

GEPHE SUMMARY

Gephebase Gene		GepheID
T-box transcription factor (TBX3) (<a +t-box+transcription+factor+(tbx3)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+T-box+transcription+factor+(TBX3)+"#gephebase-summary-title)	GP00001462	
Entry Status	Prigent	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			
Coloration (coat) (<a +coloration+(coat)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+(coat)+"#gephebase-summary-title)			
Trait State in Taxon A			
Dun horse characterized by diluted pigment with dark primitive markings - radially asymmetric deposition of pigment in hair			
Trait State in Taxon B			
Non-dun horse intensely pigmented not associated with markings - circumferential distribution of melanocytes and pigment granules in hair			
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?	
Yes		Yes	
Taxon A Description		Taxon B Description	
Dun horse characterized by diluted pigment with dark primitive markings - radially asymmetric deposition of pigment in hair		Non-dun horse intensely pigmented not associated with markings - circumferential distribution of melanocytes and pigment granules in hair	

GENOTYPIC CHANGE

Generic Gene Name		UniProtKB Homo sapiens
TBX3		O15119 (http://www.uniprot.org/uniprot/O15119)
Synonyms		GenebankID or UniProtKB
UMS; XHL; TBX3-ISO		KT896510 (https://www.ncbi.nlm.nih.gov/nuccore/KT896510)
String		
9606.ENSPP00000257566 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000257566)		
Sequence Similarities		
-		
GO - Molecular Function		
GO:0043565 : sequence-specific DNA binding		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0043565>)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000981>)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)
GO:0001085 : RNA polymerase II transcription factor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001085>)
GO:0001078 : proximal promoter DNA-binding transcription repressor activity, RNA polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0001078>)
GO:0001102 : RNA polymerase II activating transcription factor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001102>)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043066>)
GO:0006357 : regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006357>)
GO:0001501 : skeletal system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001501>)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)
GO:0045892 : negative regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045892>)
GO:0045893 : positive regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045893>)
GO:0060021 : roof of mouth development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060021>)
GO:0008284 : positive regulation of cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008284>)
GO:0001701 : in utero embryonic development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001701>)
GO:0009887 : animal organ morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009887>)
GO:0001947 : heart looping (<https://www.ebi.ac.uk/QuickGO/term/GO:0001947>)
GO:0030539 : male genitalia development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030539>)
GO:0090398 : cellular senescence (<https://www.ebi.ac.uk/QuickGO/term/GO:0090398>)
GO:0045787 : positive regulation of cell cycle
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045787>)
GO:0060412 : ventricular septum morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060412>)
GO:0019827 : stem cell population maintenance
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019827>)
GO:0035116 : embryonic hindlimb morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035116>)
GO:2000648 : positive regulation of stem cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000648>)
GO:0035136 : forelimb morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035136>)
GO:0030879 : mammary gland development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030879>)
GO:0045662 : negative regulation of myoblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045662>)
GO:0035115 : embryonic forelimb morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035115>)
GO:0048332 : mesoderm morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048332>)
GO:0008595 : anterior/posterior axis specification, embryo
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008595>)
GO:0003167 : atrioventricular bundle cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003167>)
GO:0001568 : blood vessel development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001568>)
GO:0060444 : branching involved in mammary gland duct morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060444>)
GO:0060923 : cardiac muscle cell fate commitment
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060923>)
GO:0007569 : cell aging (<https://www.ebi.ac.uk/QuickGO/term/GO:0007569>)
GO:0042733 : embryonic digit morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042733>)
GO:0030540 : female genitalia development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030540>)
GO:0046884 : follicle-stimulating hormone secretion
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046884>)
GO:0021761 : limbic system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0021761>)
GO:0032275 : luteinizing hormone secretion
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032275>)
GO:0060596 : mammary placode formation

(<https://www.ebi.ac.uk/QuickGO/term/GO:0060596>)
GO:0030857 : negative regulation of epithelial cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030857>)
GO:0003151 : outflow tract morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003151>)
GO:0060931 : sinoatrial node cell development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060931>)
GO:0010159 : specification of animal organ position
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010159>)

GO - Cellular Component	
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)	Presumptive Null
No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title)	Molecular Type
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Cis-regulatory~#gephebase-summary-title)	Aberration Type
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion~#gephebase-summary-title)	Deletion Size
1-10 kb	Molecular Details of the Mutation
missing sequence of nearly contiguous 1609bp and 8 bp segments	
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title)	Experimental Evidence
Regulatory mutations in TBX3 disrupt asymmetric hair pigmentation that underlies Dun camouflage color in horses. (2016) (https://pubmed.ncbi.nlm.nih.gov/26691985)	Main Reference
Imsland F; McGowan K; Rubin CJ; Henegar C; Sundstr�m E; Berglund J; Schwochow D; Gustafson U; Imsland P; Lindblad-Toh K; Lindgren G; Mikko S; Millon L; Wade C; Schubert M; Orlando L; Penedo MC; Barsh GS; Andersson L	Authors

Abstract

Dun is a wild-type coat color in horses characterized by pigment dilution with a striking pattern of dark areas termed primitive markings. Here we show that pigment dilution in Dun horses is due to radially asymmetric deposition of pigment in the growing hair caused by localized expression of the T-box 3 (TBX3) transcription factor in hair follicles, which in turn determines the distribution of hair follicle melanocytes. Most domestic horses are non-dun, a more intensely pigmented phenotype caused by regulatory mutations impairing TBX3 expression in the hair follicle, resulting in a more circumferential distribution of melanocytes and pigment granules in individual hairs. We identified two different alleles (non-dun1 and non-dun2) causing non-dun color. non-dun2 is a recently derived allele, whereas the Dun and non-dun1 alleles are found in ancient horse DNA, demonstrating that this polymorphism predates horse domestication. These findings uncover a new developmental role for T-box genes and new aspects of hair follicle biology and pigmentation.

Additional References

RELATED GEPHE

13 (Agouti, Endothelin receptor B, Kit (type III receptor protein-tyrosine kinase), MC1R, MFSD12, Microphthalmia-associated transcription factor, Pax3, PMEL17, SLC24A, SLC36A1, SLC45A2=MATP, syntaxin-17, TRPM1) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~9796~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~T-box transcription factor (TBX3)~/and+Taxon ID=~9796~/or+Gene Gephebase=~T-box transcription factor (TBX3)~/and+Taxon ID=~9796~#gephebase-summary-title)	Related Haplotypes

EXTERNAL LINKS

COMMENTS

@AllelicSeries Dun is dominant over non-dun ; <https://omia.org/OMIA001972/9796/>