

GEPHE SUMMARY

DMRT3 (https://www.gephebase.org/search-criteria/?and+Gene+Gephebase=%DMRT3%#gephebase-summary-title)	Gephebase Gene	GP00000230	GephelD
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Behavior (https://www.gephebase.org/search-criteria/?and+Trait+Category=%Behavior%#gephebase-summary-title)	Trait		
Gait (ability to pace) (https://www.gephebase.org/search-criteria/?and+Trait=%Gait+(ability+to+pace)%#gephebase-summary-title)	Trait State in Taxon A		
Equus caballus	Trait State in Taxon B		
Equus caballus	Ancestral State		
Data not curated	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria/?and+Taxonomic+Status=%Domesticated%#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Equus caballus (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Equus+caballus%#gephebase-summary-title)		Equus caballus (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Equus+caballus%#gephebase-summary-title)	
horse	Common Name	horse	Common Name
	Synonyms		Synonyms
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758		Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	
Rank		Rank	
species	Lineage	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		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		Parent	
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)		Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)	
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)	NCBI Taxonomy ID	9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

DMRT3	Generic Gene Name	UniProtKB Equus caballus
-	Synonyms	GenebankID or UniProtKB
9796.ENSECAP00000020841 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9796.ENSECAP00000020841)	String	JQ922371 (https://www.ncbi.nlm.nih.gov/nuccore/JQ922371)
Belongs to the DMRT family.	Sequence Similarities	
GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)	GO - Molecular Function	
GO:0043565 : sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043565)		
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 - Biological Process

GO:0007548 : sex differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0007548>)GO:0008344 : adult locomotory behavior
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008344>)GO:0019226 : transmission of nerve impulse
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019226>)GO:0021521 : ventral spinal cord interneuron specification
(<https://www.ebi.ac.uk/QuickGO/term/GO:0021521>)

GO - Cellular Component

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

Presumptive Null

Yes ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^Yes))

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

g.22999655C>A p.S301* Premature stop codon (Ser301Stop) resulting in truncated protein: a single base change at nucleotide position chr23:22999655 causing a premature stop at codon 301 in DMRT3 (DMRT3_Ser301STOP). The allele is expected to encode a truncated protein lacking 1

Experimental Evidence

Linkage Mapping ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=^Linkage+Mapping))

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ser	STP	301

Main Reference

Mutations in DMRT3 affect locomotion in horses and spinal circuit function in mice. (2012) (<https://pubmed.ncbi.nlm.nih.gov/22932389>)

Authors

Andersson LS; Larhammar M; Memic F; Wootz H; Schwochow D; Rubin CJ; Patra K; Arnason T; Wellbring L; HjÃ¶lm G; Imsland F; Petersen JL; McCue ME; Mickelson JR; Cothran G; Ahituv N; Roepstorff L; Mikko S; Vallstedt A; Lindgren G; Andersson L; Kullander K

Abstract

Locomotion in mammals relies on a central pattern-generating circuitry of spinal interneurons established during development that coordinates limb movement. These networks produce left-right alternation of limbs as well as coordinated activation of flexor and extensor muscles. Here we show that a premature stop codon in the DMRT3 gene has a major effect on the pattern of locomotion in horses. The mutation is permissive for the ability to perform alternate gaits and has a favourable effect on harness racing performance. Examination of wild-type and Dmrt3-null mice demonstrates that Dmrt3 is expressed in the dl6 subdivision of spinal cord neurons, takes part in neuronal specification within this subdivision, and is critical for the normal development of a coordinated locomotor network controlling limb movements. Our discovery positions Dmrt3 in a pivotal role for configuring the spinal circuits controlling stride in vertebrates. The DMRT3 mutation has had a major effect on the diversification of the domestic horse, as the altered gait characteristics of a number of breeds apparently require this mutation.

Additional References

Genome-wide analysis reveals selection for important traits in domestic horse breeds. (2013) (<https://pubmed.ncbi.nlm.nih.gov/23349635>)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001715/9796/>

