GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00002230

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 ; dominant

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

Krt25

Synonyms

K25; Ka38; CK-25; Krt25a; mIRSa1; 4631426H08Rik

String

10090.ENSMUSP00000048439 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000048439)

Sequence Similarities

Belongs to the intermediate filament family.

GO - Molecular Function

GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)
GO:0005198 : structural molecule activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005198)

GO - Biological Process

GO:0007010 : cytoskeleton organization

UniProtKB Mus musculus

Q8VCW2 (http://www.uniprot.org/uniprot/Q8VCW2)

GenebankID or UniProtKB

()

(https://www.ebi.ac.uk/QuickGO/term/GO:0007010)
GO:0007568 : aging (https://www.ebi.ac.uk/QuickGO/term/GO:0007568)
GO:0031069 : hair follicle morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0031069)
GO:0045109 : intermediate filament organization
(https://www.ebi.ac.uk/QuickGO/term/GO:0045109)
GO:0042633 : hair cycle (https://www.ebi.ac.uk/QuickGO/term/GO:0042633)

GO - Cellular Component

GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0005882 : intermediate filament (https://www.ebi.ac.uk/QuickGO/term/GO:0005882)

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.266G>A p.Arg89His

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	Arg	His	89

Main Reference

A missense variant in the coil1A domain of the keratin 25 gene is associated with the dominant curly hair coat trait (Crd) in horse. (2017) (https://pubmed.ncbi.nlm.nih.gov/29141579)

Authors

Morgenthaler C; Diribarne M; Capitan A; Legendre R; Saintilan R; Gilles M; EsquerrÃ© D; Juras R; Khanshour A; Schibler L; Cothran G

Abstract

Curly horses present a variety of curl phenotypes that are associated with various degrees of curliness of coat, mane, tail and ear hairs. Their origin is still a matter of debate and several genetic hypotheses have been formulated to explain the diversity in phenotype, including the combination of autosomal dominant and recessive alleles. Our purpose was to map the autosomal dominant curly hair locus and identify the causal variant using genome-wide association study (GWAS) and whole-genome sequencing approaches.

A GWAS was performed using a Bayesian sparse linear mixed model, based on 51 curly and 19 straight-haired French and North American horses from 13 paternal families genotyped on the Illumina EquineSNP50 BeadChip. A single strong signal was observed on equine chromosome 11, in a region that encompasses the type I keratin gene cluster. This region was refined by haplotype analysis to a segment including 36 genes, among which are 10 keratin genes (KRT-10, -12, -20, -23, -24, -25, -26, -27, -28, -222). To comprehensively identify candidate causal variants within all these genes, whole-genome sequences were obtained for one heterozygous curly stallion and its straight-haired son. Among the four non-synonymous candidate variants identified and validated in the curly region, only variant g.21891160G>A in the KRT25 gene (KRT25:p.R89H) was in perfect agreement with haplotype status in the whole pedigree. Genetic association was then confirmed by genotyping a larger population consisting of 353 horses. However, five discordant curly horses were observed, which carried neither the variant nor the main haplotype associated with curliness. Sequencing of KRT25 for two discordant horses did not identify any other deleterious variant, which suggests locus rather than allelic heterogeneity for the curly phenotype.

We identified the KRT25:p.R89H variant as responsible for the dominant curly trait, but a second dominant locus may also be involved in the shape of hairs within North American Curly horses.

Additional References

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

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

@Dominance https://omia.org/OMIA000245/9796/