GEPHE SUMMARY

Gephebase Gene
TRPM1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^TRPM1^#gephebase-summary-title)

GepheID
GP00001140

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^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+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name
Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Equus caballus^#gephebase-summary-title)

Common Name
horse

Synonyms
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758

Rank
species

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

Parent
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)

NCBI Taxonomy ID
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Equus caballus^#gephebase-summary-title)

Common Name
horse

Synonyms
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758

Rank
species

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

Parent
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)

NCBI Taxonomy ID
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
TRPM1

Synonyms
MLSN1; CSNB1C; LTRPC1; MLSN

String
9606.ENSP00000380897 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000380897)

Sequence Similarities
Belongs to the transient receptor (TC 1.A.4) family. LTrpC subfamily. TRPM1 sub-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

UniProtKB Homo sapiens
Q7Z4N2 (http://www.uniprot.org/uniprot/Q7Z4N2)

GenebankID or UniProtKB
XP_005602903 (https://www.ncbi.nlm.nih.gov/nuccore/XP_005602903)

(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)

Presumptive Null

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)

Molecular Type

Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)

Aberration Type

Insertion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Insertion^#gephebase-summary-title)

Insertion Size

1-10 kb

Molecular Details of the Mutation

a 1378 bp retroviral LTR insertion in intron 1 of TRPM1

Experimental Evidence

Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title)

Main Reference

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)

Authors

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

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.

Additional References

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)

RELATED GEPHE

Related Genes

13 (Agouti, Endothelin receptor B, Kit (type III receptor protein-tyrosine kinase), MC1R, MFSD12, Microphtalmia-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 Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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