffisance ovarienne précoce génétique non acquise](#)

Gènes associés au critère « Premature ovarian failure »

Your search resulted in multiple matches. Please select a position:

Gencode Genes

[POF1B \(ENST00000262753.9\)](#) at [chrX:85277396-85379665](#) - Homo sapiens POF1B actin binding protein (POF1B), trans

[NOBOX \(ENST00000644387.2\)](#) at [chr7_KZ208913v1_alt:446710-459685](#) - Homo sapiens NOBOX oogenesis homeobox (NOBOX

[FOXL2 \(ENST00000648323.1\)](#) at [chr3:138944224-138947137](#) - Homo sapiens forkhead box L2 (FOXL2), mRNA. (from Ref

[HFM1 \(ENST00000370425.8\)](#) at [chr1:91260766-91404837](#) - Homo sapiens helicase for meiosis 1 (HFM1), transcript v

[STAG3 \(ENST00000426455.5\)](#) at [chr7:100177563-100214380](#) - Homo sapiens stromal antigen 3 (STAG3), transcript va

[FIGLA \(ENST00000332372.6\)](#) at [chr2:70777310-70790643](#) - Homo sapiens folliculogenesis specific bHLH transcripti

[DIAPH2 \(ENST00000373049.8\)](#) at [chrX:96684712-97469835](#) - Homo sapiens diaphanous related formin 2 (DIAPH2), tra

[PABPC5 \(ENST00000312600.4\)](#) at [chrX:91434839-91438584](#) - Homo sapiens poly(A) binding protein cytoplasmic 5 (PA

[INHA \(ENST00000243786.3\)](#) at [chr2:219572310-219575711](#) - Homo sapiens inhibin subunit alpha (INHA), mRNA. (from

[MSH5 \(ENST00000443040.5\)](#) at [chr6_GL000253v2_alt:3045068-3052473](#) - Homo sapiens mutS homolog 5 (MSH5), transcr

[MSH5 \(ENST00000457917.6\)](#) at [chr6_GL000255v2_alt:2995840-3017438](#) - Homo sapiens mutS homolog 5 (MSH5), transcr

[FMR1 \(ENST00000370475.9\)](#) at [chrX:147911919-147951125](#) - Homo sapiens FMRP translational regulator 1 (FMR1), tr

[SYCE1 \(ENST00000303903.10\)](#) at [chr10:133554290-133565583](#) - Homo sapiens synaptonemal complex central element p

[MSH5 \(ENST00000427735.5\)](#) at [chr6_GL000252v2_alt:2987763-3010450](#) - Homo sapiens mutS homolog 5 (MSH5), transcr

[MSH5 \(ENST00000441395.5\)](#) at [chr6_GL000254v2_alt:3081873-3104565](#) - Homo sapiens mutS homolog 5 (MSH5), transcr

[PCMT1 \(ENST00000464889.7\)](#) at [chr6:149749741-149811419](#) - Homo sapiens protein-L-isoaspartate (D-aspartate) O-m

