GEPHE SUMMARY

Gephebase Gene

Microphtalmia-associated transcription factor (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Microphtalmia-associated transcription factor^#gephebase-summary-title)

GepheID

GP00002127

Entry Status

Published

Main curator

Santos

PHENOTYPIC CHANGE

Trait Category

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait

Coloration (feathers) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (feathers)^#gephebase-summary-title)

Trait State in Taxon A

Mallard pigmentation pattern

Trait State in Taxon B

Pekin pigmentation (white)

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Anas platyrhynchos (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Anas platyrhynchos^#gephebase-summary-title)

Common Name

mallard

Synonyms

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

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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)

NCBI Taxonomy ID

8839 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8839)

is Taxon A an Infraspecies?

Yes

Taxon A Description

Wild mallard ducks

Taxon B

Latin Name

Anas platyrhynchos (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Anas platyrhynchos^#gephebase-summary-title)

Common Name

mallard

Synonyms

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

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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)

NCBI Taxonomy ID

8839 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8839)

is Taxon B an Infraspecies?

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 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000044938)

Sequence Similarities

Belongs to the MiT/TFE family.

GO - Molecular Function

GO:0046983 : protein dimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046983)

UniProtKB Mus musculus

Q08874 (http://www.uniprot.org/uniprot/Q08874)

GenebankID or UniProtKB

()

GO:0003700 : DNA-binding transcription factor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0003700)
GO:0043565 : sequence-specific DNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0043565)
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)
GO:0003682 : chromatin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003682)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
(https://www.ebi.ac.uk/QuickGO/term/GO:0000981)
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA
polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001077)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0000978)
GO:0070888 : E-box binding (https://www.ebi.ac.uk/QuickGO/term/GO:0070888)
GO:0003705 : transcription factor activity, RNA polymerase II distal enhancer sequence-
specific binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003705)

GO - Biological Process

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

GO - Cellular Component

GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0032991 : protein-containing complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0032991)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

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

Main Reference

An intercross population study reveals genes associated with body size and plumage color in ducks. (2018) (https://pubmed.ncbi.nlm.nih.gov/30018292)

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 wildâ€‰Ã—â€‰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

2 (Endothelin receptor B2, MC1R) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^8839^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@parallelism