

## GEPHE SUMMARY

<p>DMRT3 (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^DMRT3^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^DMRT3^#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00000230</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

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

## GENOTYPIC CHANGE

<p>DMRT3</p>	<p>Generic Gene Name</p>	<p>F6W2R2 (<a href="http://www.uniprot.org/uniprot/F6W2R2">http://www.uniprot.org/uniprot/F6W2R2</a>)</p>	<p>UniProtKB Equus caballus</p>
<p>-</p>	<p>Synonyms</p>	<p>JQ922371 (<a href="https://www.ncbi.nlm.nih.gov/nuccore/JQ922371">https://www.ncbi.nlm.nih.gov/nuccore/JQ922371</a>)</p>	<p>GenebankID or UniProtKB</p>
<p>9796.ENSECAP00000020841 (<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9796.ENSECAP00000020841">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9796.ENSECAP00000020841</a>)</p>	<p>String</p>		
<p>Belongs to the DMRT family.</p>	<p>Sequence Similarities</p>		
<p>GO:0003700 : DNA-binding transcription factor activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003700">https://www.ebi.ac.uk/QuickGO/term/GO:0003700</a>)</p> <p>GO:0043565 : sequence-specific DNA binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043565">https://www.ebi.ac.uk/QuickGO/term/GO:0043565</a>)</p> <p>GO:0046872 : metal ion binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046872">https://www.ebi.ac.uk/QuickGO/term/GO:0046872</a>)</p> <p>GO:0003677 : DNA binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003677">https://www.ebi.ac.uk/QuickGO/term/GO:0003677</a>)</p>	<p>GO - Molecular Function</p>		

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

Molecular Type

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

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

	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; Hjälm 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

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

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

