PLP1 proteolipid protein 1 [*Homo sapiens* (human)]

Download Datasets

Gene ID: 5354, updated on 12-Sep-2024

Summary

Official Symbol	PLP1 provided by HGNC
Official Full Name	proteolipid protein 1 provided by HGNC
Primary source	HGNC:HGNC:9086
See related	Ensembl:ENSG00000123560 MIM:300401; AllianceGenome:HGNC:9086
Gene type	protein coding
RefSeq status	REVIEWED
Organism	Homo sapiens
Lineage	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as	PLP; PMD; HLD1; MMPL; SPG2; GPM6C; PLP/DM20
Summary	This gene encodes a transmembrane proteolipid protein that is the predominant component of myelin. The encoded protein may play a role in the compaction, stabilization, and maintenance of myelin sheaths, as well as in oligodendrocyte development and axonal survival. Mutations in this gene cause Pelizaeus-Merzbacher disease and spastic paraplegia type 2. Alternatively splicing results in multiple transcript variants, including the DM20 splice variant. [provided by RefSeq, Feb 2015]
Expression	Restricted expression toward brain (RPKM 1259.3) See more
Orthologs	mouse all

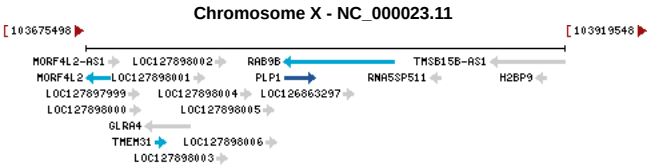
NEW **Try the new Gene table**
Try the new Transcript table

Genomic context

Location: Xq22.2

Exon count: 8

Annotation release	Status	Assembly	Chr	Location
RS_2024_08	current	GRCh38.p14 (GCF_000001405.40)	X	NC_000023.11 (103776506..103792619)
RS_2024_08	current	T2T-CHM13v2.0 (GCF_009914755.1)	X	NC_060947.1 (102223463..102239585)
RS_2024_09	previous assembly	GRCh37.p13 (GCF_000001405.25)	X	NC_000023.10 (103031434..103047548)

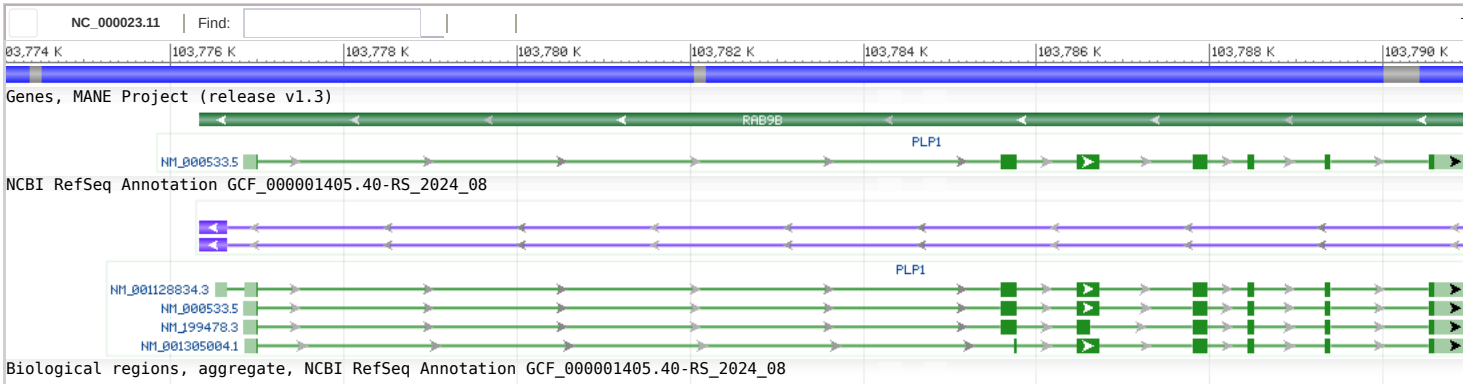


Genomic regions, transcripts, and products

Go to reference sequence details

Genomic Sequence: NC_000023.11 Chromosome X Reference GRCh38.p14 Primary Assembly

