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LCA5

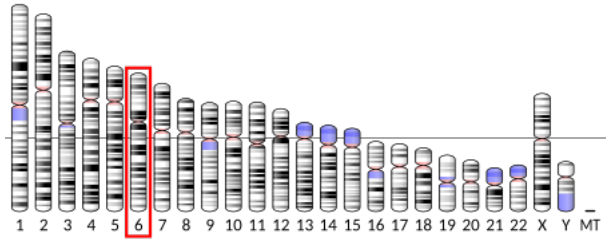
Lebercilin, also known as **leber congenital amaurosis 5** (LCA5), is a protein that in humans is encoded by the *LCA5* gene.[5][6][7] This protein is thought to be involved in centrosomal or ciliary functions.

Clinical significance

Mutations in the *LCA5* gene are associated with Leber's congenital amaurosis.

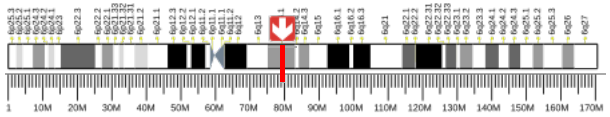
References

1. GRCh38: Ensembl release 89: ENSG00000135338 (http://May2017.archive.ensembl.org/Homo_sapiens/Gene/Summary?db=core;g=ENSG00000135338) – Ensembl, May 2017
2. GRCm38: Ensembl release 89: ENSMUSG00000032258 (http://May2017.archive.ensembl.org/Mus_musculus/Gene/Summary?db=core;g=ENSMUSG00000032258) – Ensembl, May 2017
3. "Human PubMed Reference:" (https://www.ncbi.nlm.nih.gov/sites/entrez?db=gene&cmd=Link&LinkName=gene_pubmed&from_uid=167691). *National Center for Biotechnology Information, U.S. National Library of Medicine*.
4. "Mouse PubMed Reference:" (https://www.ncbi.nlm.nih.gov/sites/entrez?db=gene&cmd=Link&LinkName=gene_pubmed&from_uid=75782). *National Center for Biotechnology Information, U.S. National Library*

LCA5	
Identifiers	
Aliases	LCA5 (https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/31923), C6orf152, Leber congenital amaurosis 5, lebercilin, lebercilin LCA5
External IDs	OMIM: 611408 (https://omim.org/entry/611408); MGI: 1923032 (http://www.informatics.jax.org/marker/MGI:1923032); HomoloGene: 32718 (https://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=homologene&dopt=HomoloGene&list_uids=32718); GeneCards: LCA5 (https://www.genecards.org/cgi-bin/carddisp.pl?gene=LCA5); OMA:LCA5 - orthologs (https://omabrowser.org/oma/vps/ENSG00000135338)
Gene location (Human)	
Chr.	Chromosome 6 (human)[1]

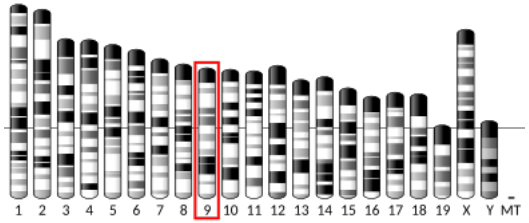
of Medicine.

5. "Entrez Gene: Leber congenital amaurosis 5" (https://www.ncbi.nlm.nih.gov/sites/entrez?Db=gene&Cmd=ShowDetailView&TermToSearch=167691).
6. Dharmaraj S, Li Y, Robitaille JM, Silva E, Zhu D, Mitchell TN, Maltby LP, Baffoe-Bonnie AB, Maumenee IH (January 2000). "A novel locus for Leber congenital amaurosis maps to chromosome 6q" (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1288337). *Am. J. Hum. Genet.* **66** (1): 319–26. doi:10.1086/302719 (https://doi.org/10.1086%2F302719). PMC 1288337 (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1288337). PMID 10631161 (https://pubmed.ncbi.nlm.nih.gov/10631161).
7. den Hollander AI, Koenekoop RK, Mohamed MD, Arts HH, Boldt K, Towns KV, Sedmak T, Beer M, Nagel-Wolfrum K, McKibbin M, Dharmaraj S, Lopez I, Ivings L, Williams GA, Springell K, Woods CG, Jafri H, Rashid Y, Strom TM, van der Zwaag B, Gosens I, Kersten FF, van Wijk E, Veltman JA, Zonneveld MN, van Beersum SE, Maumenee IH, Wolfrum U, Cheetham ME, Ueffing M, Cremers FP, Inglehearn CF, Roepman R (July 2007). "Mutations in LCA5, encoding the ciliary protein lebercilin, cause Leber congenital amaurosis". *Nat. Genet.* **39** (7): 889–95. doi:10.1038/ng2066 (https://doi.org/10.1038%2Fng2066). PMID 17546029 (https://pubmed.ncbi.nlm.nih.gov/17546029). S2CID 13772221 (https://api.semanticscholar.org/CorpusID:13772221).

