# Literature databases

# OMIM

## Online Mendelian Inheritance in Man

## OMIM

**OMIM is a database that catalogues all the known diseases with a genetic component, and—when possible—links them to the relevant genes in the human genome and provides references for further research and tools for genomic analysis of a catalogued**

"Mendelian inheritance" refers to the transmission of inherited characters from generation to generation through the transmission of genes.

# OMIM history

**1960:** database start by Dr. Victor A. McKusick as a catalog of mendelian traits and disorders, entitled Mendelian Inheritance in Man (MIM).

**1966-1998:** Twelve book editions of MIM

**1985:** Online version, OMIM, created by a collaboration between the National Library of Medicine and the William H. Welch Medical Library at Johns Hopkins. It was made generally available on the internet starting in 1987.

**1995:** OMIM was developed for the World Wide Web by NCBI, the National Center for Biotechnology Information.

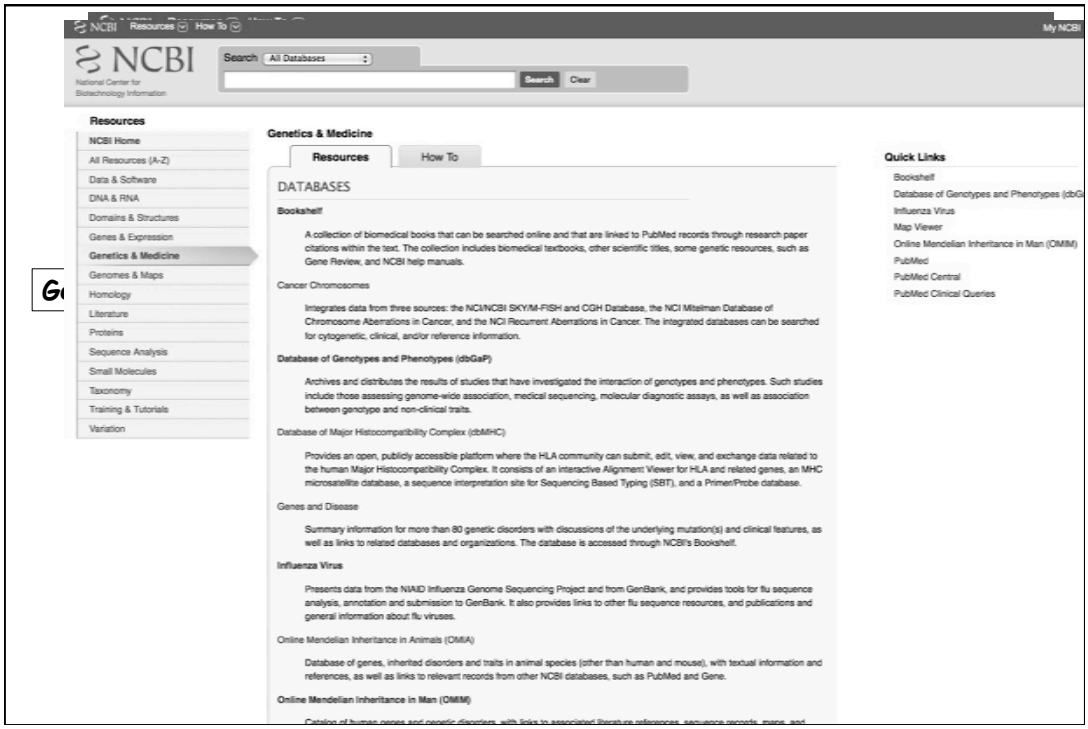
**OMIM Statistics for August 14, 2010**

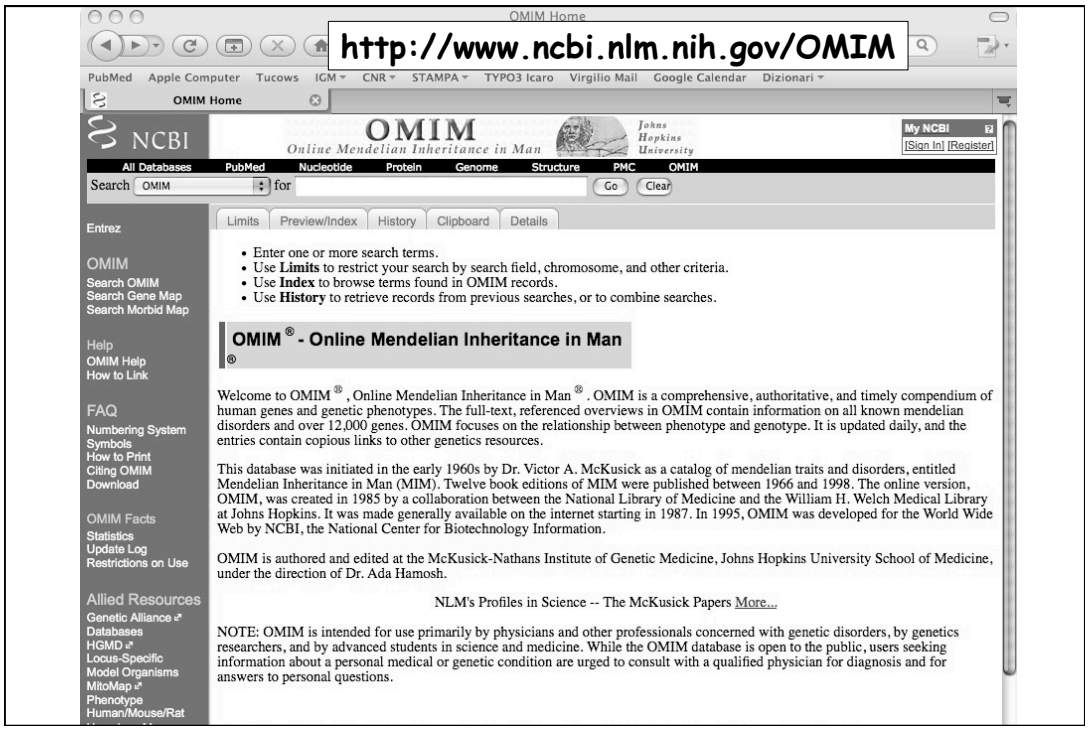
**Number of Entries**

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

**Synopsis of the Human Gene Map**

| Chr. | Loci | Chr. | Loci | Chr. | Loci |
|---|---|---|---|---|---|
| 1 | 1222 | 9 | 477 | 17 | 730 |
| 2 | 798 | 10 | 461 | 18 | 180 |
| 3 | 662 | 11 | 780 | 19 | 789 |
| 4 | 480 | 12 | 656 | 20 | 312 |
| 5 | 593 | 13 | 233 | 21 | 145 |
| 6 | 749 | 14 | 387 | 22 | 313 |
| 7 | 553 | 15 | 375 | X | 700 |
| 8 | 453 | 16 | 491 | Y | 45 |
| Total number of loci: **12584** | | | | | |


NCBI
Resources
How To
My NCBI
Search
All Databases
Search
Clear
Resources
NCBI Home
All Resources (A-Z)
Data & Software
DNA & RNA
Domains & Structures
Genes & Expression
Genetics & Medicine
Genomes & Maps
Homology
Literature
Proteins
Sequence Analysis
Small Molecules
Taxonomy
Training & Tutorials
Variation
Genetics & Medicine
Resources
How To
DATABASES
Bookshelf
