

Genome Database

Genome Databases

- The study of genes and proteins has become an extremely important area in the modern day biology and they are better known as genomics and proteomics
- Genome databases store this information and differently from gene databases the genome databases contain both coding and non-coding inter-genic sequences.

Types of databases

- Many types of databases available for researchers in the field of biology
 - a) Primary sequence databases - for storage of raw experimental data
 - b) Secondary databases - contain information on sequence patterns and motifs
 - c) Organism specific databases and other databases like Saccharomyces genome database, Mouse genome database, Human genome database, European mutant mouse pathology database

Organism-specific genomic databases

| Organism | Database/resource | URL |
|---------------------------------|--|---|
| <i>Escherichia coli</i> | EcoGene EcoCyc (Encyclopedia of <i>E. coli</i> genes and metabolism) Colibri | http://bmb.med.miami.edu/EcoGene/EcoWeb http://ecocyc.pangeasystems.com/ecocyc/ecocyc.html http://genolist.pasteur.fr/Colibri |
| <i>Bacillus subtilis</i> | SubtiList | http://genolist.pasteur.fr/SubtiList |
| <i>Saccharomyces cerevisiae</i> | Saccharomyces Genome Database (SGD) | http://genome-www.stanford.edu/Saccharmyces |
| <i>Plasmodium falciparum</i> | PlasmoDB | http://PlasmoDB.org |
| <i>Arabidopsis thaliana</i> | MIPS <i>Arabidopsis thaliana</i> Database (MAtdB) The Arabidopsis information resource (TAIR) | http://mips.gsf.de/proj/thal/db http://www.arabidopsis.org |
| <i>Drosophila melanogaster</i> | FlyBase | http://flybase.bio.indiana.edu |
| <i>Caenorhabditis elegans</i> | A <i>C. elegans</i> DataBase (ACeDB) | http://www.acedb.org |
| Mouse | Mouse Genome Database (MGD) | http://www.informatics.jax.org |
| Human | OnLine Mendelian Inheritance in Man (OMIM) | http://www.ncbi.nlm.nih.gov/omim |

OMIM

Online Mendelian Inheritance in Man (OMIM)

information on all known mendelian disorders
linked to over 12,000 genes”

- “Started at 1960s by Dr. Victor A. McKusick as a catalog of mendelian traits and disorders”
- Linked disease data
- Links disease phenotypes and causative genes
- Used by physicians and geneticists

INTRODUCTION

- Online Mendelian Inheritance in Man (OMIM) is a continuously updated catalogue of human genes and genetic disorders.
- OMIM focuses primarily on inherited, or heritable, genetic diseases.
- It is considered to be a phenotypic companion to the human genome project.

- During search exclude common words like genes or diseases
- Putting + in front of multiple words.

Inbox x | Fwd: x | Front x | GENC x | Viral x | Down x | BOOK x | omim x | Nucl x | final x | ([]) x | OMIM x

omim.org/search/advanced/entry

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OMIM Advanced Search

Search OMIM...

Relevance Date updated Date created Results per page: 10

Search In:
 MIM Number
 Title
 Text
 Allelic Variants
 HGNC Symbol
 Contributors

Only Records With:
 Allelic Variants
 Clinical Synopsis
 Gene Map Locus

MIM Number Prefix:
 * gene with known sequence
 + gene with known sequence and phenotype
 # phenotype description, molecular basis known
 % mendelian phenotype or locus, molecular basis unknown
 none - other, mainly phenotypes with suspected mendelian basis

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
 Mitochondrial Autosomal Unknown

Dates:
Created From: YYYY/MM/DD To: YYYY/MM/DD

Genome_Databases.pdf Show all

15:45 07-12-2020

OMIM Advanced Search

Relevance Date updated Date created Results per page: 10

Search In:

- MIM Number
- Title
- Text
- Allelic Variants
- HGNC Symbol
- Contributors

Only Records With:

- Allelic Variants
- Clinical Synopsis
- Gene Map Locus

MIM Number Prefix:

- * gene with known sequence
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Chromosome:

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Dates:

Created From: YYYY/MM/DD To: YYYY/MM/DD

OMIM Presentation

Press **Esc** to exit full screen

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5 YEARS
OMIM
Human Genetics Knowledge
for the World

OMIM®

Online Mendelian Inheritance in Man®

An Online Catalog of Human Genes and Genetic Disorders

Updated October 4, 2019

SLC6A1

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

Need help? : [Example Searches](#), [OMIM Search Help](#), [OMIM Video Tutorials](#)

Mirror site : <https://mirror.omim.org>

OMIM is supported by a grant from NHGRI, licensing fees, and generous contributions from people like you.

