

# GEPHE SUMMARY

	Gephebase Gene	GephelD
Agouti ( <a href="https://www.gephebase.org/search-criteria/?and+Gene">https://www.gephebase.org/search-criteria/?and+Gene</a> Gephebase="Agouti">#gephebase-summary-title)	GP00000062	
	Entry Status	Main curator
Published	Martin	

## PHENOTYPIC CHANGE

	Trait Category	
Morphology ( <a href="https://www.gephebase.org/search-criteria/?and+Trait">https://www.gephebase.org/search-criteria/?and+Trait</a> Category="Morphology">#gephebase-summary-title)	Trait	
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria/?and+Trait=^Coloration">https://www.gephebase.org/search-criteria/?and+Trait=^Coloration</a> (coat)#gephebase-summary-title)	Trait State in Taxon A	
Equus caballus	Trait State in Taxon B	
Equus caballus - modern breed and predomestic fossils	Ancestral State	
Taxon A	Taxonomic Status	
Intraspecific ( <a href="https://www.gephebase.org/search-criteria/?and+Taxonomic">https://www.gephebase.org/search-criteria/?and+Taxonomic</a> Status="Intraspecific">#gephebase-summary-title)		
Taxon A		Taxon B
	Latin Name	Latin Name
Equus caballus ( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Equus+caballus">#gephebase-summary-title)</a>	Equus caballus ( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Equus+caballus">#gephebase-summary-title)</a>	
horse	Common Name	Common Name
	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	
species	Rank	Rank
	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) ( <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> )	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> )	
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 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> )
No	is Taxon A an Infraspecies?	is Taxon B an Infraspecies?
	Yes	Yes
		Taxon B Description
		Equus caballus - modern breed and predomestic fossils

## GENOTYPIC CHANGE

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

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>)

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

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

Deletion Size

10-99 bp

Molecular Details of the Mutation

11bp deletion in exon 2: g.2174\_2184del11 c.191\_201del

Experimental Evidence

Linkage Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=%27Linkage Mapping%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental%20Evidence=%27Linkage%20Mapping%27#gephebase-summary-title))

Main Reference

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>)

Authors

Rieder S; Taourit S; Mariat D; Langlois B; Guérin G

Abstract

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.

Additional References

Genotypes of predomestic horses match phenotypes painted in Paleolithic works of cave art. (2011) (<https://pubmed.ncbi.nlm.nih.gov/22065780>)

## RELATED GEPHE

#### Related Genes

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+TaxonID=%279796%27/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

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

