GEPHE SUMMARY

Gephebase Gene
MFSD12 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MFSD12^#gephebase-summary-title)

GepheID
GP00002243

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
brown coat ; Shetland pony

Trait State in Taxon B
light brown (Mushroom dilution phenotype) ; Shetland pony ; autosomal recessive

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?
No

GENOTYPIC CHANGE

Generic Gene Name
MFSD12

Synonyms
PP3501; C19orf28

String
9606.ENSP00000347583 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000347583)

Sequence Similarities
Belongs to the major facilitator superfamily.

GO - Molecular Function
GO:0015293 : symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015293)

GO - Biological Process
GO:0048022 : negative regulation of melanin biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0048022)
GO:0008643 : carbohydrate transport (https://www.ebi.ac.uk/QuickGO/term/GO:0008643)

UniProtKB Homo sapiens
Q6NUT3 (http://www.uniprot.org/uniprot/Q6NUT3)

GenebankID or UniProtKB
()

GO:0071702 : organic substance transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0071702)

GO - Cellular Component

GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0005765 : lysosomal membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005765)
GO:0005770 : late endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005770)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Insertion Size

1-9 bp

Molecular Details of the Mutation

p.(Asp201fs) due to c.600Cins

Experimental Evidence

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

Main Reference

Frameshift Variant in MFSD12 Explains the Mushroom Coat Color Dilution in Shetland Ponies. (2019) (https://pubmed.ncbi.nlm.nih.gov/31635058)

Authors

Tanaka J; Leeb T; Rushton J; Famula TR; Mack M; Jagannathan V; Flury C; Bachmann I; Eberth J; McDonnell SM; Penedo MCT; Bellone RR

Abstract

Mushroom is a unique coat color phenotype in Shetland Ponies characterized by the dilution of the chestnut coat color to a sepia tone and is hypothesized to be a recessive trait. A genome wide association study (GWAS), utilizing the Affymetrix 670K array (MNEc670k) and a single locus mixed linear model analysis (EMMAX), identified a locus on ECA7 for further investigation (P = 2.08 Ã— 10). This locus contained a 3 Mb run of homozygosity in the 12 mushroom ponies tested. Analysis of high throughput Illumina sequencing data from one mushroom Shetland pony compared to 87 genomes from horses of various breeds, uncovered a frameshift variant, p.Asp201fs, in the MFSD12 gene encoding the major facilitator superfamily domain containing 12 protein. This variant was perfectly concordant with phenotype in 96 Shetland Ponies (P = 1.15 Ã— 10), was identified in the closely related Miniature Horse for which the mushroom phenotype is suspected to occur (f = 0.02), and was absent in 252 individuals from seven additional breeds not reported to have the mushroom phenotype. MFSD12 is highly expressed in melanocytes and variants in this gene in humans, mice, and dogs impact pigmentation. Given the role of MFSD12 in melanogenesis, we propose that p.Asp201fs is causal for the dilution observed in mushroom ponies.

Additional References

RELATED GEPHE

Related Genes

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

@Parallelism https://omia.org/OMIA002197/9796/