

GEPHE SUMMARY

aggrecan (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^aggrecan^#gephebase-summary-title)	Gephebase Gene	GP00002144	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)	Trait Category		
Body size (dwarfism) (https://www.gephebase.org/search-criteria?/and+Trait=^Body size (dwarfism)^#gephebase-summary-title)	Trait		
Miniature horses	Trait State in Taxon A		
Miniature horses with dwarf phenotype (chondrodysplasia)	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus caballus^#gephebase-summary-title)	Latin Name	Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus caballus^#gephebase-summary-title)	Latin Name
horse	Common Name	horse	Common Name
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	Synonyms	Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	Synonyms
species	Rank	species	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	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
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)	Parent	Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)	Parent
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 Infrappecies?	No	is Taxon B an Infrappecies?

GENOTYPIC CHANGE

ACAN	Generic Gene Name	P16112 (http://www.uniprot.org/uniprot/P16112)	UniProtKB Homo sapiens
AGC1; SEDK; AGCAN; CSPG1; MSK16; CSPGCP; SSOAOD	Synonyms	()	GenebankID or UniProtKB
9606.ENSPP0000387356 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP0000387356)	String		
Belongs to the aggrecan/versican proteoglycan family.	Sequence Similarities		
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)	GO - Molecular Function		
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>)

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null="No" #gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive Null=)) Presumptive Null
Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type="Coding" #gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular Type=)) Molecular Type
SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type="SNP" #gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration Type=)) Aberration Type
Nonsynonymous SNP Coding Change
g.95284530C>T ; p.Val424Met Molecular Details of the Mutation
Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence="Association Mapping" #gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental Evidence=)) Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Val	Met	424

Multiple alleles of ACAN associated with chondrodysplastic dwarfism in Miniature horses. (2018) (<https://pubmed.ncbi.nlm.nih.gov/30058072>) Main Reference
Eberth JE; Graves KT; MacLeod JN; Bailey E Authors

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 abortuses 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

3 (B4GALT7, HMGA2, LCORL) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9796^/and+Trait=Body size/and+groupHaplotypes=true#gephebase-summary-title>)

3 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^aggreca^/and+Taxon ID=^9796^/or+Gene Gephebase=^aggreca^/and+Taxon ID=^9796^#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

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