Go to nucleotide: Graphics FASTA GenBank

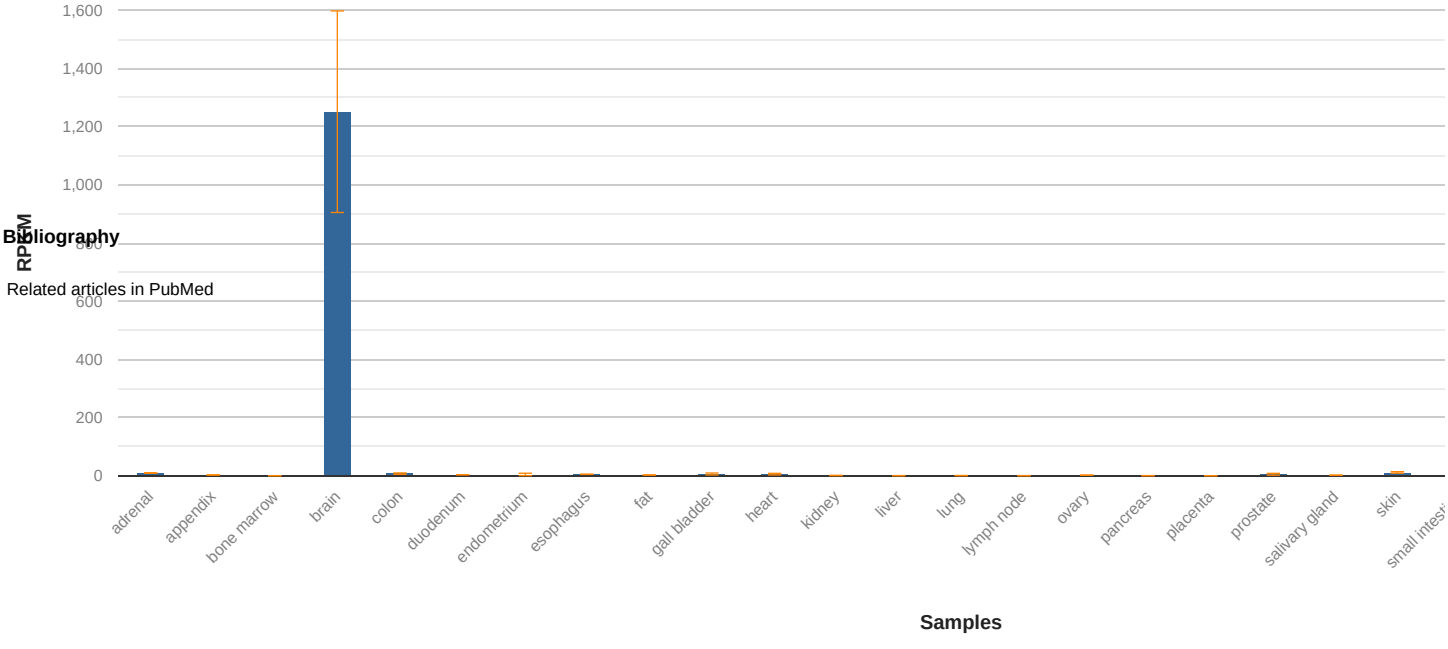


Expression

See details

HPA RNA-seq normal tissues

- Project title: HPA RNA-seq normal tissues HPA RNA-seq normal tissues
- Description: RNA-seq was performed of tissue samples from 95 human individuals representing 27 different tissues in order to determine tissue-specificity of all protein-coding genes
- BioProject: PRJEB4337
- Publication: PMID 24309898
- Analysis date: Wed Apr 4 07:08:55 2018



Bibliography

Related articles in PubMed

GeneRIFs: Gene References Into Functions

What's a GeneRIF?

1. Microdeletion in distal PLP1 enhancers causes hereditary spastic paraplegia 2.
2. PLP1 gene mutations cause spastic paraplegia type 2 in three families.
3. Identifying oligodendrocyte enhancers governing Plp1 expression.
4. Novel Insight into the Potential Pathogenicity of Mitochondrial Dysfunction Resulting from PLP1 Duplication Mutations in Patients with Pelizaeus-Merzbacher Disease.
5. Correlation Between Anti-Myelin Proteolipid Protein (PLP) Antibodies and Disease Severity in Multiple Sclerosis Patients With PLP Response-Permissive HLA Types.
6. These findings support the assumption that genetic variation in PLP1 affects white matter myelination in the healthy human brain.
7. demonstrate for the first time that the wmN1 enhancer region: (1) is functional in hPLP1
8. PLP1 mutation is associated with hereditary spastic paraplegia.
9. Findings suggest that PLP1 and CNTN1 gene variations modulate specific aspects of callosal microstructure that are in line with their gene function.
10. three single nucleotide polymorphisms in PLP1 that were associated with interhemispheric integration via the corpus callosum in a previous study also are relevant for functional hemispheric asymmetries.

