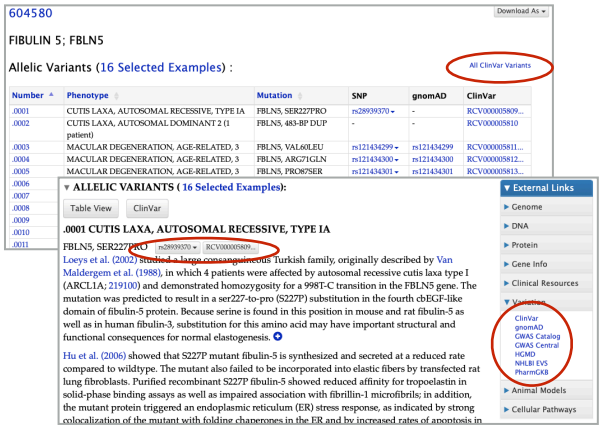
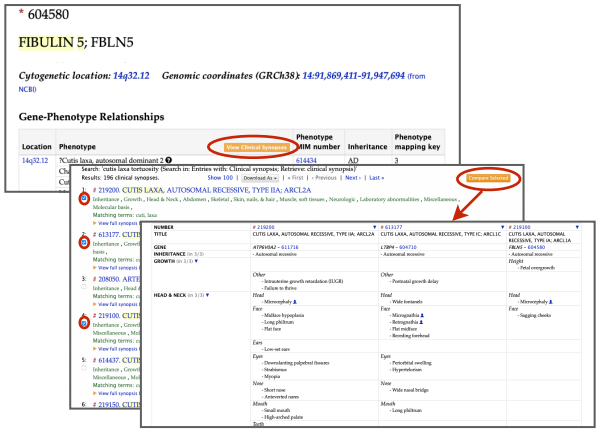
OMIM Allelic Variants

Mutations are cataloged in OMIM in the Allelic Variants section of gene entries. Only select mutations are included. Selection criteria 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 particular common disorders. To see more variants in a gene, follow links in OMIM to ClinVar, gnomAD, and many other variant resources.



Side-by-side Clinical Synopsis viewer



Compare clinical features among phenotypes side-by-side by selecting entries from a Clinical Synopsis Quick View page, from each Phenotypic Series page, from the link within the Gene-Phenotype Relationship table in gene entries with more than one phenotype, and at the top of the Phenotype MIM# column in gene map table view. Brief video tutorials explain this and other strategies for optimizing searches and using MIMmatch. These tutorials and other help are available from the “Help” menu option at the top of every OMIM.org page.

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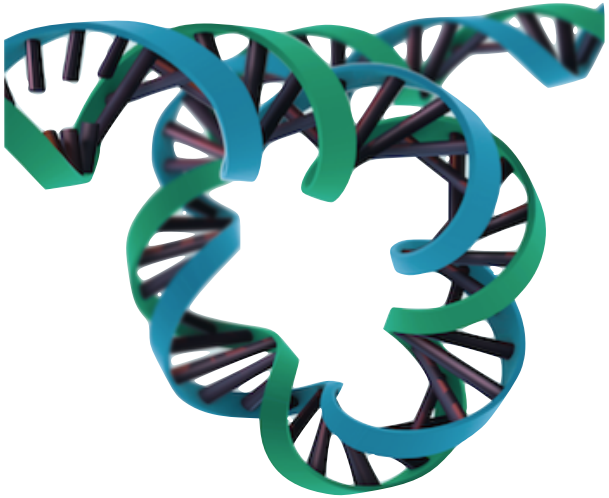
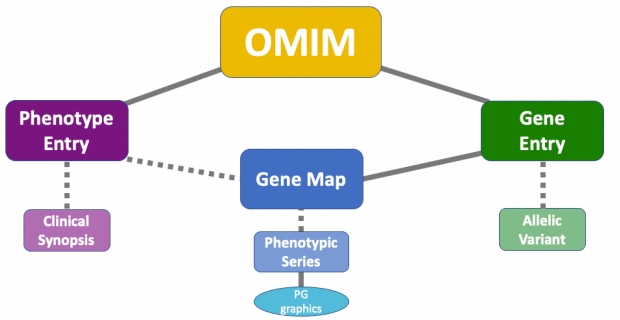
Human Genetics Knowledge for the World



omim.org

OMIM® (Online *Mendelian Inheritance in Man)* is a continuously updated authoritative compendium of human genes and genetic phenotypes (disorders and traits) with full-text, referenced overviews of over 8,800 phenotypes and over 17,200 genes. OMIM focuses on the relationships between phenotypes and genes. In addition to the descriptive entries, **OMIM.org** provides additional unique displays of information including the Clinical Synopses, Gene Map, Phenotypic Series, and Phene-Gene Graphics.