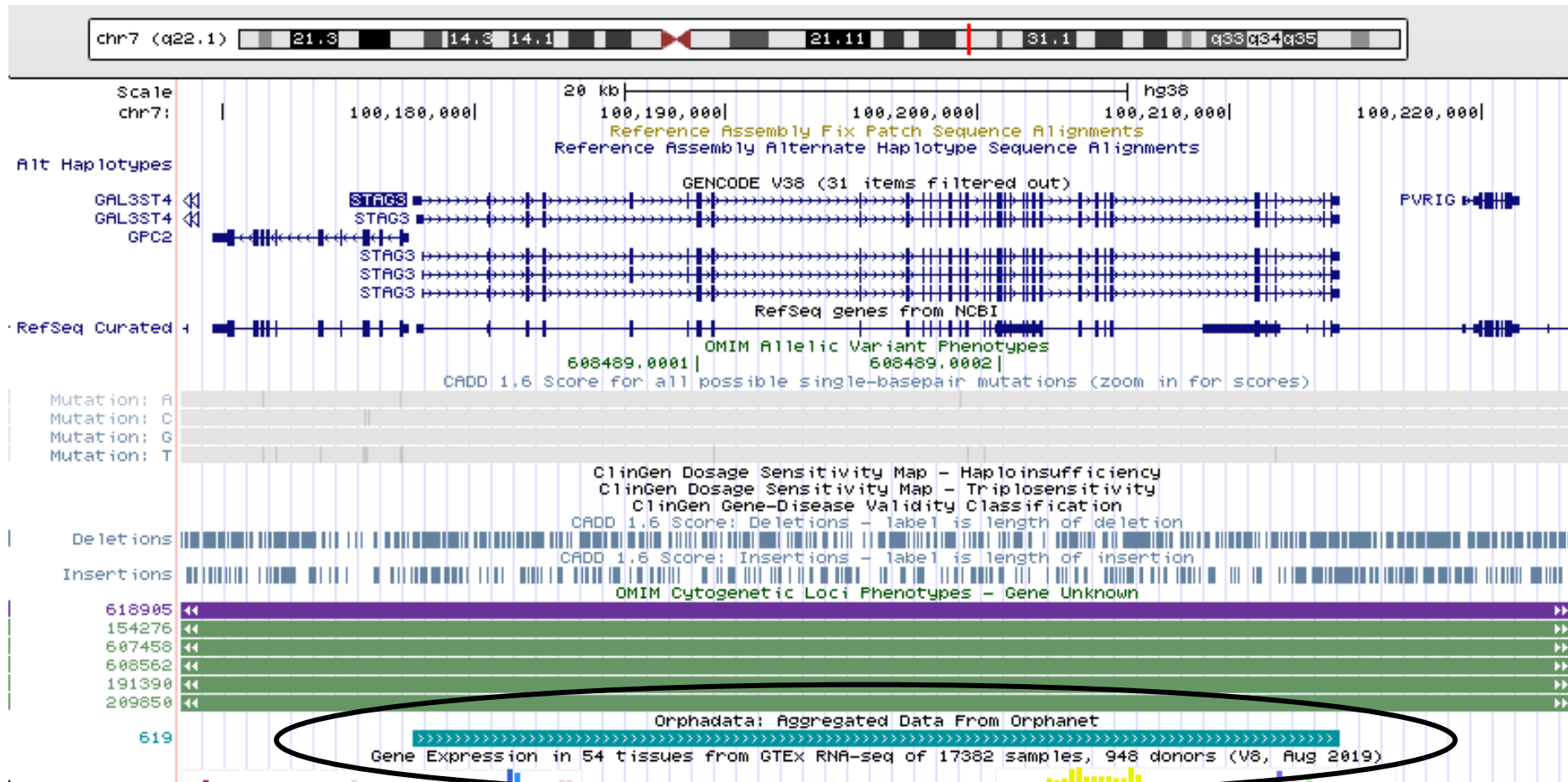
[MSH5 \(ENST00000375755.8\)](#) at [chr6:31739948-31762676](#) - Homo sapiens mutS homolog 5 (MSH5), transcript variant 3

[BMP15 \(ENST00000252677.4\)](#) at [chrX:50910735-50916641](#) - Homo sapiens bone morphogenetic protein 15 (BMP15), mRN

[DACH2 \(ENST00000373125.9\)](#) at [chrX:86148451-86832602](#) - Homo sapiens dachshund family transcription factor 2 (D

[GDF9 \(ENST00000378673.2\)](#) at [chr5:132861181-132866637](#) - Homo sapiens growth differentiation factor 9 (GDF9), t

[NR5A1 \(ENST00000373588.9\)](#) at [chr9:124481236-124507399](#) - Homo sapiens nuclear receptor subfamily 5 group A mem



Localisation de : Aggregated Data from Orphanet