

GEPHE SUMMARY

	Gephebase Gene		GepheID
TRPM1 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~TRPM1~#gephebase-summary-title)		GP00001140	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology~#gephebase-summary-title)	Trait		
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(coat)~#gephebase-summary-title)	Trait State in Taxon A		
Equus caballus	Trait State in Taxon B		
Equus caballus - Appaloosa and pre-domestication era fossils ; Stationary congenital night blindness (homozygotes) & Leopard Complex/Appaloosa spotting (incompletely dominant)	Ancestral State		
Taxon A	Taxonomic Status		
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific~#gephebase-summary-title)			
Taxon A		Taxon B	
Latin Name		Latin Name	
Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Equus caballus~#gephebase-summary-title)		Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Equus caballus~#gephebase-summary-title)	
Common Name		Common Name	
horse		horse	
Synonyms		Synonyms	
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758		Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	
Rank		Rank	
species		species	
Lineage		Lineage	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Perissodactyla; Equidae; Equus; Equus		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Perissodactyla; Equidae; Equus; Equus	
Parent		Parent	
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)		Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)		9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)	
is Taxon A an Infraspecies?		is Taxon B an Infraspecies?	
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
TRPM1		Q7Z4N2 (http://www.uniprot.org/uniprot/Q7Z4N2)	
	Synonyms		GenebankID or UniProtKB
MLSN1; CSNB1C; LTRPC1; MLSN		XP_005602903 (https://www.ncbi.nlm.nih.gov/nuccore/XP_005602903)	
	String		
9606.ENSPO0000380897 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000380897)			
	Sequence Similarities		
Belongs to the transient receptor (TC 1.A.4) family. LTrpC subfamily. TRPM1 subfamily.			
	GO - Molecular Function		
GO:0005262 : calcium channel activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005262)			
	GO - Biological Process		
GO:0070588 : calcium ion transmembrane transport			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0070588>)
GO:0007601 : visual perception (<https://www.ebi.ac.uk/QuickGO/term/GO:0007601>)
GO:0051262 : protein tetramerization (<https://www.ebi.ac.uk/QuickGO/term/GO:0051262>)
GO:0071482 : cellular response to light stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071482>)
GO:0060402 : calcium ion transport into cytosol
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060402>)
GO:0007216 : G protein-coupled glutamate receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007216>)
GO:0046548 : retinal rod cell development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046548>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005887 : integral component of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)
GO:0035841 : new growing cell tip (<https://www.ebi.ac.uk/QuickGO/term/GO:0035841>)

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)	Presumptive Null
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)	Molecular Type
Insertion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Insertion^#gephebase-summary-title)	Aberration Type
1-10 kb	Insertion Size
a 1378 bp retroviral LTR insertion in intron 1 of TRPM1	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title)	Experimental Evidence
Fine-mapping and mutation analysis of TRPM1: a candidate gene for leopard complex (LP) spotting and congenital stationary night blindness in horses. (2010) (https://pubmed.ncbi.nlm.nih.gov/20353955)	Main Reference
Bellone RR; Forsyth G; Leeb T; Archer S; Sigurdsson S; Imsland F; Mauceli E; Engensteiner M; Bailey E; Sandmeyer L; Grahn B; Lindblad-Toh K; Wade CM	Authors

Abstract

Leopard Complex spotting occurs in several breeds of horses and is caused by an incompletely dominant allele (LP). Homozygosity for LP is also associated with congenital stationary night blindness (CSNB) in Appaloosa horses. Previously, LP was mapped to a 6 cm region on ECA1 containing the candidate gene TRPM1 (Transient Receptor Potential Cation Channel, Subfamily M, Member 1) and decreased expression of this gene, measured by qRT-PCR, was identified as the likely cause of both spotting and ocular phenotypes. This study describes investigations for a mutation causing or associated with the Leopard Complex and CSNB phenotype in horses. Re-sequencing of the gene and associated splice sites within the 105 624 bp genomic region of TRPM1 led to the discovery of 18 SNPs. Most of the SNPs did not have a predictive value for the presence of LP. However, one SNP (ECA1:108,249,293 C>T) found within intron 11 had a strong ($P < 0.0005$), but not complete, association with LP and CSNB and thus is a good marker but unlikely to be causative. To further localize the association, 70 SNPs spanning over two Mb including the TRPM1 gene were genotyped in 192 horses from three different breeds segregating for LP. A single 173 kb haplotype associated with LP and CSNB (ECA1: 108,197,355-108,370,150) was identified. Illumina sequencing of 300 kb surrounding this haplotype revealed 57 SNP variants. Based on their localization within expressed sequences or regions of high sequence conservation across mammals, six of these SNPs were considered to be the most likely candidate mutations. While the precise function of TRPM1 remains to be elucidated, this work solidifies its functional role in both pigmentation and night vision. Further, this work has identified several potential regulatory elements of the TRPM1 gene that should be investigated further in this and other species.

Genotypes of predomestic horses match phenotypes painted in Paleolithic works of cave art. (2011) (https://pubmed.ncbi.nlm.nih.gov/22065780) Evidence for a retroviral insertion in TRPM1 as the cause of congenital stationary night blindness and leopard complex spotting in the horse. (2013) (https://pubmed.ncbi.nlm.nih.gov/24167615) Phenotypic and Genetic Analysis of the Leopard Complex Spotting in Noriker Horses. (2017) (https://pubmed.ncbi.nlm.nih.gov/28453641)	Additional References
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RELATED GEPHE

13 (Agouti, Endothelin receptor B, Kit (type III receptor protein-tyrosine kinase), MC1R, MFSD12, Microphthalmia-associated transcription factor, Pax3, PMEL17, SLC24A, SLC36A1, SLC45A2=MATP, syntaxin-17, T-box transcription factor (TBX3)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9796^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

@TE @HeterozygoteAdvantage <https://omia.org/OMIA002139/9796/>

