GEPHE SUMMARY

Gephebase Gene
KRT25 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^KRT25^#gephebase-summary-title)

GepheID
GP00002288

Entry Status
Published

Main curator
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
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Equus caballus^#gephebase-summary-title)

Common Name
horse

Synonyms
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758

Rank
species

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

Parent
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)

NCBI Taxonomy ID
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Equus caballus^#gephebase-summary-title)

Common Name
horse

Synonyms
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758

Rank
species

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

Parent
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)

NCBI Taxonomy ID
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Sp6

Synonyms
Epfn; Klf14; AA591031; AI592962; 1110025J03Rik

String
10090.ENSMUSP00000039307 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000039307)

Sequence Similarities
Belongs to the Sp1 C2H2-type zinc-finger protein family.

GO - Molecular Function
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 - Biological Process
GO:0006357 : regulation of transcription by RNA polymerase II

UniProtKB Mus musculus
Q9ESX2 (http://www.uniprot.org/uniprot/Q9ESX2)

GenebankID or UniProtKB
()

(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 (pâ€‰=â€‰1.412â€‰Ã—â€‰10-1.102â€‰Ã—â€‰10) on horse chromosome 11 at 22-35â€‰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/