

GEPHE SUMMARY

Gephebase Gene
Microphthalmia-associated transcription factor

Entry Status
Published

GepheID
GP00002127

Main curator
Santos

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (feathers)

Trait State in Taxon A
Mallard pigmentation pattern

Trait State in Taxon B
Pekin pigmentation (white)

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Anas platyrhynchos

Common Name
mallard

Synonyms
Anas boschas; Anas domesticus; Anas platyrhynchos f. domestica; mallard; duck; mallard duck; mallard ducks; Anas platyrhynchos Linnaeus 1758; Anas platyrhynchos

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; Anseriformes; Anatidae; Anatinae; Anas

Parent
Anas (ducks) - (Rank: genus)

NCBI Taxonomy ID
8839

is Taxon A an Intraspecies?
Yes

Taxon A Description
Wild mallard ducks

Taxon B

Latin Name
Anas platyrhynchos

Common Name
mallard

Synonyms
Anas boschas; Anas domesticus; Anas platyrhynchos f. domestica; mallard; duck; mallard duck; mallard ducks; Anas platyrhynchos Linnaeus 1758; Anas platyrhynchos

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; Anseriformes; Anatidae; Anatinae; Anas

Parent
Anas (ducks) - (Rank: genus)

NCBI Taxonomy ID
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is Taxon B an Intraspecies?
Yes

Taxon B Description
Domesticated breed

GENOTYPIC CHANGE

Generic Gene Name
Mitf

Synonyms
Wh; bw; mi; vit; BCC2; Bhlhe32; Gsfbcc2; Vitiligo; Bw; Mi; Vit

String
10090.ENSMUSP00000044938

Sequence Similarities
Belongs to the MiT/TFE family.

GO - Molecular Function
GO:0046983 : protein dimerization activity
GO:0003700 : DNA-binding transcription factor activity
GO:0043565 : sequence-specific DNA binding
GO:0003677 : DNA binding
GO:0003682 : chromatin binding
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific

UniProtKB Mus musculus
Q08874

GenebankID or UniProtKB

GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding
GO:0070888 : E-box binding
GO:0003705 : transcription factor activity, RNA polymerase II distal enhancer sequence-specific binding

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
GO:0045944 : positive regulation of transcription by RNA polymerase II
GO:0006357 : regulation of transcription by RNA polymerase II
GO:0006355 : regulation of transcription, DNA-templated
GO:0030154 : cell differentiation
GO:0043473 : pigmentation
GO:0000122 : negative regulation of transcription by RNA polymerase II
GO:0045893 : positive regulation of transcription, DNA-templated
GO:0010628 : positive regulation of gene expression
GO:0006351 : transcription, DNA-templated
GO:0045165 : cell fate commitment
GO:0010468 : regulation of gene expression
GO:0030318 : melanocyte differentiation
GO:0043010 : camera-type eye development
GO:0030316 : osteoclast differentiation
GO:0042127 : regulation of cell proliferation
GO:0046849 : bone remodeling
GO:0044336 : canonical Wnt signaling pathway involved in negative regulation of apoptotic process
GO:0030336 : negative regulation of cell migration
GO:2000144 : positive regulation of DNA-templated transcription, initiation
GO:0065003 : protein-containing complex assembly
GO:0045670 : regulation of osteoclast differentiation
GO:2001141 : regulation of RNA biosynthetic process
GO:0016055 : Wnt signaling pathway

GO - Cellular Component

GO:0005634 : nucleus
GO:0032991 : protein-containing complex

Presumptive Null

No

Molecular Type

Cis-regulatory

Aberration Type

Insertion

Insertion Size

1-10 kb

Molecular Details of the Mutation

6.6kb insertion between exon 1M and exon 2, leads to lower expression of isoform MITF M in pekin white ducks

Experimental Evidence

Linkage Mapping

Main Reference

An intercross population study reveals genes associated with body size and plumage color in ducks. (2018)

Authors

Zhou Z; Li M; Cheng H; Fan W; Yuan Z; Gao Q; Xu Y; Guo Z; Zhang Y; Hu J; Liu H; Liu D; Chen W; Zheng Z; Jiang Y; Wen Z; Liu Y; Chen H; Xie M; Zhang Q; Huang W; Wang W; Hou S; Jiang Y

Abstract

Comparative population genomics offers an opportunity to discover the signatures of artificial selection during animal domestication, however, their function cannot be directly revealed. We discover the selection signatures using genome-wide comparisons among 40 mallards, 36 indigenous-breed ducks, and 30 Pekin ducks. Then, the phenotypes are fine-mapped based on resequencing of 1026 ducks from an F segregating population generated by wild × domestic crosses. Interestingly, the two key economic traits of Pekin duck are associated with two selective sweeps with fixed mutations. A novel intronic insertion most possibly leads to a splicing change in MITF accounted for white duck down feathers. And a putative long-distance regulatory mutation causes continuous expression of the IGF2BP1 gene after birth which increases body size by 15% and feed efficiency by 6%. This study provides new insights into genotype-phenotype associations in animal research and constitutes a promising resource on economically important genes in fowl.

Additional References

RELATED GEPHE

Related Genes

1 (MC1R)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@parallelism