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SCRIPING 4:11 / 6:06

Scroll for details



Search Results

← → ↻ omim.org/search/?index=entry/locus+score+desc%2C+prefix_sort+desc&start=1&limit=10&search=slc6a1



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slc6a1



Options

View Results at [Gene Map Table](#)

[Clinical Synopsis](#)

Display Highlights

Search: 'slc6a1'

Results: 6 entries

Download As

1: * 137165. SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER, GABA), MEMBER 1; SLC6A1

Cytogenetic location: 3p25.3, Genomic coordinates (GRCh38): 328,000,700-328,001,240

Matching terms: slc6a1

► [Gene-Phenotype Relationships](#) ► [Links](#)

2: † 616421. MYOCLONIC-ATONIC EPILEPSY; MAE

Cytogenetic location: 3p25.3

Matching terms: slc6a1

► [Phenotype-Gene Relationships](#) ► [ICD+](#) ► [Links](#)

3: * 137163. GAMMA-AMINOBUTYRIC ACID RECEPTOR, DELTA; GABRD

Cytogenetic location: 1p36.33, Genomic coordinates (GRCh38): 12,019,344-12,020,757

Matching terms: slc6a1

► [Gene-Phenotype Relationships](#) ► [Links](#)

4: † 600131. EPILEPSY, CHILDHOOD ABSENCE, SUSCEPTIBILITY TO, 1; ECA1

Cytogenetic location: 8q24, Genomic coordinates (GRCh38): 8126,700,000-145,136,626

Matching terms: slc6a1

► [Gene-Phenotype Relationships](#) ► [Phenotypic Series](#) ► [ICD+](#) ► [Links](#)

5: * 607952. SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER, GABA), MEMBER 11; SLC6A11

Cytogenetic location: 3p25.3, Genomic coordinates (GRCh38): 328,804,199-328,940,713

Matching terms: slc6a1

► [Links](#)

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SCREENCAST



MYTIC



Genome_Databases.pdf



Show all

Search OMIM... [Search Icon] Options | Display [Highlights]

***137165**
Table of Contents

- Title
- Gene-Phenotype Relationships
- Text
 - Description
 - Cloning and Expression
 - Gene Structure
 - Gene Function
 - Mapping
 - Molecular Genetics
 - Animal Model
- Allelic Variants
- Table View
- References
- Contributors
- Creation Date
- Edit History

*** 137165**

SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER, GABA), MEMBER 1; SLC6A1

Alternative titles; symbols

**GAMMA-AMINOBUTYRIC ACID TRANSPORTER
GABA TRANSPORTER; GABATR
GABATHG**

HGNC Approved Gene Symbol: SLC6A1

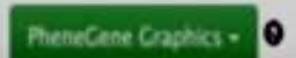
Cytogenetic location: 3p25.3 Genomic coordinates (GRCh38): 3:10,992,733-11,039,248 (from NCBI)

Gene-Phenotype Relationships

| Location | Phenotype | Phenotype MIM number | Inheritance | Phenotype mapping key |
|----------|----------------------------|----------------------|-------------|-----------------------|
| 3p25.3 | Myoclonic-astonic epilepsy | 616421 | AD | 3 |

External Links

- Genome
 - Ensembl
 - NCBI Genome Data Viewer
 - UCSC Genome Browser
- DNA
- Protein
- Gene Info
- Clinical Resources
- Variation
- Animal Models
- Cell Lines
- Cellular Pathways



Search OMIM... [Search] [Options] [Display] [Highlights]

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GABA TRANSPORTER; GABATR
GABATHG

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Gene-Phenotype Relationships

