

## GEPHE SUMMARY

MFSD12 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~MFSD12~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~MFSD12~#gephebase-summary-title</a> )	Gephebase Gene	GP00002243	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title</a> )	Trait Category		
Coloration (coat) ( <a href="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</a> )	Trait		
brown coat ; Shetland pony	Trait State in Taxon A		
light brown (Mushroom dilution phenotype) ; Shetland pony ; autosomal recessive	Trait State in Taxon B		
	Ancestral State		
	Taxon A		
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title</a> )	Taxonomic Status		
	Taxon A		Taxon B
Equus caballus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Equus+caballus~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Equus+caballus~#gephebase-summary-title</a> )	Latin Name	Equus caballus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Equus+caballus~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Equus+caballus~#gephebase-summary-title</a> )	Latin Name
horse	Common Name	horse	Common Name
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	Synonyms	Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	Synonyms
species	Rank	species	Rank
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	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	Lineage
Equus () - (Rank: subgenus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510</a> )	Parent	Equus () - (Rank: subgenus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510</a> )	Parent
9796 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796</a> )	NCBI Taxonomy ID	9796 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

MFSD12	Generic Gene Name	Q6NUT3 ( <a href="http://www.uniprot.org/uniprot/Q6NUT3">http://www.uniprot.org/uniprot/Q6NUT3</a> )	UniProtKB Homo sapiens
PP3501; C19orf28	Synonyms	()	GenebankID or UniProtKB
9606.ENSPO0000347583 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000347583">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000347583</a> )	String		
Belongs to the major facilitator superfamily.	Sequence Similarities		
GO:0015293 : symporter activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0015293">https://www.ebi.ac.uk/QuickGO/term/GO:0015293</a> )	GO - Molecular Function		
GO:0048022 : negative regulation of melanin biosynthetic process ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0048022">https://www.ebi.ac.uk/QuickGO/term/GO:0048022</a> )	GO - Biological Process		
GO:0008643 : carbohydrate transport ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008643">https://www.ebi.ac.uk/QuickGO/term/GO:0008643</a> )			

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](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](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](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](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 ( $P = 2.08 \times 10^{-10}$ ). 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+T axon ID=~9796^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+T+axon+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/>