

Agouti (https://www.gephebase.org/search-criteria?and+GeneGephebase=^Agouti^#gephebase-summary-title)	Gephebase Gene	GP00000062	GepheID
Published	Entry Status	Martin	Main curator

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)		Trait Category	
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+coat^#gephebase-summary-title)		Trait	
Equus caballus		Trait State in Taxon A	
Equus caballus - modern breed and predomestic fossils		Trait State in Taxon B	
Taxon A		Ancestral State	
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus+caballus^#gephebase-summary-title)		Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus+caballus^#gephebase-summary-title)	
Common Name		Common Name	
horse		horse	
Synonyms		Synonyms	
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758		Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	
Rank		Rank	
species		species	
Lineage		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		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		Parent	
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)		Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)		9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		Yes	
		Taxon B Description	
		Equus caballus - modern breed and predomestic fossils	

Asip	Generic Gene Name	UniProtKB Mus musculus
	Synonyms	Q03288 (http://www.uniprot.org/uniprot/Q03288)
As; ASP; A<y>; ASIP; a	String	GenebankID or UniProtKB
10090.ENSMUSP00000029123 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)		AAG01635 (https://www.ncbi.nlm.nih.gov/nuccore/AAG01635)
-	Sequence Similarities	
GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779)	GO - Molecular Function	
GO:0031781 : type 3 melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031781)		

GO:0031782 : type 4 melanocortin receptor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031782>)

GO - Biological Process

GO:0008343 : adult feeding behavior
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008343>)
GO:0006091 : generation of precursor metabolites and energy
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006091>)
GO:0071514 : genetic imprinting (<https://www.ebi.ac.uk/QuickGO/term/GO:0071514>)
GO:0009755 : hormone-mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009755>)
GO:0042438 : melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
GO:0032438 : melanosome organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)
GO:0032402 : melanosome transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032402>)
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
GO:0048023 : positive regulation of melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)
GO:0040030 : regulation of molecular function, epigenetic
(<https://www.ebi.ac.uk/QuickGO/term/GO:0040030>)

GO - Cellular Component

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
GO:0005623 : cell (<https://www.ebi.ac.uk/QuickGO/term/GO:0005623>)

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes~#gephebase-summary-title)	Presumptive Null
	Molecular Type
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title)	Aberration Type
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion~#gephebase-summary-title)	Deletion Size
10-99 bp	Molecular Details of the Mutation
11bp deletion in exon 2: g.2174_2184del11 c.191_201del	
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title)	Experimental Evidence
Mutations in the agouti (ASIP), the extension (MC1R), and the brown (TYRP1) loci and their association to coat color phenotypes in horses (Equus caballus). (2001) (https://pubmed.ncbi.nlm.nih.gov/11353392)	Main Reference
Rieder S; Taourit S; Mariat D; Langlois B; GuÃ©rin G	Authors
Coat color genetics, when successfully adapted and applied to different mammalian species, provides a good demonstration of the powerful concept of comparative genetics. Using cross-species techniques, we have cloned, sequenced, and characterized equine melanocortin-1-receptor (MC1R) and agouti-signaling-protein (ASIP), and completed a partial sequence of tyrosinase-related protein 1 (TYRP1). The coding sequences and parts of the flanking regions of those genes were systematically analyzed in 40 horses and mutations typed in a total of 120 horses. Our panel represented 22 different horse breeds, including 11 different coat colors of Equus caballus. The comparison of a 1721-bp genomic fragment of MC1R among the 11 coat color phenotypes revealed no sequence difference apart from the known chestnut allele (C901T). In particular, no dominant black (ED) mutation was found. In a 4994-bp genomic fragment covering the three putative exons, two introns and parts of the 5'- and 3'-UTRs of ASIP, two intronic base substitutions (SNP-A845G and C2374A), a point mutation in the 3'-UTRs (A4734G), and an 11-bp deletion in exon 2 (ADEx2) were detected. The deletion was found to be homozygous and completely associated with horse recessive black coat color (Aa/Aa) in 24 black horses out of 9 different breeds from our panel. The frameshift initiated by ADEx2 is believed to alter the regular coding sequence, acting as a loss-of-function ASIP mutation. In TYRP1 a base substitution was detected in exon 2 (C189T), causing a threonine to methionine change of yet unknown function, and an SNP (A1188G) was found in intron 2.	Abstract
Genotypes of predomestic horses match phenotypes painted in Paleolithic works of cave art. (2011) (https://pubmed.ncbi.nlm.nih.gov/22065780)	Additional References

RELATED GEPHE

13 (Endothelin receptor B, Kit (type III receptor protein-tyrosine kinase), MC1R, MFSD12, 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+Taxon ID=~9796~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA000201/9796/>