A collection of biomedical books that can be searched online and that are linked to PubMed records through research paper citations within the text. The collection includes biomedical textbooks, other scientific titles, some genetic resources, such as Gene Review, and NCBI help manuals.
Cancer Chromosomes
Integrates data from three sources: the NCI/NCBI SKY/M-FISH and CGH Database, the NCI Mitelman Database of Chromosome Aberrations in Cancer, and the NCI Recurrent Aberrations in Cancer. The integrated databases can be searched for cytogenetic, clinical, and/or reference information.
Database of Genotypes and Phenotypes (dbGaP)
Archives and distributes the results of studies that have investigated the interaction of genotypes and phenotypes. Such studies include those assessing genome-wide association, medical sequencing, molecular diagnostic assays, as well as association between genotype and non-clinical traits.
Database of Major Histocompatibility Complex (dbMHC)
Provides an open, publicly accessible platform where the HLA community can submit, edit, view, and exchange data related to the human Major Histocompatibility Complex. It consists of an interactive Alignment Viewer for HLA and related genes, an MHC microsatellite database, a sequence interpretation site for Sequencing Based Typing (SBT), and a Primer/Probe database.
Genes and Disease
Summary information for more than 80 genetic disorders with discussions of the underlying mutation(s) and clinical features, as well as links to related databases and organizations. The database is accessed through NCBI's Bookshelf.
Influenza Virus
Presents data from the NIAID Influenza Genome Sequencing Project and from GenBank, and provides tools for flu sequence analysis, annotation and submission to GenBank. It also provides links to other flu sequence resources, and publications and general information about flu viruses.
Online Mendelian Inheritance in Animals (OMIA)
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)
Quick Links
Bookshelf
Database of Genotypes and Phenotypes (dbG
Influenza Virus
Map Viewer
Online Mendelian Inheritance in Man (OMIM)
PubMed
PubMed Central
PubMed Clinical Queries

