

GEPHE SUMMARY

TBXT (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+TBXT+Gephebase-summary-title)	Gephebase Gene	GP00002293	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		
Organ size (tail; short) (https://www.gephebase.org/search-criteria?/and+Trait+Organ+size+(tail;+short)+Gephebase-summary-title)	Trait		
WT	Trait State in Taxon A		
Bob tail in Manx cats ; dominant ; recessive embryonic lethal	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
Felis catus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Felis+catus+Gephebase-summary-title)	Latin Name	Felis catus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Felis+catus+Gephebase-summary-title)	Latin Name
domestic cat	Common Name	domestic cat	Common Name
Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.	Synonyms	Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.	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; Carnivora; Feliformia; Felidae; Felinae; Felis	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Feliformia; Felidae; Felinae; Felis	Lineage
Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682)	Parent	Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682)	Parent
9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685)	NCBI Taxonomy ID	9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

Tbxt	Generic Gene Name	P20293 (http://www.uniprot.org/uniprot/P20293)	UniProtKB Mus musculus
Lr; T1; Bra; Low; Tl2; Tl3; cou; Tbxt; me75; D17Mit170; T	Synonyms	()	GenebankID or UniProtKB
10090.ENSMUSP00000074236 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000074236)	String		
-	Sequence Similarities		
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001228)	GO - Molecular Function		
GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000977)			
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:0001102 : RNA polymerase II activating transcription factor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001102>)

GO - Biological Process

GO:0009653 : anatomical structure morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009653>)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
GO:0006357 : regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006357>)
GO:0009952 : anterior/posterior pattern specification
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009952>)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)
GO:0001843 : neural tube closure (<https://www.ebi.ac.uk/QuickGO/term/GO:0001843>)
GO:0008284 : positive regulation of cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008284>)
GO:0001756 : somitogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0001756>)
GO:0060395 : SMAD protein signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060395>)
GO:0061371 : determination of heart left/right asymmetry
(<https://www.ebi.ac.uk/QuickGO/term/GO:0061371>)
GO:0001707 : mesoderm formation (<https://www.ebi.ac.uk/QuickGO/term/GO:0001707>)
GO:0030903 : notochord development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030903>)
GO:0001708 : cell fate specification (<https://www.ebi.ac.uk/QuickGO/term/GO:0001708>)
GO:0007498 : mesoderm development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007498>)
GO:0043433 : negative regulation of DNA-binding transcription factor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043433>)
GO:0023019 : signal transduction involved in regulation of gene expression
(<https://www.ebi.ac.uk/QuickGO/term/GO:0023019>)
GO:0036342 : post-anal tail morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0036342>)
GO:0060349 : bone morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0060349>)
GO:0071300 : cellular response to retinoic acid
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071300>)
GO:0001570 : vasculogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0001570>)
GO:0022414 : reproductive process (<https://www.ebi.ac.uk/QuickGO/term/GO:0022414>)
GO:0048706 : embryonic skeletal system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048706>)
GO:0055007 : cardiac muscle cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0055007>)
GO:0003007 : heart morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0003007>)
GO:0007509 : mesoderm migration involved in gastrulation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007509>)
GO:0001839 : neural plate morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001839>)
GO:0014028 : notochord formation (<https://www.ebi.ac.uk/QuickGO/term/GO:0014028>)
GO:0007341 : penetration of zona pellucida
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007341>)
GO:0003257 : positive regulation of transcription from RNA polymerase II promoter
involved in myocardial precursor cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003257>)

GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0000790 : nuclear chromatin (<https://www.ebi.ac.uk/QuickGO/term/GO:0000790>)
GO:0000785 : chromatin (<https://www.ebi.ac.uk/QuickGO/term/GO:0000785>)

Presumptive Null

Yes (<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>)

Aberration Type

Indel (<https://www.gephebase.org/search-criteria?/and+Aberration+Type=~Indel^#gephebase-summary-title>)

Indel Size

10-99 bp

Molecular Details of the Mutation

c.998_1014dup17delGCC

Experimental Evidence

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Candidate+Gene^#gephebase-summary-title>)

Main Reference

Buckingham KJ; McMillin MJ; Brassil MM; Shively KM; Magnaye KM; Cortes A; Weinmann AS; Lyons LA; Barnshad MJ

Most mammals possess a tail, humans and the Great Apes being notable exceptions. One approach to understanding the mechanisms and evolutionary forces influencing development of a tail is to identify the genetic factors that influence extreme tail length variation within a species. In mice, the Tailless locus has proven to be complex, with evidence of multiple different genes and mutations with pleiotropic effects on tail length, fertility, embryogenesis, male transmission ratio, and meiotic recombination. Five cat breeds have abnormal tail length phenotypes: the American Bobtail, the Manx, the Pixie-Bob, the Kurilian Bobtail, and the Japanese Bobtail. We sequenced the T gene in several independent lineages of Manx cats from both the US and the Isle of Man and identified three 1-bp deletions and one duplication/deletion, each predicted to cause a frameshift that leads to premature termination and truncation of the carboxy terminal end of the Brachyury protein. Ninety-five percent of Manx cats with short-tail phenotypes were heterozygous for T mutations, mutant alleles appeared to be largely lineage-specific, and a maximum LOD score of 6.21 with T was obtained at a recombination fraction (\hat{r}) of 0.00. One mutant T allele was shared with American Bobtails and Pixie-Bobs; both breeds developed more recently in the US. The ability of mutant Brachyury protein to activate transcription of a downstream target was substantially lower than wild-type protein. Collectively, these results suggest that haploinsufficiency of Brachyury is one mechanism underlying variable tail length in domesticated cats.

RELATED GEPHE

1 (HES7) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9685~/and+Trait=Organ+size/and+groupHaplotypes=true#gephebase-summary-title>)

3 (<https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=~TBXT~/and+Taxon+ID=~9685~/or+Gene+Gephebase=~TBXT~/and+Taxon+ID=~9685~/#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage @AllelicSeries @Parallelism <https://omia.org/OMIA000975/9685/>