Human Gene Description (OMIM)

No summary description provided by OMIM

Gene-Phenotype Relationships

OMIM

No OMIM gene-phenotype relationships found

Reported Alleles From OMIM

OGDHL

No OMIM allelic variants found

Control Population Gene Summary

OGDHL (ExAC Gene Table)

Constraint from ExAC	Expected no. variants	Observed no. variants	ConstraintMetric
Synonymous	189.3	196	z=-0.30
Missense	417.8	414	z=0.09
LoF	31.1	13	pLI=0.00
CNV	9.4	23	z=-1.07

Disease Population (Geno2MP Database)

Chr10:50946295 G>A

No matches found

Gene-Phenotype Relationships (Geno2MP)

Chr10:50946295 G>A

No matches found

Population Allele Frequencies (ExAC Database)

Chr10:50946295 G>A

Allele count	1
Allele number	121070
Homozygous count	0
Allele frequency	0.00000825968
Gene	OGDHL

Population Allele Frequencies (GnomAD Database)

Chr10:50946295 G>A

Allele Count	3
Allele Number	277128
Homozygous count	0
Allele frequency	0.000010825
Gene	OGDHL

Benign (ClinVar)	Likely Benign (ClinVar)	Pathogenic (ClinVar)	Likely Pathogenic (ClinVar)	Risk Factor (ClinVar)	Uncertain Significance (ClinVar)	Conflicting Interpretations (ClinVar)
0	0	16	5	0	5	0

Reported Alleles From ClinVar

OGDHL / Chr10:50946295 G>A

Variation	Location	Condition(s)	Frequency	Clinical Significance	Review Status
NC_000010.10:g.(?_46491169)_(51081560_?)del	**Chr10:46491169-51081560**	See cases		Pathogenic	criteria provided, single submitter
NC_000010.10:g.(?_46242057)_(51595050_?)dup	**Chr10:46242057-51595050**	See cases		Pathogenic	criteria provided, single submitter
GRCh38/hg38 10q11.21-21.3(chr10:42112187-67400675)x3	**Chr10:42607635-69160433**	See cases		Pathogenic	no assertion criteria provided
GRCh38/hg38 10q11.22-11.23(chr10:45710242-49929364)x1	**Chr10:46205690-51330432**	See cases		Pathogenic	no assertion criteria provided
GRCh38/hg38 10q11.22-11.23(chr10:45931517-50035809)x3	**Chr10:46966533-51795569**	See cases		Uncertain significance	no assertion criteria provided
GRCh38/hg38 10q11.22-11.23(chr10:45788078-	**Chr10:46476965-**	See cases		Uncertain	no assertion criteria provided

Copy Number Variation In Control Population (DGV Database)

OGDHL

Show 10 entries Search:

Position	Size	Type	Subtype	Frequency	Gain	Loss	Sample Size	References
10 46692238 51472468	4780230	OTHER	inversion	0.00000000	0	0	9	18451855
10 49201820 51136375	1934555	CNV	loss	0.00003438	0	1	29084	25217958
10 50928727 51019190	90463	CNV	gain	0.00003438	1	0	29084	25217958
10 50949446 50953144	3698	CNV	duplication	0.00540541	1	0	185	20981092
10 50950375 50950772	397	CNV	insertion	1.00000000	1	0	1	19546169

Showing 1 to 5 of 5 entries Previous 1 Next

Common Copy Number Variants (DECIPHER Database)

10:50946295

No matches found

Gene Function Table

OGDHL

☑ Show only best DIOPT v6 score gene

	Homolog	DIOPT Score	Expression	Molecular function	Cellular component	Biological process
Human	OGDHL PubMed Monarch	NA	• cerebellum • kidney • parathyroid gland Show all / GTEx	• protein binding	No term based on experiment	No term based on experiment
Rat	Ogdhl PubMed	11/11	Show all (6)	No term based on experiment	No term based on experiment	No term based on experiment
Mouse	Ogdhl PubMed IMPC	12/13	No data available Open on MGI	No term based on experiment	No term based on experiment	No term based on experiment
Zebrafish	ogdhl PubMed	12/12	No structure expressed in wild-type Open on ZFIN	No term based on experiment	No term based on experiment	No term based on experiment
Drosophila	Nc73EF PubMed	9/12	• Head • Eye • Brain • Thoracic-Abdominal Ganglion • Crop • Midgut • Hindgut • Malpighian Tubules • Fat Body • Salivary Gland • Heart • Carcass • Ovary • Testis • VirginFemale Spermatheca • InseminatedFemale Spermatheca • Male Accessory Gland Show all (25)	No term based on experiment	• microtubule associated complex	No term based on experiment
C Elegans	ogdh-1 PubMed	8/12	Open on WormBase	• oxoglutarate dehydrogenase (succinyl-transferring) activity • oxidoreductase activity, acting on the aldehyde or oxo group of donors, disulfide as acceptor • thiamine pyrophosphate binding	• mitochondrion • mitochondrial matrix • cytosol • mitochondrial membrane • oxoglutarate dehydrogenase complex	• generation of precursor metabolites and energy • glycolytic process • tricarboxylic acid cycle
Budding Yeast	KGD1 PubMed	9/11	Open on SGD	No term based on experiment	• mitochondrion • mitochondrial oxoglutarate dehydrogenase complex • mitochondrial nucleoid	No term based on experiment
Fission Yeast	SPBC3H7.03c PubMed	6/8	Open on PomBase	No term based on experiment	• mitochondrion	No term based on experiment

