GEPHE SUMMARY

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

GepheID
GP00002291
Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)
Trait
Organ size (tail; short) (https://www.gephebase.org/search-criteria?/and+Trait=^Organ size (tail; short)^#gephebase-summary-title)
Trait State in Taxon A
WT
Trait State in Taxon B
Bob tail in Manx cats ; dominant ; recessive embryonic lethal
Ancestral State
Taxon A
Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Felis catus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Felis catus^#gephebase-summary-title)
Common Name
domestic cat
Synonyms
Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.
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; Carnivora; Feliformia; Felidae; Felinae; Felis
Parent
Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682)
NCBI Taxonomy ID
9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685)
is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Felis catus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Felis catus^#gephebase-summary-title)
Common Name
domestic cat
Synonyms
Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.
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; Carnivora; Feliformia; Felidae; Felinae; Felis
Parent
Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682)
NCBI Taxonomy ID
9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685)
is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Tbxt
Synonyms
Lr; T1; Bra; Low; Tl2; Tl3; cou; Tbxt; me75; D17Mit170; T
String
10090.ENSMUSP00000074236 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000074236)
Sequence Similarities
-
GO - Molecular Function
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001228)
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)

UniProtKB Mus musculus
P20293 (http://www.uniprot.org/uniprot/P20293)
GenebankID or UniProtKB
()

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

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

c.1199delC

Experimental Evidence

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

Main Reference

Multiple mutant T alleles cause haploinsufficiency of Brachyury and short tails in Manx cats. (2013) (https://pubmed.ncbi.nlm.nih.gov/23949773)

Authors

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

Abstract

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 (Î˜) 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.

Additional References

RELATED GEPHE

Related Genes

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

Related Haplotypes

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/