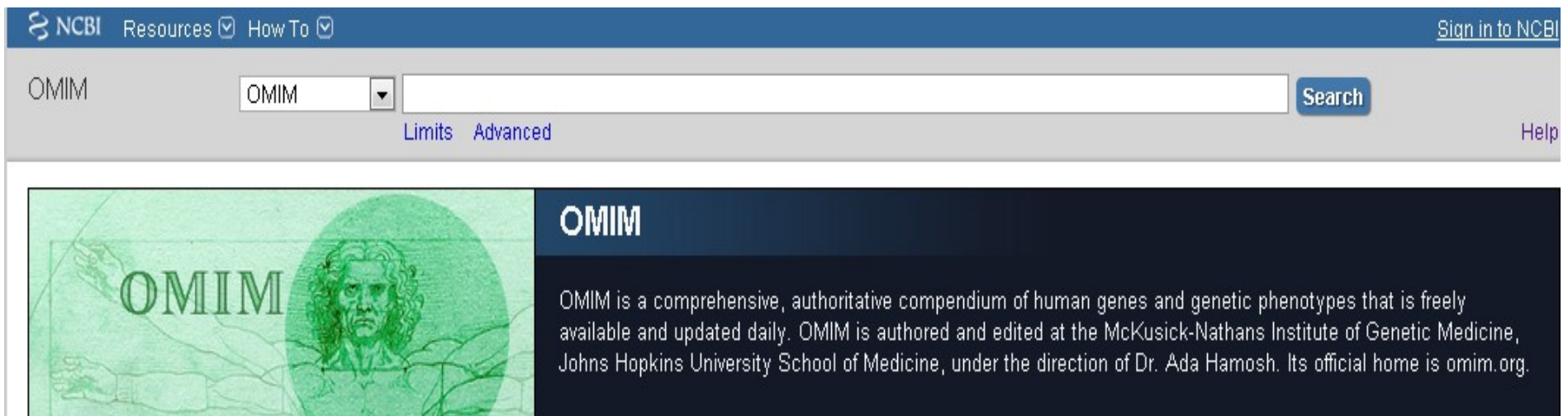
| Location | Phenotype | Phenotype MIM number | Inheritance | Phenotype mapping key |
|----------|---------------------------|----------------------|-------------|-----------------------|
| 3p25.3 | Myoclonic-atonic epilepsy | 616421 | AD | 3 |



PheneGene Graphics

OMIM database

- [Online Mendelian Inheritance in Man \(OMIM\)](#) information on all known mendelian disorders linked to over 12,000 genes”
- “Started at 1960s by Dr. Victor A. McKusick as a catalog of mendelian traits and disorders”
- Linked disease data
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The screenshot shows the top navigation bar of the OMIM website. On the left, there is the NCBI logo and links for "Resources" and "How To". On the right, there is a "Sign in to NCBI" link. Below the navigation bar is a search area with the text "OMIM" on the left, a dropdown menu set to "OMIM", a search input field, and a "Search" button. Below the search field are links for "Limits" and "Advanced". On the far right of this section is a "Help" link. The main content area features a large green-tinted image of Leonardo da Vinci's Vitruvian Man on the left, with the text "OMIM" overlaid. To the right of the image is a dark blue header with the text "OMIM" in white. Below this header is a white text box containing the following description: "OMIM is a comprehensive, authoritative compendium of human genes and genetic phenotypes that is freely available and updated daily. OMIM is authored and edited at the McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, under the direction of Dr. Ada Hamosh. Its official home is omim.org."

OMIM – basic search

- Each search results entry has *, +, # or % symbol

OMIM Entry Statistics:

| Number of Entries in OMIM (Updated 12 September 2013) : | | | | | |
|---|-----------|----------|----------|---------------|--------|
| Prefix | Autosomal | X Linked | Y Linked | Mitochondrial | Totals |
| • Gene description | 13,615 | 664 | 48 | 35 | 14,362 |
| + Gene and phenotype, combined | 112 | 4 | 0 | 2 | 118 |
| # Phenotype description, molecular basis known | 3,578 | 278 | 4 | 28 | 3,888 |
| % Phenotype description or locus, molecular basis unknown | 1,600 | 133 | 5 | 0 | 1,738 |
| Other, mainly phenotypes with suspected mendelian basis | 1,754 | 119 | 2 | 0 | 1,875 |
| Totals | 20,659 | 1,198 | 59 | 65 | 21,981 |

- # entries are the most informative as molecular basis of phenotype – genotype association is known is known
- Will do search on: Ankylosing spondylitis (AS)
- AS characterized by chronic inflammation of spine

