GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00000269

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 - "overo" coat

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#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?

Yes

Taxon B Description

Equus caballus - "overo" coat

GENOTYPIC CHANGE

Generic Gene Name

Ednrb

Synonyms

ETb; ET-B; ET-BR; ETR-b; Sox10m1

String

10090.ENSMUSP00000022718 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000022718)

Sequence Similarities

Belongs to the G-protein coupled receptor 1 family. Endothelin receptor subfamily. EDNRB sub-subfamily.

GO - Molecular Function

GO:0017046 : peptide hormone binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017046)
GO:0004962 : endothelin receptor activity

UniProtKB Mus musculus

P48302 (http://www.uniprot.org/uniprot/P48302)

GenebankID or UniProtKB

O62709 (https://www.ncbi.nlm.nih.gov/nuccore/O62709)

(https://www.ebi.ac.uk/QuickGO/term/GO:0004962)
GO:0031702 : type 1 angiotensin receptor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0031702)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0007422 : peripheral nervous system development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007422)
GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0000122)
GO:0001755 : neural crest cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0001755)
GO:0019934 : cGMP-mediated signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0019934)
GO:0007186 : G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007186)
GO:0032269 : negative regulation of cellular protein metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0032269)
GO:0042311 : vasodilation (https://www.ebi.ac.uk/QuickGO/term/GO:0042311)
GO:0008284 : positive regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0008284)
GO:0019722 : calcium-mediated signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0019722)
GO:0007204 : positive regulation of cytosolic calcium ion concentration
(https://www.ebi.ac.uk/QuickGO/term/GO:0007204)
GO:0035815 : positive regulation of renal sodium excretion
(https://www.ebi.ac.uk/QuickGO/term/GO:0035815)
GO:0051930 : regulation of sensory perception of pain
(https://www.ebi.ac.uk/QuickGO/term/GO:0051930)
GO:0032496 : response to lipopolysaccharide
(https://www.ebi.ac.uk/QuickGO/term/GO:0032496)
GO:0014070 : response to organic cyclic compound
(https://www.ebi.ac.uk/QuickGO/term/GO:0014070)
GO:0048265 : response to pain (https://www.ebi.ac.uk/QuickGO/term/GO:0048265)
GO:0008217 : regulation of blood pressure
(https://www.ebi.ac.uk/QuickGO/term/GO:0008217)
GO:0048066 : developmental pigmentation
(https://www.ebi.ac.uk/QuickGO/term/GO:0048066)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0042310 : vasoconstriction (https://www.ebi.ac.uk/QuickGO/term/GO:0042310)
GO:0014826 : vein smooth muscle contraction
(https://www.ebi.ac.uk/QuickGO/term/GO:0014826)
GO:0007568 : aging (https://www.ebi.ac.uk/QuickGO/term/GO:0007568)
GO:0071222 : cellular response to lipopolysaccharide
(https://www.ebi.ac.uk/QuickGO/term/GO:0071222)
GO:0086100 : endothelin receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0086100)
GO:0048484 : enteric nervous system development
(https://www.ebi.ac.uk/QuickGO/term/GO:0048484)
GO:0035645 : enteric smooth muscle cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0035645)
GO:0042045 : epithelial fluid transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0042045)
GO:0048246 : macrophage chemotaxis
(https://www.ebi.ac.uk/QuickGO/term/GO:0048246)
GO:0014043 : negative regulation of neuron maturation
(https://www.ebi.ac.uk/QuickGO/term/GO:0014043)
GO:0007200 : phospholipase C-activating G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007200)
GO:0060406 : positive regulation of penile erection
(https://www.ebi.ac.uk/QuickGO/term/GO:0060406)
GO:0001934 : positive regulation of protein phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0001934)
GO:0035810 : positive regulation of urine volume
(https://www.ebi.ac.uk/QuickGO/term/GO:0035810)
GO:0007497 : posterior midgut development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007497)
GO:0050678 : regulation of epithelial cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0050678)
GO:0031620 : regulation of fever generation
(https://www.ebi.ac.uk/QuickGO/term/GO:0031620)
GO:0006885 : regulation of pH (https://www.ebi.ac.uk/QuickGO/term/GO:0006885)
GO:1990839 : response to endothelin (https://www.ebi.ac.uk/QuickGO/term/GO:1990839)
GO:0019233 : sensory perception of pain
(https://www.ebi.ac.uk/QuickGO/term/GO:0019233)

GO - Cellular Component

GO:0016021 : integral component of membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0016020 : membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)
GO:0045121 : membrane raft (https://www.ebi.ac.uk/QuickGO/term/GO:0045121)
GO:0031965 : nuclear membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0031965)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Ile118Lys

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ile	Lys	118

Main Reference

A missense mutation in the endothelin-B receptor gene is associated with Lethal White Foal Syndrome: an equine version of Hirschsprung disease. (1998) (https://pubmed.ncbi.nlm.nih.gov/9585428)

Authors

Metallinos DL; Bowling AT; Rine J

Abstract

Lethal White Foal Syndrome is a disease associated with horse breeds that register white coat spotting patterns. Breedings between particular spotted horses, generally described as frame overo, produce some foals that, in contrast to their parents, are all white or nearly all white and die shortly after birth of severe intestinal blockage. These foals have aganglionosis characterized by a lack of submucosal and myenteric ganglia from the distal small intestine to the large intestine, similar to human Hirschsprung Disease. Some sporadic and familial cases of Hirschsprung Disease are due to mutations in the endothelin B receptor gene (EDNRB). In this study, we investigate the role of EDNRB in Lethal White Foal Syndrome. A cDNA for the wild-type horse endothelin-B receptor gene was cloned and sequenced. In three unrelated lethal white foals, the EDNRB gene contained a 2-bp nucleotide change leading to a missense mutation (I118K) in the first transmembrane domain of the receptor, a highly conserved region of this protein among different species. Seven additional unrelated lethal white foal samples were found to be homozygous for this mutation. No other homozygotes were identified in 138 samples analyzed, suggesting that homozygosity was restricted to lethal white foals. All (40/40) horses with the frame overo pattern (a distinct coat color pattern that is a subset of overo horses) that were tested were heterozygous for this allele, defining a heterozygous coat color phenotype for this mutation. Horses with tobiano markings included some carriers, indicating that tobiano is epistatic to frame overo. In addition, horses were identified that were carriers but had no recognized overo coat pattern phenotype, demonstrating the variable penetrance of the mutation. The test for this mutant allele can be utilized in all breeds where heterozygous animals may be unknowingly bred to each other including the Paint Horse, Pinto horse, Quarter Horse, Miniature Horse, and Thoroughbred.

Additional References

Endothelin receptor B polymorphism associated with lethal white foal syndrome in horses. (1998) (https://pubmed.ncbi.nlm.nih.gov/9530628)
A dinucleotide mutation in the endothelin-B receptor gene is associated with lethal white foal syndrome (LWFS); a horse variant of Hirschsprung disease. (1998) (https://pubmed.ncbi.nlm.nih.gov/9580670)

RELATED GEPHE

Related Genes

13 (Agouti, 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), TRPM1) (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

@Epistasis ; @Pleiotropy @HeterozygoteAdvantage ; with effects analogous to Hirschsprung Disease in human