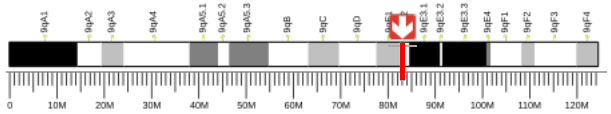


Band 6q14.1 **Start** 79,484,991 bp[1]
End 79,537,458 bp[1]

Gene location (Mouse)



Chr. Chromosome 9 (mouse)[2]



Band 9|9 E2 **Start** 83,272,346 bp[2]
End 83,323,180 bp[2]

RNA expression pattern

Bgee (https://www.bgee.org/)	Human	Mouse (ortholog)
	Top expressed in (https://www.bgee.org/gene/ENSG00000135338)	Top expressed in (https://www.bgee.org/gene/ENSMUSG00000032258)
	▪ mucosa of paranasal sinus	▪ superior cervical ganglion
	▪ bronchial epithelial cell	▪ neural layer of retina

	▪ caput epididymis ▪ testicle ▪ Achilles tendon ▪ secondary oocyte ▪ right uterine tube ▪ ventricular zone ▪ ganglionic eminence ▪ gonad ▪ hand ▪ spermatid ▪ tail of embryo ▪ genital tubercle ▪ spermatocyte ▪ zygote ▪ trigeminal ganglion ▪ retinal pigment epithelium More reference expression data (https://www.bgee.org/gene/ENSG00000135338)
BioGPS (http://biogps.org/)	n/a
	Gene ontology
Molecular function	▪ protein binding (http://amigo.geneontology.org/amigo/term/GO:0005515) ▪ protein-containing complex binding (http://amigo.geneontology.org/amigo/term/GO:0044877)
Cellular component	▪ cytoplasm (http://amigo.geneontology.org/amigo/term/GO:0005737) ▪ cell projection (http://amigo.geneontology.org/amigo/term/GO:0042995)

	▪ cilium (http://amigo.geneontology.org/amigo/term/GO:0005929) ▪ cytoskeleton (http://amigo.geneontology.org/amigo/term/GO:0005856) ▪ microtubule organizing center (http://amigo.geneontology.org/amigo/term/GO:0005815) ▪ axoneme (http://amigo.geneontology.org/amigo/term/GO:0005930) ▪ photoreceptor connecting cilium (http://amigo.geneontology.org/amigo/term/GO:0032391) ▪ ciliary basal body (http://amigo.geneontology.org/amigo/term/GO:0036064)
Biological process	▪ protein transport (http://amigo.geneontology.org/amigo/term/GO:0015031) ▪ intraciliary transport (http://amigo.geneontology.org/amigo/term/GO:0042073) ▪ photoreceptor cell maintenance (http://amigo.geneontology.org/amigo/term/GO:0045494)
Sources:Amigo (http://amigo.geneontology.org/) / QuickGO (https://www.ebi.ac.uk/QuickGO/)	

Orthologs

Species	Human	Mouse

Entrez	167691 (https://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=gene&cmd=retrieve&dopt=default&list_uids=167691&rn=1)	75782 (https://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=gene&cmd=retrieve&dopt=default&list_uids=75782&rn=1)
Ensembl	ENSG00000135338 (http://www.ensembl.org/Homo_sapiens/geneview?gene=ENSG00000135338;db=core)	ENSMUSG00000032258 (http://www.ensembl.org/Mus_musculus/geneview?gene=ENSMUSG00000032258;db=core)
UniProt	Q86VQ0 (https://www.uniprot.org/uniprot/Q86VQ0)	Q80ST9 (https://www.uniprot.org/uniprot/Q80ST9)
RefSeq (mRNA)	NM_001122769 (https://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NM_001122769) NM_181714 (https://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NM_181714)	NM_027448 (https://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NM_027448) NM_029434 (https://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NM_029434)
RefSeq	NP_001116241 (http	NP_081724 (https://ww

(protein)	s://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NP_001116241) NP_859065 (https://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NP_859065)	w.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NP_081724) NP_083710 (https://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?val=NP_083710)
Location (UCSC)	Chr 6: 79.48 – 79.54 Mb (https://genome.ucsc.edu/cgi-bin/hgTracks?org=Human&db=hg38&position=chr6:79484991-79537458)	Chr 9: 83.27 – 83.32 Mb (https://genome.ucsc.edu/cgi-bin/hgTracks?org=Mouse&db=mm0&position=chr9:83272346-83323180)
PubMed search	[3]	[4]
Wikidata		
View/Edit Human		View/Edit Mouse

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