

## GEPHE SUMMARY

**Gephebase Gene**  
TBXT

**Entry Status**  
Published

**GepheID**  
GP00002289

**Main curator**  
Martin

## PHENOTYPIC CHANGE

**Trait Category**  
Morphology

**Trait**  
Organ size (tail; short)

**Trait State in Taxon A**  
WT

**Trait State in Taxon B**  
Short Tail

**Ancestral State**  
Taxon A

**Taxonomic Status**  
Domesticated

### Taxon A

**Latin Name**  
*Ovis aries*

**Common Name**  
sheep

**Synonyms**  
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep;  
Ovis aries 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; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

**Parent**  
Ovis () - (Rank: genus)

**NCBI Taxonomy ID**  
9940

**is Taxon A an Intraspecies?**  
Yes

**Taxon A Description**  
Barag sheep

### Taxon B

**Latin Name**  
*Ovis aries*

**Common Name**  
sheep

**Synonyms**  
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep;  
Ovis aries 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; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

**Parent**  
Ovis () - (Rank: genus)

**NCBI Taxonomy ID**  
9940

**is Taxon B an Intraspecies?**  
Yes

**Taxon B Description**  
Hulunbair sheep; Kazalh and other breeds

## GENOTYPIC CHANGE

**Generic Gene Name**  
Tbxt

**Synonyms**  
Lr; T1; Bra; Low; Tl2; Tl3; cou; Tbxt; me75; D17Mit170; T

**String**  
10090.ENSMUSP00000074236

**Sequence Similarities**  
-

**GO - Molecular Function**  
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific  
GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding  
GO:0043565 : sequence-specific DNA binding  
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific  
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding  
GO:0001085 : RNA polymerase II transcription factor binding  
GO:0001102 : RNA polymerase II activating transcription factor binding

**UniProtKB Mus musculus**  
P20293

**GenebankID or UniProtKB**

#### GO - Biological Process

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

#### GO - Cellular Component

GO:0005737 : cytoplasm  
GO:0005654 : nucleoplasm  
GO:0005634 : nucleus  
GO:0000790 : nuclear chromatin  
GO:0000785 : chromatin

#### Presumptive Null

No

#### Molecular Type

Coding

#### Aberration Type

SNP

#### SNP Coding Change

Nonsynonymous

#### Molecular Details of the Mutation

c.G334T p.G112W

#### Experimental Evidence

Linkage Mapping

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gly	Trp	112

#### Main Reference

Whole Genome Sequencing of Hulunbuir Short-Tailed Sheep for Identifying Candidate Genes Related to the Short-Tail Phenotype. (2018)

#### Authors

Zhi D; Da L; Liu M; Cheng C; Zhang Y; Wang X; Li X; Tian Z; Yang Y; He T; Long X; Wei W; Cao G

#### Abstract

The Hulunbuir short-tailed sheep (*Ovis aries*) is a breed native to China, in which the short-tail phenotype is the result of artificial and natural selection favoring a specific set of genetic mutations. Here, we analyzed the genetic differences between short-tail and normal-tail phenotypes at the genomic level. Selection signals were identified in genome-wide sequences. From 16 sheep, we identified 72,101,346 single nucleotide polymorphisms. Selection signals were detected based on the fixation index and heterozygosity. Seven genomic regions under putative selection were identified, and these regions contained nine genes. Among these genes, T was the strongest candidate as T is related to vertebral development. In T, a nonsynonymous mutation at c.G334T resulted in p.G112W substitution. We inferred that the c.G334T mutation in T leads to functional changes in Brachyury-encoded by this gene-resulting in the short-tail phenotype. Our findings provide a valuable insight into the development of the short-tail phenotype in sheep and other short-tailed animals.

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#### Additional References

Two linked TBXT (brachyury) gene polymorphisms are associated with the tailless phenotype in fat-rumped sheep. (2019)

## RELATED GEPHE

### Related Genes

No matches found.

### Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

@Parallelism @HeterozygoteAdvantage