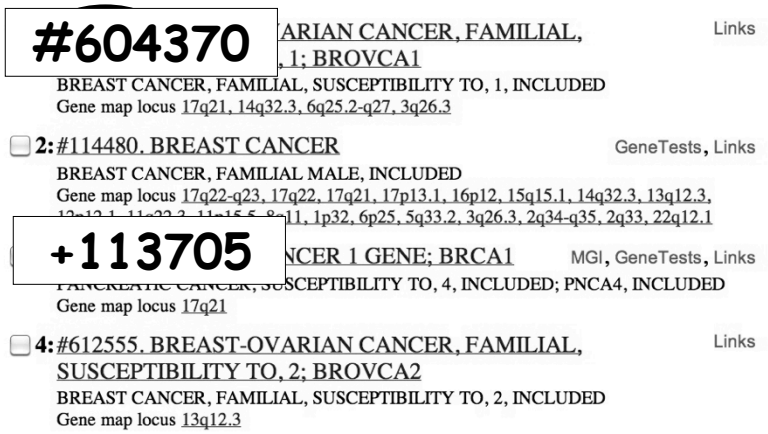

http://www.ncbi.nlm.nih.gov/OMIM
OMIM Home
PubMed
Apple Computer
Tucows
IGM
CNR
STAMPA
TYPO3 Icaro
Virgilio Mail
Google Calendar
Dizionari
NCBI
OMIM
Online Mendelian Inheritance in Man
Johns Hopkins University
My NCBI
[Sign In] [Register]
All Databases
PubMed
Nucleotide
Protein
Genome
Structure
PMC
OMIM
Search
OMIM
for
Go
Clear
Limits
Preview/Index
History
Clipboard
Details
Entrez
OMIM
Search OMIM
Search Gene Map
Search Morbid Map
Help
OMIM Help
How to Link
FAQ
Numbering System
Symbols
How to Print
Citing OMIM
Download
OMIM Facts
Statistics
Update Log
Restrictions on Use
Allied Resources
Genetic Alliance
Databases
HGMD
Locus-Specific
Model Organisms
MitoMap
Phenotype
Human/Mouse/Rat
Enter one or more search terms.
Use Limits to restrict your search by search field, chromosome, and other criteria.
Use Index to browse terms found in OMIM records.
Use History to retrieve records from previous searches, or to combine searches.
OMIM ® - Online Mendelian Inheritance in Man ®
Welcome to OMIM ®, Online Mendelian Inheritance in Man ®. OMIM is a comprehensive, authoritative, and timely compendium of human genes and genetic phenotypes. The full-text, referenced overviews in OMIM contain information on all known mendelian disorders and over 12,000 genes. OMIM focuses on the relationship between phenotype and genotype. It is updated daily, and the entries contain copious links to other genetics resources.
This database was initiated in the early 1960s by Dr. Victor A. McKusick as a catalog of mendelian traits and disorders, entitled Mendelian Inheritance in Man (MIM). Twelve book editions of MIM were published between 1966 and 1998. The online version, OMIM, was created in 1985 by a collaboration between the National Library of Medicine and the William H. Welch Medical Library at Johns Hopkins. It was made generally available on the internet starting in 1987. In 1995, OMIM was developed for the World Wide Web by NCBI, the National Center for Biotechnology Information.
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.
NLM's Profiles in Science -- The McKusick Papers More...
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.

# BASIC SEARCH IN OMIM

**How many entries regarding cancer are present in OMIM??**

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

# Accession codes in OMIM




**Each entry has an unique ID formed by a six-digit number (MIM number) preceded by a symbol.**

# MIM number "translation"

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

# MIM number prefix "translation"

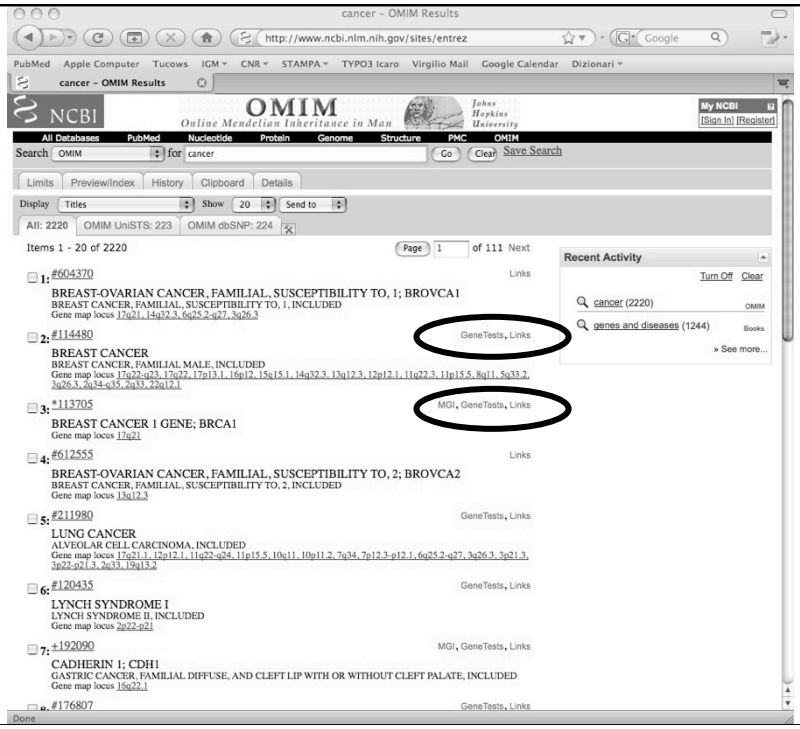
* An **asterisk** indicates a gene of known sequence.

