GEPHE SUMMARY

Gephebase Gene
aggrecan (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^aggrecan^#gephebase-summary-title)

GepheID
GP00002144

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Body size (dwarfism) (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 (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Equus caballus^#gephebase-summary-title)

Common Name
horse

Synonyms
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758

Rank
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

Parent
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)

NCBI Taxonomy ID
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Equus caballus^#gephebase-summary-title)

Common Name
horse

Synonyms
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758

Rank
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

Parent
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)

NCBI Taxonomy ID
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
ACAN

Synonyms
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)

UniProtKB Homo sapiens
P16112 (http://www.uniprot.org/uniprot/P16112)

GenebankID or UniProtKB
()

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

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Coding (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)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

g.95284530C>T ; p.Val424Met

Experimental Evidence

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

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

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 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

Related Genes

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

Related Haplotypes

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

EXTERNAL LINKS

COMMENTS

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