Human Gene Protein Domains DIOPT V6

OGDHL

Index	Domain name	Domain start	Domain stop	Domain description	Protein ID	External ID
hs1	Transket_pyr	636	852	Transketolase, pyrimidine binding domain; pfam02779	NP_060715.2	CDD:202390
hs1	TPP_E1_OGDC_like	251	514	Thiamine pyrophosphate (TPP) family, E1 of OGDC-like subfamily, TPP-binding module; composed of proteins similar to the E1 component of the 2-oxoglutarate dehydrogenase multienzyme complex (OGDC). OGDC catalyzes the oxidative decarboxylation of...; cd02016	NP_060715.2	CDD:48179
hs1	sucA	46	1001	2-oxoglutarate dehydrogenase E1 component; Reviewed; PRK09404	NP_060715.2	CDD:181824

Multiple Protein Alignment DIOPT V6

OGDHL

```
hs1 ------MSQLRLLP--SRLGVQAARLLAAHDVPVFGWRSRSSGPPATFPSS---------------KGGGGSSYMEEMYFAWLENPQSVHKSWDSFFREASEEAFSGSAQP--RPPSV--[93]
mm1 ------MSQLRLLP--FRLGPRATKLLATRAIPVFSGCRRSSGPPTTIPRS---------------RSGVSSSYVEEMYFAWLENPQSVHKSWDSFFQRASKEASVGPAQP--QLPAV--[93]
rn1 ------MSQLRLLL--FRLGPQARKLLATRDIAAFGGRRRSSGPPTTIPRS---------------RGGVSPSYVEEMYFAWLENPQSVHKSWDNFFQRATKEASVGPAQP--QPPAV--[93]
dr1 ------MNQLRPLA--GALRSSSQWM-----------RGHVGTGKRVFDLR-RNCSSGVT---EPSLAACSSSYVVEMYYAWLEDHKNVHESWDAYFRNAE----ASSS-----------[82]
dm1 MHRAHTAFSLALSP--MAHKNFATWLLKS--------SSSQQMAKVTAAAAVRTYNSAAA---EPFANGSTASYVEEMYNAWLRDPTSVHTSWDAYFRSNS---YVSPPNL--AP-----[97]
ce1 MHRASLICRLASPSRINAIRNAS--------------SGKSHISASTLVQH-RNQSVAAAVKHEPFLNGSSSIYIEQMYEAWLQDPSSVHTSWDAYFRNVE--AGAGPGQAFQAPPAT--[101]
sc1 --------MLRFVSS-QTCRYSSRGLLKTSL----------LKNASTVKIVGRGLATTGT---DNFLSTSNATYIDEMYQAWQKDPSSVHVSWDAYFKNMS-NPKIPATKAFQAPPS---[94]
sp1 --------MLRFIPS-SA---KARALRRSAVTAY---RLNRLTCLSSLQ-QNRTFATQPT---DDFLTGGAADYVDEMYDAWKKDPNSVHASWQAYFKNVQ-ERGVSPSKAFQAPPL---[97]
          *           .                                    . *: :** ** .:  .** **: :*:         .
hs1 ---VHESRSAVSSR----------T------KTSKLVEDHLAVQSLIRAYQIRGHHVAQLDPLGILDADLD-SFVPSDLITTIDKL-------------------AFYDLQEADLDKEFQ[174]
mm1 ---LQESRTSVSSC----------T------KTSKLVEDHLAVQSLIRAYQIRGHHVAQLDPLGILDADLD-SFVPSDLITTIDKLGKSWDPSLLSFYAALASFPAFYDLQEADLDKEFR[193]
rn1 ---IQESRASVSSC----------T------KTSKLVEDHLAVQSLIRAYQIRGHHVAQLDPLGILDADLD-SFVPSDLITTIDKL-------------------AFYDLQEADLDKEFR[174]
dr1 ---VESGEKPLSMLLQGRTMSQTPA------MSEKVVEDHLAVHTLIRAYQVRGHHVARLDPLGILTADLD-SFVPSDLITSIDKL-------------------ASYGLEESDLDKSFQ[173]
dm1 ---VQANTLPLTAFNFGGAVAGA-A------PDSKTIDDHLAVQAIIRSYQIRGHNIAHLDPLEINTPEL-----PGN--SSTKSI-----------Y-------ANFSFGEQDMDRQFK[182]
```

Highlight from Integer (1~n) to Integer (1~n) for ☑ hs ☐ rn ☐ mm ☐ dr ☐ dm ☐ ce ☐ sc ☐ sp