\# A number symbol an () indicates that it is a descriptive entry, usually of a phenotype. The reason for the use of the #-sign is given in the first paragraph of the entry. Discussion of any gene(s) related to the phenotype resides in another entry(ies) as described in the first paragraph.

\+ A plus sign indicates that the entry contains the description of a gene of known sequence and a phenotype.

% A percent sign indicates that the entry describes a confirmed mendelian phenotype or phenotypic locus for which the underlying molecular basis is not known.

No symbol generally indicates a description of a phenotype for which the mendelian basis, although suspected, has not been clearly established or that the separateness of this phenotype from that in another entry is unclear.



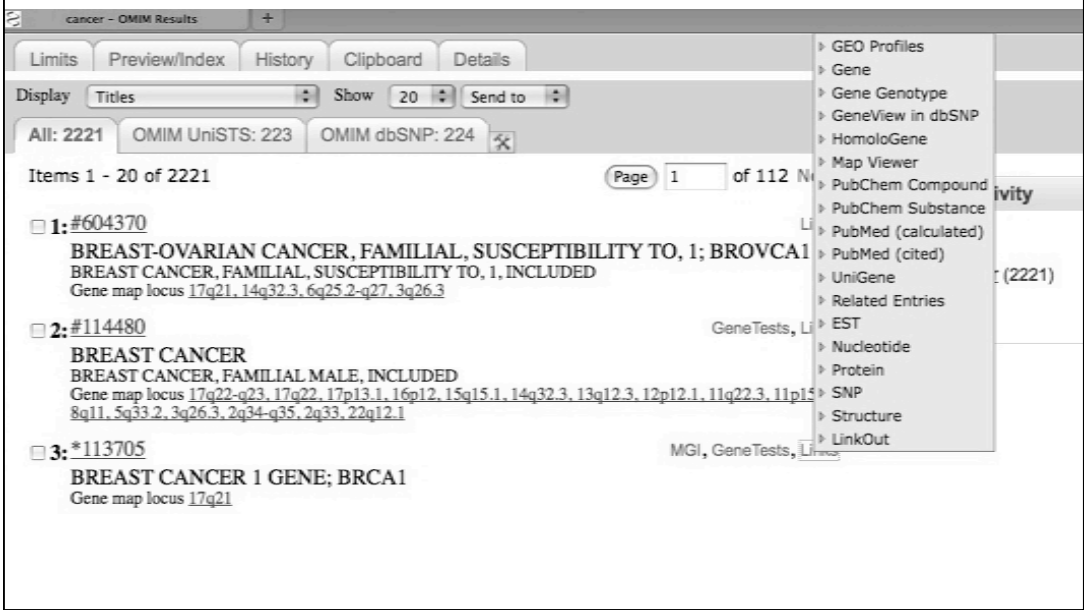

GENETests promotes the appropriate use of genetic services in patient care and personal decision making, by providing current, authoritative information on genetic testing and its use in diagnosis, management, and genetic counseling.

## MGI Mouse Genome Informatics

MGI is the international database resource for the laboratory mouse, providing integrated genetic, genomic, and biological data to facilitate the study of human health and disease.

Mouse Genome Database (MGD) Project
Gene Expression Database (GXD) Project
Mouse Tumor Biology (MTB) Database Project
Gene Ontology (GO) Project at MGI
MouseCyc Project at MGI

http://www.informatics.jax.org/




# ADVANCED SEARCH IN OMIM

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

# MARFAN SYNDROME

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

## Genes & Expression

# Genes and Diseases

*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.

From *Genes and Disease* you reach many online related resources with free and full access.

For example:

a) visit the human genome to see the location of the genes implicated in each disorder

b) find related gene sequences in different organisms

c) look in other books in the NCBI Bookshelf.

Currently over 80 genetic disorders have been summarized, and the content of *Genes and Disease* is continually growing.

## Marfan Syndrome

http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gnd

## Where to study

**OMIM Help**
**http://www.ncbi.nlm.nih.gov/Omim/omimhelp.html**

**OMIM FAQ**
**http://www.ncbi.nlm.nih.gov/Omim/omimfaq.html**

# NORRIE DISEASE