

## HGMD®: Human Gene Mutation Data

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

HGMD<sup>®</sup> 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-

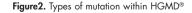
## Key Capabilities

- 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

HGMD® Professional 2016.1									
HGMD	Gene Mu	tation Phe	notype Referen	ce Batch	h Advanced	Statistics Infor	mation Support	Hor	
+		,							
<b>HGMD</b> accession	Reported disease	phenotype	Varia	ut class	Gene symb	d Coden change	Amino acid cl	ange	
CM060874	Cardio-facio-cutaneous syndrom			BRAF	CAG-CGG	Gin-Arg			
The Q257X substitution exhibits a shi	it is polarity from polar to positively charged and di	taglayn a decrease in l	Kyte-Dool Ittle hydrophobicity B	on-3.5 to -4.5. Ap	proximitally 0.63% of the	insense maintiers in MCO.D are Ols-Arg	The motation occurs 510 amino a	cids them the en	
Literature citation				Cit	Citation type Comments/s				
1. Nahoni (2006) Nat Genet 38: 294 Pattas: 10474404				Primary linearum opon No comments					
2. Aeby (2007) J Inherit Metab Dir 30: 827 Publie: 17700371				Addisonal phenotype		CFC syndrome with muscular CoQ10 deficiency; Mut. descr. as			
3. Carcavilla (2013) Rev Exp Cardiol 66: 350 Public: 2475314				A655	oral phenotype	LEOPARD syndrome.	OPARD syndrome.		
4. Kiel (2014) Mol Synt Biol 10: 727 Publish 2402000				Addition	of Steature report	Structure-energy-based predictions and network modelling - su			
5. Moniya (2015) Hum Mol Gener 24: 7349 Publist 16/2012				Addition	al Steature report	Mouse model	use model		
6. Rodriguez-Niciana (2008) Methods Enzymol 438: 277 publist 1341225				Punction	al characterisation	None			
7. Wong Ramsey (2014) Am J Med Genet A 164: 2036 Pueton: 24710072				Addition	al 2 tentors report	None			
					Extra informati	lon			
Coding strand genomic sequence (GRCh38) TTTGTG.				ACTITIGTOGAAAGCIGCTITICC(A/G)666TITICC6CIGTCAAACAIGIGGTTATAA					
Genomic coordinate (GRCh38)			chir7:140801502						
Genome viewers			UCSC: UCSC (corden): NCBI MapViewer: NCBI SeqViewer						
HOVS nomenclature			<u>NM_004333.4</u> ; c.770A>0; <u>NP_004324.2</u> ; p.Q257R						
Variant Call Format (VCF)			CHRON FOR ID REF ALT 7 140801802 CH090974 T C						
Protein structures			P15056: INstruct: multi						
dbSNP mamber			m150177035 (8)						
Variant class			Disease causing mutation						
CpG			Ne						
	GRCh37 legacy data 11								
	Amino acid compar	isen					Orthologous amine a-	cid <u>conserv</u>	
	Arg(R)			11	atonomy 🕑   Organism	Protei			
Amino acid name		glutanine	arginine			Tatonomy	Protein ID		
Polarity/charge		polar	positively charged			Homo saplens	NP 004324.2	247 8	
pH		neutral	basic		dilare	goda melanoleuca	XP_011226355.1	223 E	
Residue weight		128	156		dilitza	tor minimizerents	XP 006264783.1	243 1	
Hydrophobicity score		-3.5	-4.5			ligator strengts	XP 006019605.1	225 8	
Hydrophilicity acore		0.2	3.0			u elaterhenches	XP 005021554.1	145 2	

Figure 1. HGMD® Professional sample mutation report

Disease-causing mutation (DM) Disease Likely associated disease-causing mutation (DM?) polymorphism with functional vidence (DFF HGMD® Variant Classes In vitro or Disease in vivo associated functional polymorphism polymorphism (DP) (FP)



## From Our Customers

"HGMD<sup>®</sup> professional provides the most comprehensive database of human disease associations and is an invaluable resource in both clinical and researchgrade genetics and genomics activties."

– Dr. Ali Torkamani, CSO at Cypher Genomics

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

- We Yaping Yang, PhD, Baylor College of Medicine

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ested in a particular inherited condition in a

given patient or family. HGMD® is a widely

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## Sample to Insight