Submit: New GeneRIF Correction See all GeneRIFs (82)

Phenotypes

BioGRID CRISPR Screen Phenotypes (21 hits/885 screens)

Find tests for this gene in the NIH Genetic Testing Registry (GTR)

Review eQTL and phenotype association data in this region using PheGenI

Associated conditions

Description	Tests
Hereditary spastic paraplegia 2 MedGen: C1839264, OMIM: 312920, GeneReviews: PLP1 Disorders	Compare labs
Pelizaeus-Merzbacher disease MedGen: C0205711, OMIM: 312080, GeneReviews: PLP1 Disorders	Compare labs

Copy number response

Description
Copy number response Haploinsufficency Sufficient evidence for dosage pathogenicity (Last evaluated 2021-04-12) ClinGen Genome Curation Page, PubMed
Triplosensitivity Sufficient evidence for dosage pathogenicity (Last evaluated 2021-04-12) ClinGen Genome Curation Page, PubMed

Variation

See variants in ClinVar

See studies and variants in dbVar

See Variation Viewer (GRCh37.p13)

See Variation Viewer (GRCh38)

Pathways from PubChem

3 items Download

Glial cell differentiation
Data Source: WikiPathways External ID: WP2276
Taxonomy Name: Homo sapiens (human)

Oligodendrocyte specification and differentiation, leading to myelin components for CNS
Data Source: WikiPathways External ID: WP4304
Taxonomy Name: Homo sapiens (human)

Sudden infant death syndrome (SIDS) susceptibility pathways
Data Source: WikiPathways External ID: WP706
Taxonomy Name: Homo sapiens (human)

PubChem

Interactions

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Products	Interactant	Other Gene	Complex	Source	Pubs	Description
P60201	P27797	CALR		HPRD	PubMed	
P60201	P27824	CANX		HPRD	PubMed	
P60201	P06756	ITGAV		HPRD	PubMed	
P60201	P20916	MAG		HPRD	PubMed	
P60201	P02686	MBP		HPRD	PubMed	
BioGRID:111368	BioGRID:128799	ADGRG3		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:106710	AKT1		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:106848	APP		BioGRID	PubMed	Affinity Capture-Western; Two-hybrid
BioGRID:111368	BioGRID:124627	AQP10		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:106859	AQP6		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:128353	ARL13B		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:117285	BCL2L13		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:107107	BIK		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:123732	CALN1		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:107262	CALR		BioGRID	PubMed	Affinity Capture-Western
BioGRID:111368	BioGRID:107396	CD40		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:107410	CD74		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:115692	CDIPT		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:114532	CLDN2		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:119070	CLDN20		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:118886	CLEC2D		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:108369	CLN8		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:115751	CREB3		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:124786	CREB3L1		BioGRID	PubMed	Two-hybrid
BioGRID:111368	BioGRID:107889	CTSD		BioGRID	PubMed	Co-localization

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General gene information

Markers

Homology

NCBI Orthologs
Orthologs from OrthoDB

Gene Ontology Provided by GOA

Function	Evidence Code	Pubs
enables protein binding	IPI	PubMed
enables protein-containing complex binding	IEA	
enables structural constituent of myelin sheath	IBA	
enables structural molecule activity	TAS	PubMed

Process	Evidence Code	Pubs
involved_in AMPA selective glutamate receptor signaling pathway	IEA	
involved_in astrocyte development	IEA	
involved_in axon development	IBA	
involved_in axon ensheathment	TAS	PubMed
involved_in central nervous system myelination	IBA	
involved_in chemical synaptic transmission	TAS	PubMed
involved_in inflammatory response	IEA	
involved_in long-chain fatty acid biosynthetic process	IEA	
involved_in positive regulation of calcium ion transmembrane transport	IEA	
involved_in positive regulation of cell migration	IEA	
involved_in positive regulation of gene expression	IEA	
involved_in substantia nigra development	HEP	PubMed

Component	Evidence Code	Pubs
part_of integrin alphav-beta3 complex	IEA	
is_active_in myelin sheath	IBA	
is_active_in plasma membrane	IBA	
located_in plasma membrane	IDA	PubMed
located_in synapse	IEA	

General protein information

Preferred Names
myelin proteolipid protein

Names
lipophilin
major myelin proteolipid protein

NCBI Reference Sequences (RefSeq)

NEW **Try the new Transcript table**

RefSeqs maintained independently of Annotated Genomes

These reference sequences exist independently of genome builds. Explain

Genomic

1. **NG_008863.2 RefSeqGene**

Range	5330..21109
Download	GenBank, FASTA, Sequence Viewer (Graphics)

mRNA and Protein(s)

1. **NM_000533.5** → **NP_000524.3** **myelin proteolipid protein isoform 1**

   See identical proteins and their annotated locations for NP_000524.3

   Status: REVIEWED

Description	Transcript Variant: This variant (1, also known as PLP) encodes the longest isoform (1). Variants 1 and 3 encode the same isoform (1).
Source sequence(s)	AK292728, BC095452, DA299940
Consensus CDS	CCDS14513.1

UniProtKB/Swiss-Prot	P04400, P06905, P60201, Q502Y1, Q6FHZ6
UniProtKB/TrEMBL	A8K9L3, B4DI21, E7EMV6
Related	ENSP00000484450.1, ENST00000621218.5

