GEPHE SUMMARY

Gephebase Gene
EOMES (eomesodermin)

GepheID
GP00000271

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Bird head comb (reduced)

Trait State in Taxon A
Gallus gallus

Trait State in Taxon B
Gallus gallus - Duplex Comb

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Gallus gallus

Common Name
chicken

Synonyms
Gallus gallus domesticus; chicken; bantam; chickens

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus

Parent
Gallus () - (Rank: genus)

NCBI Taxonomy ID
9031

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Gallus gallus

Common Name
chicken

Synonyms
Gallus gallus domesticus; chicken; bantam; chickens

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus

Parent
Gallus () - (Rank: genus)

NCBI Taxonomy ID
9031

is Taxon B an Infraspecies?
Yes

Taxon B Description
Gallus gallus - Duplex Comb

GENOTYPIC CHANGE

Generic Gene Name
Eomes

Synonyms
Tbr2; TBR-2; C77258

String
10090.ENSMUSP00000035020

Sequence Similarities
-

GO - Molecular Function
GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding
GO:0043565 : sequence-specific DNA binding
GO:0003677 : DNA binding
GO:0003682 : chromatin binding
GO:0001227 : DNA-binding transcription repressor activity, RNA polymerase II-specific
GO:0001102 : RNA polymerase II activating transcription factor binding

GO - Biological Process

UniProtKB Mus musculus
O54839

GenebankID or UniProtKB
NP_001308484

GO:0045944 : positive regulation of transcription by RNA polymerase II
GO:0050767 : regulation of neurogenesis
GO:0006357 : regulation of transcription by RNA polymerase II
GO:0030154 : cell differentiation
GO:0000122 : negative regulation of transcription by RNA polymerase II
GO:0045893 : positive regulation of transcription, DNA-templated
GO:0045597 : positive regulation of cell differentiation
GO:0001707 : mesoderm formation
GO:0010468 : regulation of gene expression
GO:0043433 : negative regulation of DNA-binding transcription factor activity
GO:0002250 : adaptive immune response
GO:0001824 : blastocyst development
GO:0007420 : brain development
GO:0010002 : cardioblast differentiation
GO:0002302 : CD8-positive, alpha-beta T cell differentiation involved in immune response
GO:0060706 : cell differentiation involved in embryonic placenta development
GO:0021895 : cerebral cortex neuron differentiation
GO:0021796 : cerebral cortex regionalization
GO:0001706 : endoderm formation
GO:0001714 : endodermal cell fate specification
GO:0032609 : interferon-gamma production
GO:0060809 : mesodermal to mesenchymal transition involved in gastrulation
GO:0021772 : olfactory bulb development
GO:0045664 : regulation of neuron differentiation
GO:0035914 : skeletal muscle cell differentiation
GO:0019827 : stem cell population maintenance
GO:0001829 : trophectodermal cell differentiation

GO - Cellular Component
GO:0005634 : nucleus
GO:0000790 : nuclear chromatin

Presumptive Null
No

Molecular Type
Gene Amplification

Aberration Type
Insertion

Insertion Size
10-100 kb

Molecular Details of the Mutation
a 20 Kb tandem duplication containing several conserved putative regulatory elements located 200 Kb upstream of the eomesodermin gene

Experimental Evidence
Linkage Mapping

Main Reference
A genomic duplication is associated with ectopic eomesodermin expression in the embryonic chicken comb and two duplex-comb phenotypes. (2015)

Authors
Dorshorst B; Harun-Or-Rashid M; Bagherpoor AJ; Rubin CJ; Ashwell C; Gourichon D; Tixier-Boichard M; HallbÃ¶Ã¶k F; Andersson L

Abstract
Duplex-comb (D) is one of three major loci affecting comb morphology in the domestic chicken. Here we show that the two Duplex-comb alleles, V-shaped (D*V) and Buttercup (D*C), are both associated with a 20 Kb tandem duplication containing several conserved putative regulatory elements located 200 Kb upstream of the eomesodermin gene (EOMES). EOMES is a T-box transcription factor that is involved in mesoderm specification during gastrulation. In D*V and D*C chicken embryos we find that EOMES is ectopically expressed in the ectoderm of the comb-developing region as compared to wild-type embryos. The confinement of the ectopic expression of EOMES to the ectoderm is in stark contrast to the causal mechanisms underlying the two other major comb loci in the chicken (Rose-comb and Pea-comb) in which the transcription factors MNR2 and SOX5 are ectopically expressed strictly in the mesenchyme. Interestingly, the causal mutations of all three major comb loci in the chicken are now known to be composed of large-scale structural genomic variants that each result in ectopic expression of transcription factors. The Duplex-comb locus also illustrates the evolution of alleles in domestic animals, which means that alleles evolve by the accumulation of two or more consecutive mutations affecting the phenotype. We do not yet know whether the V-shaped or Buttercup allele correspond to the second mutation that occurred on the haplotype of the original duplication event.

Additional References

RELATED GEPHE

Related Genes
2 (MNR2, SOX5)
Related Haplotypes
No matches found.

EXTERNAL LINKS

COMMENTS

Multiple alleles (not identified) OMIA 000296-9031