

GEPHE SUMMARY

KRT25 (#gephebase-summary-title)	Gephebase Gene	GP00002230	GephelD
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Trait Category			
Morphology (#gephebase-summary-title)	Trait		
Hair type (curly) (<a ?and+taxon+and+synonyms='^Equus+caballus"' href="https://www.gephebase.org/search-criteria/?and+Trait=^Hair+type(curly)#gephebase-summary-title)</td><td>Trait State in Taxon A</td><td></td><td></td></tr> <tr> <td>Straight hair</td><td>Trait State in Taxon B</td><td></td><td></td></tr> <tr> <td>Curly Hair including in Bashkir Curly Horse ; dominant</td><td>Ancestral State</td><td></td><td></td></tr> <tr> <td>Taxon A</td><td>Taxonomic Status</td><td></td><td></td></tr> <tr> <td>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

Krt25	Generic Gene Name	UniProtKB Mus musculus
K25; Ka38; CK-25; Krt25a; mIRSa1; 4631426H08Rik	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000048439 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000048439)	String	
Belongs to the intermediate filament family.	Sequence Similarities	
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)	GO - Molecular Function	
GO:0005198 : structural molecule activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005198)	GO - Biological Process	
GO:0007010 : cytoskeleton organization		

(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)		Presumptive Null
GO:0005882 : intermediate filament (https://www.ebi.ac.uk/QuickGO/term/GO:0005882)		
No (https://www.gephebase.org/search-criteria/?and+Presumptive Null=%22No%22#gephebase-summary-title)		Molecular Type
Coding (https://www.gephebase.org/search-criteria/?and+Molecular Type=%22Coding%22#gephebase-summary-title)		Aberration Type
SNP (https://www.gephebase.org/search-criteria/?and+Aberration Type=%22SNP%22#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=%22Association Mapping%22#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; Esquerré 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=%22KRT25%22 /and+Taxon ID=%229796%22 /or+Gene Gephebase=%22KRT25%22 /and+Taxon ID=%229796%22#gephebase-summary-title](https://www.gephebase.org/search-criteria/?or+Gene%20Gephebase=%22KRT25%22%20and%20Taxon%20ID=%229796%22%20or+Gene%20Gephebase=%22KRT25%22%20and%20Taxon%20ID=%229796%22#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

@Dominance <https://omnia.org/OMIA000245/9796/>