OMIM-entries

Ankylosing spondylitis

Search

Sort by: Relevance Date updated

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

Search History: View, Clear

#106300

Entry ID - same as phenotype ID below

SPONDYLOARTHROPATHY, SUSCEPTIBILITY TO, 1; SPDA1

Alternative titles; symbols

ANKYLOSING SPONDYLITIS, SUSCEPTIBILITY TO
MARIE-STRUMPELL SPONDYLITIS
BECHTEREW SYNDROME

Links to other databases

Table of Contents - #106300

External Links:

Clinical Resources

Animal Models

Cellular Pathways

Centers for Mendelian Genomics

Associated gene

Phenotype ID

Gene ID

Phenotype Gene Relationships

| Location | Phenotype | Phenotype MIM number | Gene/Locus | Gene/Locus MIM number |
|----------|---|----------------------|------------|-----------------------|
| 6p21.33 | {Spondyloarthritis, susceptibility to, 1} | 106300 | HLA-B | 142830 |

Phenotypic Series

related phenotypes

Clinical Synopsis

detailed description of the phenotype divided into categories

TEXT

A number sign (#) is used with this entry because of evidence that susceptibility to ankylosing spondylitis can be conferred by variation in the HLA-B27 allele (142830.0001) on chromosome 6p21.3.

Description

Spondyloarthritis (SpA), one of the commonest chronic rheumatic diseases, includes a spectrum of related

OMIM Gene ID -entries

+142830

MAJOR HISTOCOMPATIBILITY COMPLEX, CLASS I, B; HLA-B

⇒ Full name of the gene

Alternative titles; symbols

HLA-B HISTOCOMPATIBILITY TYPE

Other entities represented in this entry:

ABACAVIR HYPERSENSITIVITY, SUSCEPTIBILITY TO, INCLUDED

SYNOVITIS, CHRONIC, SUSCEPTIBILITY TO, INCLUDED

DRUG-INDUCED LIVER INJURY DUE TO FLUCLOXACILLIN, INCLUDED

HGNC Approved Gene Symbol: HLA-B

Cytogenetic location: 6p21.33 Genomic coordinates (GRCh37): 6:31,321,648 - 31,324,988 (from NCBI)

Link to other databases to
obtain DNA or protein sequences and
any other information



- Table of Contents - +142830
- External Links:
- Genome
- DNA
- Protein
- Gene Info
- Clinical Resources
- Variation
- Animal Models
- Cellular Pathways

Centers for Mendelian Genomics

Gene Phenotype Relationships

| Location | Phenotype | Phenotype MIM number |
|----------|---|----------------------|
| 6p21.33 | {Abacavir hypersensitivity, susceptibility to} | |
| | {Drug-induced liver injury due to flucloxacillin} | |
| | {Spondyloarthropathy, susceptibility to, 1} | 106300 |
| | {Stevens-Johnson syndrome, susceptibility to} | 608579 |
| | {Synovitis, chronic, susceptibility to} | |
| | {Toxic epidermal necrolysis, susceptibility to} | 608579 |

Other phenotypes
associated with
the gene

TEXT

For background information on the major histocompatibility complex (MHC) and human leukocyte antigens

OMIM-Finding disease linked genes

- Read the report of given top gene linked phenotype
 - Mapping – Linkage heterogeneity section

Mapping

Gu et al. (2009) conducted a genomewide scan followed by fine mapping analysis in a 4-generation Han Chinese family with ankylosing spondylitis and obtained a maximum lod score of 4.02 at D6S273 (theta = 0.0) on chromosome 6, verifying the HLA-B locus.

Linkage Heterogeneity

To identify major loci controlling clinical manifestations of AS, Brown et al. (2003) performed genomewide linkage analysis on 188 affected sib-pair families containing 454 affected individuals. Heritabilities of the traits studied were as follows: age at symptom onset, 0.33 ($p = 0.005$); disease activity assessed by the Bath Ankylosing Spondylitis Disease Activity Index (BASDAI), 0.49 ($p = 0.0001$); and functional impairment assessed by the Bath Ankylosing Spondylitis Functional Index (BASFI), 0.76 ($p = 0.0000001$). No linkage was observed between the MHC and any of the traits studied. Significant linkage (lod = 4.0) was observed between a region on chromosome 18p and the BASDAI. Age at symptom onset showed suggestive linkage to chromosome 11p (lod = 3.3). Maximum linkage with the BASFI was seen at chromosome 2q (lod = 2.9; see SPDA3, new). Brown et al. (2003) concluded that these clinical manifestations are largely determined by a small number of genes not encoded within the MHC.

