



HGMD®:

Human Gene Mutation Data

The human gene mutation database (HGMD®) represents an up-to-date and comprehensive collection of known and published pathogenic gene lesions responsible for human inherited disease.

HGMD® provides information of practical diagnostic importance to medical and clinical geneticists, diagnosticians, bioinformaticians, researchers in human and molecular genetics and physicians and genetic counselors inter-

ested in a particular inherited condition in a given patient or family. HGMD® is a widely used, trusted resource that has been cited in over 5000 publications in leading scientific journals.

HGMD® is available as a free public version with restricted content and limited search options for academic use only and as a fully functional professional version that requires annual subscription through BIOBASE.

Key Capabilities

- Easily verify whether an observed mutation has been previously described to be responsible for causing human inherited disease
- Obtain an overview of the pathogenic mutational spectrum of a particular gene or disease
- Quickly access detailed reports for disease-associated human inherited mutations

From Our Customers

"HGMD® professional provides the most comprehensive database of human disease associations and is an invaluable resource in both clinical and research-grade genetics and genomics activities."

- Dr. Ali Torkamani, CSO at Cypher Genomics

"We rely on HGMD® professional heavily for reporting our clinical tests. We are currently working on next generation sequencing projects, identifying genes for disease-causing mutations and disease-associated / functional polymorphisms."

- We Yaping Yang, PhD, Baylor College of Medicine

HGMD accession	Reported disease/phenotype	Variant class	Gene symbol	Chrom. change	Amino acid change
C038874	Cerebellar ataxia-osteopenia syndrome	DM	GSK3B	C10C-C10G	Gln>Arg

Literature citation	Citation type	Comments
1. Nakano (2006) Nat Genet 38: 294-298 PMID: 1653243	Primary literature report	No comments
2. Aubry (2012) J Intern Med 270: 989-997 PMID: 2231217	Additional phenotype	CFC syndrome with muscular CrQII deficiency; Mut. detect. at
3. Carverilli (2013) Am J Hum Genet 92: 103-111 PMID: 2323823	Additional phenotype	LEOPARD syndrome.
4. Kuri (2014) Mol Syst Biol 10: 127 PMID: 2463483	Additional literature report	Structure-energy based predictions and network modeling: no
5. Mistry (2013) Hum Mol Genet 22: 7199-7208 PMID: 2382323	Additional literature report	Structure model
6. Rodriguez-Viciana (2008) Methods Biochem Anal 43B: 277-288 PMID: 1881202	Patented literature report	None
7. Wang Ransay (2014) Am J Med Genet 164A: 2058-2068 PMID: 2478173	Additional literature report	None

GenBank accession	Sequence
U00002.2	TTTGTGACCTTTTCCGAAAGCTGCTTTCCGAAAGGAGTTTCCGCTGTCACACATGTTGTTATAA
chr7:140801502	
GenBank accession	CCGC: CC3C: cosmid: NCBI: MapViewer: NCBI: MapViewer
GenBank accession	NC_004533.4 (770494): 20_2043324:p.Q27R
Variant Call Format (VCF)	chr7:140801502 G>A
Protein structure	P13035: Discont: molS
UniProt member	Q27R:140801502
UniProt member	Discont: chr7:140801502
CDS	No

Transt	Gln (Q)	Arg (R)	Position	Protein ID
Amino acid name	glutamine	arginine	207	NP_201232.2
Polarity change	polar	positively charged	207	NP_201232.2
pI	neutral	basic	207	NP_201232.2
Residue weight	133	156	207	NP_201232.2
Hydrophobicity scale	-0.5	-0.5	207	NP_201232.2
Hydrophilicity scale	0.2	1.0	207	NP_201232.2

Figure1. HGMD® Professional sample mutation report

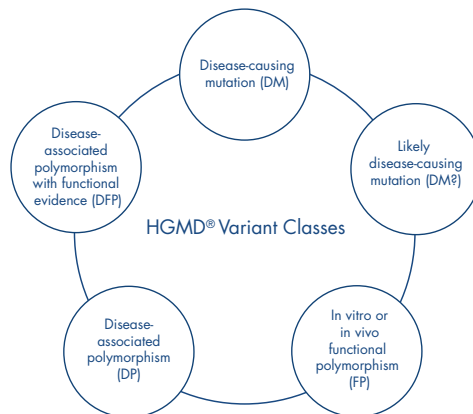


Figure2. Types of mutation within HGMD®