

GEPHE SUMMARY

	Gephebase Gene		GepheID
syntxin-17 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~syntxin-17^#gephebase-summary-title)		GP00002272	
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology^#gephebase-summary-title)			
	Trait		
Coloration (hair ; graying with age) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+(hair+;+graying+with+age)^#gephebase-summary-title)			
WT coat with late or no greying	Trait State in Taxon A		
Premature hair graying and susceptibility to melanoma	Trait State in Taxon B		
	Ancestral State		
Taxon A		Taxonomic Status	
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated^#gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		Latin Name
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 Infrappecies?		is Taxon B an Infrappecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
STX17		P56962 (http://www.uniprot.org/uniprot/P56962)	
	Synonyms		GenebankID or UniProtKB
-		()	
	String		
9606.ENSP00000259400 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000259400)			
	Sequence Similarities		
Belongs to the syntxin family.			
	GO - Molecular Function		
GO:0019901 : protein kinase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019901)			
GO:0019903 : protein phosphatase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019903)			
GO:0000149 : SNARE binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000149)			
GO:0005484 : SNAP receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005484)			

GO:0006886 : intracellular protein transport
<https://www.ebi.ac.uk/QuickGO/term/GO:0006886>
 GO:0007030 : Golgi organization (<https://www.ebi.ac.uk/QuickGO/term/GO:0007030>)
 GO:0006888 : ER to Golgi vesicle-mediated transport
<https://www.ebi.ac.uk/QuickGO/term/GO:0006888>
 GO:0097352 : autophagosome maturation
<https://www.ebi.ac.uk/QuickGO/term/GO:0097352>
 GO:0016240 : autophagosome membrane docking
<https://www.ebi.ac.uk/QuickGO/term/GO:0016240>
 GO:0097111 : endoplasmic reticulum-Golgi intermediate compartment organization
<https://www.ebi.ac.uk/QuickGO/term/GO:0097111>
 GO:0034497 : protein localization to phagophore assembly site
<https://www.ebi.ac.uk/QuickGO/term/GO:0034497>
 GO:0048278 : vesicle docking (<https://www.ebi.ac.uk/QuickGO/term/GO:0048278>)
 GO:0006906 : vesicle fusion (<https://www.ebi.ac.uk/QuickGO/term/GO:0006906>)

GO - Cellular Component

GO:0016021 : integral component of membrane
<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>
 GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
 GO:0005739 : mitochondrion (<https://www.ebi.ac.uk/QuickGO/term/GO:0005739>)
 GO:0005789 : endoplasmic reticulum membrane
<https://www.ebi.ac.uk/QuickGO/term/GO:0005789>
 GO:0012507 : ER to Golgi transport vesicle membrane
<https://www.ebi.ac.uk/QuickGO/term/GO:0012507>
 GO:0012505 : endomembrane system
<https://www.ebi.ac.uk/QuickGO/term/GO:0012505>
 GO:0005776 : autophagosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0005776>)
 GO:0031201 : SNARE complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0031201>)
 GO:0000421 : autophagosome membrane
<https://www.ebi.ac.uk/QuickGO/term/GO:0000421>
 GO:0030134 : COPII-coated ER to Golgi transport vesicle
<https://www.ebi.ac.uk/QuickGO/term/GO:0030134>
 GO:0005793 : endoplasmic reticulum-Golgi intermediate compartment
<https://www.ebi.ac.uk/QuickGO/term/GO:0005793>
 GO:0033116 : endoplasmic reticulum-Golgi intermediate compartment membrane
<https://www.ebi.ac.uk/QuickGO/term/GO:0033116>
 GO:0044233 : mitochondria-associated endoplasmic reticulum membrane
<https://www.ebi.ac.uk/QuickGO/term/GO:0044233>
 GO:0030868 : smooth endoplasmic reticulum membrane
<https://www.ebi.ac.uk/QuickGO/term/GO:0030868>

Presumptive Null

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null="+No+"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))

Molecular Type

Cis-regulatory ([https://www.gephebase.org/search-criteria?/and+Molecular Type="+Cis-regulatory+"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=))

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 Size

1-10 kb

Molecular Details of the Mutation

4.6-kb duplication in intron 6 of STX17 ; in transgenic zebrafish a construct containing two copies of the duplicated sequence acts as a strong enhancer in neural crest cells and has subsequent melanophore-specific activity whereas a single copy of the duplicated sequence acts as a weak enhancer - consistent with the phenotypic manifestation of the mutation in horses

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=))

Main Reference

A cis-acting regulatory mutation causes premature hair graying and susceptibility to melanoma in the horse. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18641652>)

Authors

Rosengren Pielberg G; Golovko A; Sundström E; Curik I; Lennartsson J; Seltenhammer MH; Druml T; Binns M; Fitzsimmons C; Lindgren G; Sandberg K; Baumung R; Vetterlein M; Strömberg S; Grabherr M; Wade C; Lindblad-Toh K; Pontón F; Heldin CH; Sällkner J; Andersson L

Abstract

In horses, graying with age is an autosomal dominant trait associated with a high incidence of melanoma and vitiligo-like depigmentation. Here we show that the Gray phenotype is caused by a 4.6-kb duplication in intron 6 of STX17 (syntaxin-17) that constitutes a cis-acting regulatory mutation. Both STX17 and the neighboring NR4A3 gene are overexpressed in melanomas from Gray horses. Gray horses carrying a loss-of-function mutation in ASIP (agouti signaling protein) had a higher incidence of melanoma, implying that increased melanocortin-1 receptor signaling promotes melanoma development in Gray horses. The Gray horse provides a notable example of how humans have cherry-picked mutations with favorable phenotypic effects in domestic animals.

Additional References

Copy number expansion of the STX17 duplication in melanoma tissue from Grey horses. (2012) (<https://pubmed.ncbi.nlm.nih.gov/22857264>)

Identification of a melanocyte-specific, microphthalmia-associated transcription factor-dependent regulatory element in the intronic duplication causing hair greying and melanoma in horses. (2012) (<https://pubmed.ncbi.nlm.nih.gov/21883983>)

RELATED GEPHE

Related Genes

13 (Agouti, Endothelin receptor B, Kit (type III receptor protein-tyrosine kinase), MC1R, MFSD12, Microphthalmia-associated transcription factor, Pax3, PMEL17, SLC24A, SLC36A1, SLC45A2=MATP, 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](https://www.gephebase.org/search-criteria?/or+Taxon+ID=))

No matches found.

EXTERNAL LINKS

COMMENTS

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