In a multistage study involving 12,701 SNPs and patients with autoimmune diseases, including ankylosing spondylitis, the Wellcome Trust Case Control Consortium and the Australo-Anglo-American Spondylitis Consortium (2007) identified significant association with SNPs in the ARTS1 gene (ERAP1; 606832) (combined results, $p = 1.2 \times 10^{-8}$ to 3.4×10^{-10}) on chromosome 5q15. Association was also found with SNPs in the IL23R gene (607562) on chromosome 1p31.3: in combined analysis, the strongest association was at rs11209032 (odds ratio, 1.3; $p = 7.5 \times 10^{-9}$). The association remained strong when only individuals who self-reported as not having inflammatory bowel disease (see IBD17, 612261) were considered, and was still strongest at rs11209032 ($p = 6.9 \times 10^{-7}$).

- Go back to the original results
 - Previously seen entry *607562 – IL23R

Introduction to TAIR

- The Arabidopsis Information Resource:
 - www.arabidopsis.org
- Curation at TAIR
 - Gene structure annotations
 - Gene / protein functional annotations
 - *Partner with the PMN*: Metabolic pathway curation
- External resource integration at TAIR
 - Data sets
 - Biological materials
 - Other databases

External resource integration at TAIR

- Data sets (viewable within TAIR)
 - Arabidopsis EST sequences
 - Other plant EST sequences (selected dicots and monocots)
 - Arabidopsis and Brassica cDNAs
 - non-coding RNAs
 - Alternative gene predictions
 - Transcript expression data
 - Peptide expression data
 - Epigenetic features
 - Ecotype-specific polymorphisms
 - Promoter elements
 - Post-translational modifications
 - Interaction partners
 - Orthologs
 - Publications
- Biological materials (provided by Arabidopsis Biological Resource Center-ABRC)
 - Seed stocks
 - DNA resources



tair

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- Microarray Experiment
- Microarray Expression
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- Polymorphisms/Alleles
- Proteins
- Protocols
- Publication
- Seed/Germplasm
- Textpresso Full Text

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- Seqviewer
- Mapviewer
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- N-Browse
- BLAST
- WU-BLAST
- FASTA
- Patmatch
- Motif Analysis
- VxInsight
- Java Tree View
- Bulk Data Retrieval
- Chromosome Map Tool
- Restriction Analysis
- Gene Symbol Registry
- Textpresso Full Text

base of genetic and molecular
 a available from TAIR
 ure, gene product information,
 aps, genetic and physical
 earch community. Gene
 ublished research literature
 1-2 times per year using
 issions of new and updated
 s to other Arabidopsis

iversity collects, reproduces,
is thaliana and related
 egrated into TAIR.

nce Department of
 ce Foundation.



- Gene
- Google TAIR website
- Metabolic Pathways
- Textpresso full text
- Exact name search
- 5' or 3' end
- Gene
- Person or lab
- Ecotype
- Gene
- Germplasm
- Keyword
- Library
- Marker
- Polymorphisms
- Protein
- Seed stock
- SNP stock
- Vector

Bre

TAIR 2010

Due to usage slow

peak use times (weekdays
 12:00-16:00 GMT) in May and
 June. If the site is slow please
 return during nonpeak hours.
**Please avoid pressing the
 reload button or resubmitting
 the same URL if you don't get
 a response; this just
 overloads the server with
 more requests for data.** We
 will migrate to faster servers in
 July, this should eliminate the
 problems. IMPORTANT: please
 do not run scripts to collect
 data from TAIR pages, contact
 us instead for an ftp file.

Plant Biology 2010
 Bringing together the global community of plant biologists.
Montreal
 July 31 - August 4, 2010

**ASPB Early Bird Registration
 Deadline [April 30, 2010]**
 The Early Bird Registration
 Deadline of this year's Plant
 Biology Meeting in Montreal is
 fast approaching! **Register** by
 Friday, May 7th.

Accessing data and resources at TAIR

- Gather information about gene(s) of interest
 - Locus page
 - GBrowse
 - NBrowse (interaction viewer)
 - Synteny viewer

THANK

YOU