

GEPHE SUMMARY

<p>KRT25 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~KRT25~#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002288</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

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

<p>Sp6</p> <p>Epfm; Klf14; AA591031; A1592962; 1110025J03Rik</p> <p>10090.ENSMUSP00000039307 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000039307)</p> <p>Belongs to the Sp1 C2H2-type zinc-finger protein family.</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> <p>GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)</p> <p>GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)</p> <p>GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)</p> <p>GO - Biological Process</p> <p>GO:0006357 : regulation of transcription by RNA polymerase II</p>	<p>UniProtKB Mus musculus</p> <p>Q9ESX2 (http://www.uniprot.org/uniprot/Q9ESX2)</p> <p>GenebankID or UniProtKB</p> <p>()</p>
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(<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 ($p \leq 1.412 \times 10^{-10}$ – 1.102×10^{-10}) 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/>