

OMIM

Online Mendelian Inheritance in Man

<http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM>

Introduction

This database is a catalog of human genes and genetic disorders, with links to literature references, sequence records, maps and related databases. Based on the book *Mendelian Inheritance in Man*, the database contains expert reviews of the evidence regarding human genetic diseases in humans. The online version is updated daily.

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 is incorporated into NCBI's Entrez system and can be queried using the same approach as the other Entrez databases such as PubMed and GenBank. This handout highlights features specific to OMIM. To learn the mechanics of searching the Entrez system, please register for one of our PubMed or InfoHub courses.

The Limits Page

Like all Entrez system databases, OMIM has a set of limits that can be used to improve your search. Click on the word "Limits" under the search entry box to view the Limits page.

1. Searches can be limited to specific database fields including:

- Title
- MIM Number
- Allelic Variants
- Text
- References
- Clinical Synopsis
- Gene Map Disorder
- Contributors

The complete list of Fields can be accessed through the pop-up menu on the Preview/Index page.

2. Chromosome number:

To confine your search to a specific chromosome number, simply click in the box beside the appropriate chromosome #. You may choose more than one.

If no boxes are checked, the system will search all OMIM records (both mapped and unmapped).

If you select multiple chromosomes, the system will retrieve records that are on any of those chromosomes.

To retrieve entries that do not have a defined chromosomal location, check unknown.

3. Records mapped to a specific cytogenetic band:

If you want to retrieve records that have been mapped to a specific cytogenetic band, search the Gene Map field using the pop-up search field menu on the Preview/Index page.

4. Records with certain attributes:

If desired, you can retrieve "Only records with" certain attributes, such as:

- Allelic Variants
- Clinical Synopsis
- Mini-MIM (A condensed OMIM report)
- Gene Map Locus

5. The Limits Page:

The Limits page presents check boxes for the most commonly used attributes.

MIM Prefixes

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

A number symbol (#) before an entry number indicates that it is a descriptive entry, usually of a phenotype, and does not represent a unique locus. 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 (+) before an entry number indicates that the entry contains the description of a gene of known sequence and a phenotype.

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

No symbol before an entry number 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.

A caret symbol (^) before an entry number means the entry no longer exists because it was removed from the database or moved to another entry as indicated.

OMIM Gene Map

The OMIM Gene Map provides a table of OMIM data, sorted by MIM location in the chromosomes. It lists genes from the p telomere of chromosome 1 through q telomere of chromosome 22, followed by genes on the X and Y chromosomes (also in the pter and qtel order). Only OMIM entries that have a known cytogenetic location are included. The OMIM Gene Map is available on the web and as an FTP'able file (<ftp://ncbi.nlm.nih.gov/repository/OMIM/genemap>). Use the Gene Map when you know the location or identity of a gene and wish to find its function or relationship to disease.

The web version of the OMIM Gene Map can be searched by gene symbol (e.g., "SOD1"), chromosomal location (e.g., "5", "1pter", "Xq"), or by disorder keyword (e.g., "alzheimer"). Capitalize X and Y to search for those chromosomes. You can specify as few characters as you like. OMIM will go to the first location that starts with the characters you give. The "Find Next" button can then be used to find subsequent instances of the term.

The OMIM Gene Map displays 20 entries at a time. You can move up or down the map, also 20 entries at a time. If a gene has been localized only to a chromosome number, rather than to a specific cytogenetic band, that entry is usually shown at the end of the genes on that chromosome.

The OMIM Gene Map data have been included in the Genes_Cytogenetic map of the Entrez Map Viewer, which presents a graphical display of various cytogenetic, genetic linkage, sequence, radiation hybrid, and other maps. When viewing a chromosome, use the "Display Settings" option to select the Genes_Cytogenetic map. The Map Viewer Help document provides additional information about how to use the viewer.

OMIM Morbid Map

The OMIM Morbid Map is an alphabetical list of diseases described in OMIM and their corresponding cytogenetic locations. The OMIM Morbid Map is available on the web and as an FTP'able file (<ftp://ncbi.nlm.nih.gov/repository/OMIM/morbidmap>). Use the morbid map to go from diseases to implicated genes.

The web version of the OMIM Morbid Map can be searched by gene symbol (e.g., "SOD1"), chromosomal location (e.g., "1pter", "Xq"), or by disorder keyword (e.g., "alzheimer"). You must capitalize "X" and "Y" to search for those chromosomes. You can also search for a number, such as "5". However, that number might appear in a column other than Location. The search system will take you to the first instance of your search term in the file, and will display the 20 entries in that region of the file. You can then scroll up and down the file 20 entries at a time. (As is true for the OMIM Gene Map, the Morbid Map is a single file.) Or, you can use the "Find Next" button to jump to the next instance of the search term.

The Morbid Map is also displayed graphically in the Entrez Map Viewer, showing the disease genes in positional order. When viewing a chromosome, use the "Display Settings" option to select the Morbid map. The Map Viewer Help document provides additional information about how to use the viewer.

Links

Each OMIM record can be linked to a variety of related resources.

Curated Links. These appear in the blue sidebar of the entry, beneath its table of content. For example: UniGene, GenBank, Protein, RefSeq.

LinkOut Links. These link to related entries in other (non-Entrez) NCBI databases, and to related resources maintained by other organizations.

Additional Help

Further information on the use of OMIM is available in the OMIM Help Document: <http://www.ncbi.nlm.nih.gov/Omim/omimhelp.html>, by contacting the NCBI, or by contacting the Eskin Biomedical Library (6-6255).