

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.

604580
FIBULIN 5; FBLN5

Allelic Variants (16 Selected Examples) :

Number	Phenotype	Mutation	SNP	gnomAD	ClinVar
.0001	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE IA	FBLN5, SER227PRO	rs2899970	-	RCV00005809..
.0002	CUTIS LAXA, AUTOSOMAL DOMINANT 2 (1 patient)	FBLN5, 485-8P-DUP	-	-	RCV00000810
.0003	MACULAR DEGENERATION, AGE-RELATED, 3	FBLN5, VAL60LEU	rs121434299	rs121434299	RCV00000811..
.0004	MACULAR DEGENERATION, AGE-RELATED, 3	FBLN5, ARG71GLN	rs121434300	rs121434300	RCV00000812..
.0005	MACULAR DEGENERATION, AGE-RELATED, 3	FBLN5, PRO78SER	rs121434301	rs121434301	RCV00000813..

External Links:

- Genome
- DNA
- Protein
- Gene Info
- Clinical Resources
- Variant
- ClinVar
- gnomAD
- OMIM Catalog
- OMIM Central
- HGMD
- HLB-EVS
- PharmGKB
- Animal Models
- Cellular Pathways

Side-by-side Clinical Synopsis viewer

* 604580
FIBULIN 5; FBLN5

Cytogenetic location: 14q32.12 Genomic coordinates (GRCh38) 14,918,691,411-91,947,694 from NCBI

Gene-Phenotype Relationships

Location	Phenotype	MIM number	Inheritance	Phenotype mapping key
14q32.12	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE IA	604580	AD	3

View Clinical Synopsis

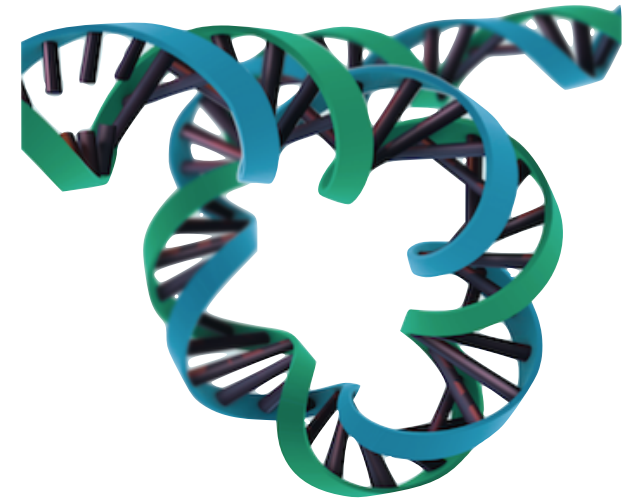
NUMBER	PHENOTYPE	MIM NUMBER	INHERITANCE	PHENOTYPE MAPPING KEY
1	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE IA	604580	AD	3
2	CUTIS LAXA, AUTOSOMAL DOMINANT 2 (1 patient)	604580	AD	3
3	MACULAR DEGENERATION, AGE-RELATED, 3	604580	AD	3
4	MACULAR DEGENERATION, AGE-RELATED, 3	604580	AD	3
5	MACULAR DEGENERATION, AGE-RELATED, 3	604580	AD	3
6	MACULAR DEGENERATION, AGE-RELATED, 3	604580	AD	3

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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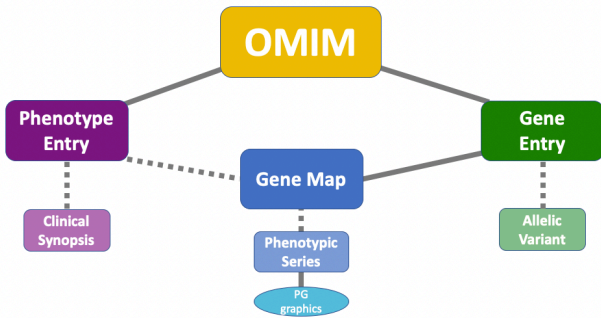
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OMIM® (Online Mendelian Inheritance in Man) is a continuously updated authoritative compendium of human genes and genetic phenotypes (disorders and traits) with full-text, referenced overviews of over 8,800 phenotypes and over 17,200 genes. OMIM focuses 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.

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.

Genomic context table	Location (from NCBI, GRCh38)	Gene/Locus name	Gene/Locus MIM number	Phenotype	Phenotype MIM number	Inheritance	Pheno map key	Comments	Mouse symbol (from MGI)
1:	1,901,915,304-1,922,213	ZNF644, MYP21	614159	Zinc finger protein 644	614167	AD	3		Zfp644
2:	1,151,281,522-1,213	ZNF687, KIAA1441, PDB6	610568	Zinc finger protein 687	616833	AD	3	fused with AML1 in t(1;21)	Zfp687
3:	1,244,048,491-1,44	ZBTB18, ZNF238, RFP58, MKDD2	608433	Zinc finger and RTB domain containing 18	612337	AD	3	Intellectual developmental disorder, autosomal dominant 22	Zbtb18
4:	2,27,377,235-2,23	ZNF513, RFP58	613598	Zinc finger protein 513	613617	AR	3	Tritanitis pigmentosa 58	Zfp513
5:	2,218,633,329-2,35	ZNF142, NEDD5HM	604083	Zinc finger protein-142	618425	AR	3	Neurodevelopmental disorder with impaired speech and hyperkinetic movements	Zfp142
6:	3,114,314,500	ZBTB20	606725	Zinc finger	259050	AD	3	Primrose syndrome	Zbtb20

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.

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.