

GEPHE SUMMARY

	Gephebase Gene	GephelD
KRT25 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^KRT25^#gephebase-summary-title)	GP00002288	Main curator
Published	Entry Status	Martin

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)	Trait		
Hair type (curly) (https://www.gephebase.org/search-criteria?/and+Trait=^Hair+type (curly)^#gephebase-summary-title)	Trait State in Taxon A		
Straight hair	Trait State in Taxon B		
Curly Hair including in Bashkir Curly Horse and Missouri Foxtrotter	Ancestral State		
Taxon A	Taxonomic Status		
Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus+caballus^#gephebase-summary-title)	Latin Name		Latin Name
horse	Common Name		Common Name
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	Synonyms		Synonyms
species	Rank		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		Lineage
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)	Parent		Parent
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)	NCBI Taxonomy ID		NCBI Taxonomy ID
No	is Taxon A an Infraspecies?		is Taxon B an Infraspecies?

GENOTYPIC CHANGE

Sp6	Generic Gene Name	UniProtKB Mus musculus
Epfn; Klf14; AA591031; Al592962; 1110025J03Rik	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000039307 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000039307)	String	
Belongs to the Sp1 C2H2-type zinc-finger protein family.	Sequence Similarities	
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872) GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677) GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)	GO - Molecular Function	
GO:0006357 : regulation of transcription by RNA polymerase II	GO - Biological Process	

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006357>)

GO:0006366 : transcription by RNA polymerase II

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006366>)

GO:0042481 : regulation of odontogenesis

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042481>)

GO - Cellular Component

GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)

GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)

Presumptive Null

No (<https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^No^#gephebase-summary-title>)

Molecular Type

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular+Type=^Coding^#gephebase-summary-title>)

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration+Type=^SNP^#gephebase-summary-title>)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.1090G>A p.Asp364Asn

Experimental Evidence

Association Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=^Association+Mapping^#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Asp	Asn	364

Main Reference

An epistatic effect of KRT25 on SP6 is involved in curly coat in horses. (2018) (<https://pubmed.ncbi.nlm.nih.gov/29686323>)

Authors

Thomer A; Gottschalk M; Christmann A; Naccache F; Jung K; Hewicker-Trautwein M; Distl O; Metzger J

Abstract

Curly coat represents an extraordinary type of coat in horses, particularly seen in American Bashkir Curly Horses and Missouri Foxtrotters. In some horses with curly coat, a hypotrichosis of variable extent was observed, making the phenotype appear more complex. In our study, we aimed at investigating the genetic background of curly coat with and without hypotrichosis using high density bead chip genotype and next generation sequencing data. Genome-wide association analysis detected significant signals ($P = 1.412 \times 10^{-11}$) on horse chromosome 11 at 22-35 Mb. In this significantly associated region, six missense variants were filtered out from whole-genome sequencing data of three curly coated horses of which two variants within KRT25 and SP6 could explain all hair phenotypes. Horses heterozygous or homozygous only for KRT25 variant showed curly coat and hypotrichosis, whereas horses with SP6 variant only, exhibited curly coat without hypotrichosis. Horses with mutant alleles in both variants developed curly hair and hypotrichosis. Thus, mutant KRT25 allele is masking SP6 allele effect, indicative for epistasis of KRT25 variant over SP6 variant. In summary, genetic variants in two different genes, KRT25 and SP6, are responsible for curly hair. All horses with KRT25 variant are additionally hypotrichotic due to the KRT25 epistatic effect on SP6.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

1 (<https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=^KRT25^/and+Taxon+ID=^9796^/or+Gene+Gephebase=^KRT25^/and+Taxon+ID=^9796^#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

@Epistasis <https://omia.org/OMIA002175/9796>