

GEPHE SUMMARY

	Gephebase Gene		GepheID
aggrecan (<a +aggrecan+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+aggrecan+"#gephebase-summary-title)		GP00002142	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)		Trait	
Body size (dwarfism) (<a +body+size+(dwarfism)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Body+size+(dwarfism)+"#gephebase-summary-title)			
	Trait State in Taxon A		
Miniature horses		Trait State in Taxon B	
Miniature horses with dwarf phenotype (chondrodysplasia)			
	Ancestral State		
Taxon A		Taxonomic Status	
Domesticated (<a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated+"#gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Equus caballus (<a +equus+caballus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Equus+caballus+"#gephebase-summary-title)		Equus caballus (<a +equus+caballus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Equus+caballus+"#gephebase-summary-title)	
	Common Name		Common Name
horse		horse	
	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		species	
	Lineage		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)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)		9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
ACAN		P16112 (http://www.uniprot.org/uniprot/P16112)	
	Synonyms		GenebankID or UniProtKB
AGC1; SEDK; AGCAN; CSPG1; MSK16; CSPGCP; SSOAOD		()	
	String		
9606.ENSP00000387356 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000387356)			
	Sequence Similarities		
Belongs to the aggrecan/versican proteoglycan family.			
	GO - Molecular Function		
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)			
GO:0030246 : carbohydrate binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030246)			
GO:0005201 : extracellular matrix structural constituent (https://www.ebi.ac.uk/QuickGO/term/GO:0005201)			
GO:0030021 : extracellular matrix structural constituent conferring compression resistance (https://www.ebi.ac.uk/QuickGO/term/GO:0030021)			

GO:0005540 : hyaluronic acid binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005540>)

GO - Biological Process

GO:0007155 : cell adhesion (<https://www.ebi.ac.uk/QuickGO/term/GO:0007155>)
GO:0007417 : central nervous system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007417>)
GO:0001502 : cartilage condensation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001502>)
GO:0002063 : chondrocyte development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002063>)
GO:0030199 : collagen fibril organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030199>)
GO:0030198 : extracellular matrix organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030198>)
GO:0007507 : heart development (<https://www.ebi.ac.uk/QuickGO/term/GO:0007507>)
GO:0018146 : keratan sulfate biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0018146>)
GO:0042340 : keratan sulfate catabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042340>)
GO:0030166 : proteoglycan biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030166>)
GO:0006508 : proteolysis (<https://www.ebi.ac.uk/QuickGO/term/GO:0006508>)
GO:0001501 : skeletal system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001501>)

GO - Cellular Component

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
GO:0062023 : collagen-containing extracellular matrix
(<https://www.ebi.ac.uk/QuickGO/term/GO:0062023>)
GO:0005604 : basement membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005604>)
GO:0031012 : extracellular matrix (<https://www.ebi.ac.uk/QuickGO/term/GO:0031012>)
GO:0098982 : GABA-ergic synapse (<https://www.ebi.ac.uk/QuickGO/term/GO:0098982>)
GO:0098978 : glutamatergic synapse
(<https://www.ebi.ac.uk/QuickGO/term/GO:0098978>)
GO:0005796 : Golgi lumen (<https://www.ebi.ac.uk/QuickGO/term/GO:0005796>)
GO:0043202 : lysosomal lumen (<https://www.ebi.ac.uk/QuickGO/term/GO:0043202>)
GO:0098966 : perisynaptic extracellular matrix
(<https://www.ebi.ac.uk/QuickGO/term/GO:0098966>)

	Presumptive Null
Yes (#gephebase-summary-title)	
	Molecular Type
Coding (#gephebase-summary-title)	
	Aberration Type
Deletion (#gephebase-summary-title)	
	Deletion Size
10-99 bp	
	Molecular Details of the Mutation
g.95257458_95257500del ; p.Phe2017-Asp2023del	
	Experimental Evidence
Association Mapping (#gephebase-summary-title)	
	Main Reference
Multiple alleles of ACAN associated with chondrodysplastic dwarfism in Miniature horses. (2018) (https://pubmed.ncbi.nlm.nih.gov/30058072)	
	Authors
Eberth JE; Graves KT; MacLeod JN; Bailey E	

Abstract

Chondrodysplastic dwarfism in Miniature horses appeared to be a recessive genetic trait based on the occurrence of affected offspring by normal parents. Dwarf phenotypes vary and range from abnormal abortions to viable offspring with evidence of skeletal dysplasia. A genome-wide association study implicated a region of ECA1 with dwarfism in Miniature horses. Aggrecan (ACAN) was a candidate gene in that region, and exons were sequenced to compare DNA sequences for dwarf and non-dwarf horses. Sequencing led to the discovery of variants in exons 2, 6, 7 and 15 associated with dwarfism. The four variants are identified with reference to Ecab 3.0 (GCF_002863925.1) as [g.95291270del](#) ([rs1095048841](#)), [g.95284530C>T](#) ([ERP107353](#)), [g.95282140C>G](#) ([rs1095048823](#)) and [g.95257480_95257500del](#) ([rs1095048839](#)) and designated here as D1, D2, D3* and D4 respectively. A previous study at another laboratory reported dwarfism associated with homozygosity for D3*. Homozygotes for those variants and compound heterozygotes for any combination of those variants always expressed a dwarfism phenotype. However, eight additional horses with dwarfism were found, seven of which were heterozygotes for D2, D3* or D4, suggesting the existence of additional ACAN alleles causing dwarfism. Among Miniature horses, the combined frequency of D1, D2, D3* and D4 was 0.163, suggesting a carrier rate of 26.2% for alleles causing chondrodysplastic dwarfism.

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Additional References

RELATED GEPHE

	Related Genes
3 (B4GALT7 , HMG2 , LCORL) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~9796#/and+Trait=Body size/and+groupHaplotypes=true#gephebase-summary-title)	
	Related Haplotypes
3 (#gephebase-summary-title)	

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001271/9796/> @AllelicSeries @HeterozygoteAdvantage