

## GEPHE SUMMARY

|  |   |                                 |                                    |
|--|---|---------------------------------|------------------------------------|
| <p>TBXT (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+TBXT+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+TBXT+Gephebase-summary-title</a>)</p> <p>Published</p> | <p>Gephebase Gene</p> <p>Entry Status</p> | <p>GP00002340</p> <p>Martin</p> | <p>GepheID</p> <p>Main curator</p> |
|--|---|---------------------------------|------------------------------------|

## PHENOTYPIC CHANGE

|  |   |   |  |   |  |
|--|---|---|--|---|--|
| <p>Morphology (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Category+Morphology+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category+Morphology+Gephebase-summary-title</a>)</p> <p>Organ size (tail; short) (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Organ+size+(tail;+short)+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Organ+size+(tail;+short)+Gephebase-summary-title</a>)</p> <p>WT</p> <p>Bob Tail in Pembroke Welsh Corgi</p> <p>Taxon A</p> <p>Domesticated (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+Gephebase-summary-title</a>)</p> | <p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p> | <p>Taxon A</p> <p>Canis lupus familiaris<br/>(<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Canis+lupus+familiaris+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Canis+lupus+familiaris+Gephebase-summary-title</a>)</p> <p>dog</p> <p>Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758</p> <p>subspecies</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Canis; Canis lupus</p> <p>Canis lupus (gray wolf) - (Rank: species)<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612</a>)</p> <p>9615<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615</a>)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p> | <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p> | <p>Taxon B</p> <p>Canis lupus familiaris<br/>(<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Canis+lupus+familiaris+Gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Canis+lupus+familiaris+Gephebase-summary-title</a>)</p> <p>dog</p> <p>Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758</p> <p>subspecies</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Canis; Canis lupus</p> <p>Canis lupus (gray wolf) - (Rank: species)<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612</a>)</p> <p>9615<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615</a>)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p> | <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p> |
|--|---|---|--|---|--|

## GENOTYPIC CHANGE

|  |  |   |  |
|--|--|---|--|
| <p>Tbxt</p> <p>Lr; T1; Bra; Low; Tl2; Tl3; cou; Tbxt; me75; D17Mit170; T</p> <p>10090.ENSMUSP00000074236<br/>(<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000074236">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000074236</a>)</p> <p>-</p>  | <p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> | <p>P20293 (<a href="http://www.uniprot.org/uniprot/P20293">http://www.uniprot.org/uniprot/P20293</a>)</p> <p>()</p> | <p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p> |
| <p>GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific<br/>(<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0001228">https://www.ebi.ac.uk/QuickGO/term/GO:0001228</a>)</p> <p>GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding<br/>(<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0000977">https://www.ebi.ac.uk/QuickGO/term/GO:0000977</a>)</p> <p>GO:0043565 : sequence-specific DNA binding<br/>(<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043565">https://www.ebi.ac.uk/QuickGO/term/GO:0043565</a>)</p> |  |   |  |

GO:000981 : DNA-binding transcription factor activity, RNA polymerase II-specific  
(<https://www.ebi.ac.uk/QuickGO/term/GO:000981>)  
GO:000978 : RNA polymerase II proximal promoter sequence-specific DNA binding  
(<https://www.ebi.ac.uk/QuickGO/term/GO:000978>)  
GO:001085 : RNA polymerase II transcription factor binding  
(<https://www.ebi.ac.uk/QuickGO/term/GO:001085>)  
GO:001102 : RNA polymerase II activating transcription factor binding  
(<https://www.ebi.ac.uk/QuickGO/term/GO:001102>)

#### 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

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

c.189C>G p.L63M

Experimental Evidence

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

|            | Taxon A | Taxon B | Position |
|------------|---------|---------|----------|
| Codon      | -       | -       | -        |
| Amino-acid | Ile     | Met     | 63       |

#### Main Reference

Canine homolog of the T-box transcription factor T; failure of the protein to bind to its DNA target leads to a short-tail phenotype. (2001) (<https://pubmed.ncbi.nlm.nih.gov/11252170>)

#### Authors

Haworth K; Putt W; Cattanach B; Breen M; Binns M; Lingaas F; Edwards YH

#### Abstract

Domestic dog breeds show a wide variety of morphologies and offer excellent opportunities to study the molecular genetics of phenotypic traits. We are interested in exploring this potential and have begun by investigating the genetic basis of a short-tail trait. Our focus has been on the T gene, which encodes a T-box transcription factor important for normal posterior mesoderm development. Haploinsufficiency of T protein underlies a short-tail phenotype in mice that is inherited in an autosomal dominant fashion. We have cloned the dog homolog of T and mapped the locus to canine Chromosome (Chr) 1q23. Full sequence analysis of the T gene from a number of different dog breeds identified several polymorphisms and a unique missense mutation in a bob-tailed dog and its bob-tailed descendants. This mutation is situated in a highly conserved region of the T-box domain and alters the ability of the T protein to bind to its consensus DNA target. Analysis of offspring from several independent bobtail x bobtail crosses indicates that the homozygous phenotype is embryonic lethal.

#### Additional References

Single-nucleotide-polymorphism-based association mapping of dog stereotypes. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18505865>)

A study of inherited short tail and taillessness in Pembroke Welsh corgi. (2008) (<https://pubmed.ncbi.nlm.nih.gov/17850278>)

Ancestral T-box mutation is present in many, but not all, short-tailed dog breeds. (2009 Mar-Apr) (<https://pubmed.ncbi.nlm.nih.gov/18854372>)

## RELATED GEPHE

#### Related Genes

1 (Dvl2) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=%5E9615%5E/and+Trait=Organ size/and+groupHaplotypes=true#gephebase-summary-title>)

#### Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

@Parallelism @HeterozygoteAdvantage <https://www.omia.org/OMIA000975/9615/>