Conserved Domains (1) **summary**

	pfam01275 Location:5 → 273	Myelin_PLP; Myelin proteolipid protein (PLP or lipophilin)

2. **NM_001128834.3** → **NP_001122306.1** **myelin proteolipid protein isoform 1**

   See identical proteins and their annotated locations for NP_001122306.1

   Status: REVIEWED

Description	Transcript Variant: This variant (3) differs in the 5' UTR compared to variant 1. Variants 1 and 3 encode the same isoform (1).
Source sequence(s)	AK292728, BC095452, DC342197
Consensus CDS	CCDS14513.1
UniProtKB/Swiss-Prot	P04400, P06905, P60201, Q502Y1, Q6FHZ6
UniProtKB/TrEMBL	A8K9L3, B4DI21, E7EMV6
Related	ENSP00000481006.1, ENST00000612423.4

Conserved Domains (1) **summary**

	pfam01275 Location:5 → 273	Myelin_PLP; Myelin proteolipid protein (PLP or lipophilin)

3. **NM_001305004.1** → **NP_001291933.1** **myelin proteolipid protein isoform 3**

   See identical proteins and their annotated locations for NP_001291933.1

   Status: REVIEWED

Description	Transcript Variant: This variant (4) has a shorter 5' UTR and uses an alternate in-frame acceptor site compared to variant 1. It encodes a shorter isoform (3) than isoform 1.
Source sequence(s)	AK292728, AK295388, BC095452, DC342996
UniProtKB/TrEMBL	B4DI30

Conserved Domains (1) **summary**

	pfam01275 Location:2 → 218	Myelin_PLP; Myelin proteolipid protein (PLP or lipophilin)

4. **NM_199478.3** → **NP_955772.1** **myelin proteolipid protein isoform 2**

   See identical proteins and their annotated locations for NP_955772.1

   Status: REVIEWED

Description	Transcript Variant: This variant (2, also known as DM20) uses an alternate in-frame donor splice site compared to variant 1. It encodes a shorter isoform (2) than isoform 1.
Source sequence(s)	AK292728, BC095452, DA299940
Consensus CDS	CCDS14514.1
UniProtKB/TrEMBL	A0A0S2Z4D4, Q5U0F2, Q5U0F3
Related	ENSP00000477619.1, ENST00000619236.1

Conserved Domains (1) **summary**

	pfam01275 Location:5 → 238	Myelin_PLP; Myelin proteolipid protein (PLP or lipophilin)

RefSeqs of Annotated Genomes: GCF_000001405.40-RS_2024_08

The following sections contain reference sequences that belong to a specific genome build. Explain

Reference GRCh38.p14 Primary Assembly

Genomic

1. **NC_000023.11 Reference GRCh38.p14 Primary Assembly**

Range	103776506..103792619
Download	GenBank, FASTA, Sequence Viewer (Graphics)

Alternate T2T-CHM13v2.0

Genomic

1. **NC_060947.1 Alternate T2T-CHM13v2.0**

Range	102223463..102239585
Download	GenBank, FASTA, Sequence Viewer (Graphics)

Related sequences

Items 1 - 25 of 43 < Prev Page 1 of 2 Next >

Nucleotide		Protein
Heading	Accession and Version	
genomic	AH002975.2	AAA60350.1
genomic	AJ006976.1	CAA07364.1
genomic	CH471190.2	EAW54690.1
		EAW54691.1
		EAW54692.1
		EAW54693.1
		EAW54694.1
genomic	CP068255.2 (102223463..102239585)	None
genomic	D13320.1	BAA02577.1
genomic	D16830.1	BAA04110.1
genomic	KF510145.1	None
genomic	KF510147.1	None
genomic	KF510609.1	None
genomic	M27111.1 (1272..1307)	None
genomic	M95057.1 (40..397)	None
genomic	S55837.1	AAD13880.1
genomic	S62086.1	AAB26927.2
genomic	S62117.1	AAB26928.1
genomic	X66420.1	CAA47050.1
genomic	Z73964.2 (6072..22181)	None
mRNA	AI205189.1	None
mRNA	AK128782.1	None
mRNA	AK292728.1	BAF85417.1
mRNA	AK295374.1	BAG58333.1
mRNA	AK295388.1	BAG58342.1
mRNA	AK308966.1	None
mRNA	AK309239.1	None
mRNA	AK312340.1	BAG35261.1
mRNA	AV731932.2	None

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Protein Accession	Links	
	GenPept Link	UniProtKB Link
P60201.2	GenPept	UniProtKB/Swiss-Prot:P60201

Additional links

Locus-specific Databases

PLP1 @ LOVD

Gene LinkOut