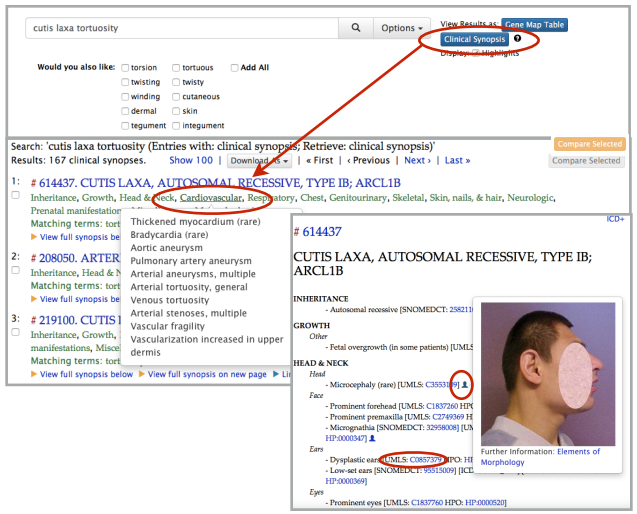
Information framework: Dashed lines indicate that not all genes have allelic variants; not all phenotypes are mapped; not all phenotypes have Clinical Synopses; and mapped phenotypes are not necessarily part of a Phenotypic Series.

Essential diagnostic tool

Clinicians search OMIM using clinical features to facilitate diagnosis of patients.

Resource for disease gene discovery

OMIM connects clinical features of a phenotype with the molecular biology of genes and their variants.



Searching clinical features

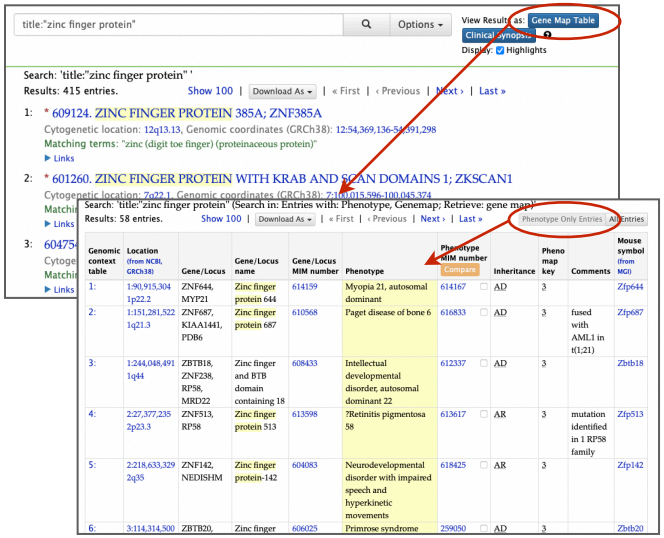
Searching on clinical features is enhanced by a thesaurus, which allows you to select additional terms to include in your search. A quick view of clinical synopses, currently available for most phenotypes, is obtained from a button next to the search box. Placing your mouse over an anatomical category reveals the underlying features. From the full view of a synopsis, mousing over the EoM link (👤) reveals the Elements of Morphology picture representation of the term. There is also an optional display of the feature identifiers from UMLS, SNOMED CT, HPO, and others.

Human- and machine-readable formats

OMIM information is structured and mapped to resources such as HPO, SNOMED CT, and Orphanet. All of this is available through the API.

Stay connected with MIMmatch

Enroll to follow updates to genes and phenotypes of interest to you, stay current on new disease-gene relationships, find other scientists with similar interests.



Easy navigation between genes and phenotypes

OMIM provides various views of gene and phenotype relationships. Finding the phenotypes associated with a particular class of gene can be as easy as searching on the class of gene, for example “zinc finger protein”, and selecting the gene map table link to the right of the search box. From the gene map table, select “phenotype only entries” to condense the table to just entries with phenotypes.