

GEPHE SUMMARY

MFSD12 (https://www.gephebase.org/search-criteria/?and+GeneGephebase=%MFSD12%#gephebase-summary-title)	Gephebase Gene	GP00002243	GephelD
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (<https://www.gephebase.org/search-criteria/?and+TraitCategory=%Morphology%#gephebase-summary-title>)

Coloration (coat) ([https://www.gephebase.org/search-criteria/?and+Trait=%Coloration\(coat\)%#gephebase-summary-title](https://www.gephebase.org/search-criteria/?and+Trait=%Coloration(coat)%#gephebase-summary-title))

brown coat ; Shetland pony

light brown (Mushroom dilution phenotype) ; Shetland pony ; autosomal recessive

Taxon A

Domesticated (<https://www.gephebase.org/search-criteria/?and+TaxonomicStatus=%Domesticated%#gephebase-summary-title>)

Equus caballus

(<https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Equus+caballus%#gephebase-summary-title>)

horse

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

species

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

Equus () - (Rank: subgenus)

(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510>)

9796

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

is Taxon A an Infraspecies?

No

GENOTYPIC CHANGE

MFSD12

PP3501; C19orf28

9606.ENSP00000347583

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

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>)

Generic Gene Name

Synonyms

String

Sequence Similarities

UniProtKB Homo sapiens

GenebankID or UniProtKB

0

GO:0015293 (<http://www.uniprot.org/uniprot/Q6NUT3>)

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>)

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=%27Yes%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive%20Null=%27Yes%27#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=%27Coding%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular%20Type=%27Coding%27#gephebase-summary-title))

Aberration Type

Insertion ([https://www.gephebase.org/search-criteria?/and+Aberration Type=%27Insertion%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration%20Type=%27Insertion%27#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=%27Association Mapping%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental%20Evidence=%27Association%20Mapping%27#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 ($P = 2.08 \times 10^{-10}$). 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 ($P = 1.15 \times 10^{-10}$), 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, Microphthalmia-associated transcription factor, Pax3, PMEL17, SLC24A, SLC36A1, SLC45A2=MATP, syntaxin-17, T-box transcription factor (TBX3), TRPM1) (<https://www.gephebase.org/search-criteria?/or+TaxonID=%279796%27/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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