GEPHE SUMMARY

Gephebase Gene
SOX10 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SOX10^#gephebase-summary-title)

GepheID
GP00001067

Entry Status
Published

Main curator
Martin

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
Columba livia - blue/black

Trait State in Taxon B
Columba livia - recessive red e1 allele

Ancestral State
Taxon A

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

Taxon A

Latin Name
Columba livia (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Columba livia^#gephebase-summary-title)

Common Name
rock pigeon

Synonyms
Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789

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; Columbiformes; Columbidae; Columba

Parent
Columba () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8931)

NCBI Taxonomy ID
8932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8932)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Columba livia (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Columba livia^#gephebase-summary-title)

Common Name
rock pigeon

Synonyms
Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789

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; Columbiformes; Columbidae; Columba

Parent
Columba () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8931)

NCBI Taxonomy ID
8932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8932)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Sox10

Synonyms
gt; Dom; Sox21; Sox-10

String
10090.ENSMUSP00000039466 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000039466)

Sequence Similarities
-

GO - Molecular Function
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001228)
GO:0003700 : DNA-binding transcription factor activity

UniProtKB Mus musculus
Q04888 (http://www.uniprot.org/uniprot/Q04888)

GenebankID or UniProtKB
XP_005514744 (https://www.ncbi.nlm.nih.gov/nuccore/XP_005514744)

(https://www.ebi.ac.uk/QuickGO/term/GO:0003700)
GO:0008134 : transcription factor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0008134)
GO:0044212 : transcription regulatory region DNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0044212)
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:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0000978)
GO:0000980 : RNA polymerase II distal enhancer sequence-specific DNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0000980)
GO:0003705 : transcription factor activity, RNA polymerase II distal enhancer sequence-specific binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003705)
GO:1990841 : promoter-specific chromatin binding
(https://www.ebi.ac.uk/QuickGO/term/GO:1990841)

GO - Biological Process

GO:0007417 : central nervous system development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007417)
GO:0043066 : negative regulation of apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0007422 : peripheral nervous system development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007422)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
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:0045892 : negative regulation of transcription, DNA-templated
(https://www.ebi.ac.uk/QuickGO/term/GO:0045892)
GO:0001755 : neural crest cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0001755)
GO:0045893 : positive regulation of transcription, DNA-templated
(https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0090090 : negative regulation of canonical Wnt signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0090090)
GO:0001701 : in utero embryonic development
(https://www.ebi.ac.uk/QuickGO/term/GO:0001701)
GO:0010628 : positive regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010628)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0048546 : digestive tract morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0048546)
GO:0002009 : morphogenesis of an epithelium
(https://www.ebi.ac.uk/QuickGO/term/GO:0002009)
GO:0048484 : enteric nervous system development
(https://www.ebi.ac.uk/QuickGO/term/GO:0048484)
GO:0048469 : cell maturation (https://www.ebi.ac.uk/QuickGO/term/GO:0048469)
GO:0032808 : lacrimal gland development
(https://www.ebi.ac.uk/QuickGO/term/GO:0032808)
GO:0048589 : developmental growth
(https://www.ebi.ac.uk/QuickGO/term/GO:0048589)
GO:0071393 : cellular response to progesterone stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:0071393)
GO:0022010 : central nervous system myelination
(https://www.ebi.ac.uk/QuickGO/term/GO:0022010)
GO:0061138 : morphogenesis of a branching epithelium
(https://www.ebi.ac.uk/QuickGO/term/GO:0061138)
GO:0010626 : negative regulation of Schwann cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0010626)
GO:0014003 : oligodendrocyte development
(https://www.ebi.ac.uk/QuickGO/term/GO:0014003)
GO:0048709 : oligodendrocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0048709)
GO:0014015 : positive regulation of gliogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0014015)
GO:0031643 : positive regulation of myelination
(https://www.ebi.ac.uk/QuickGO/term/GO:0031643)
GO:0002052 : positive regulation of neuroblast proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0002052)
GO:0006368 : transcription elongation from RNA polymerase II promoter
(https://www.ebi.ac.uk/QuickGO/term/GO:0006368)

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:0000785 : chromatin (https://www.ebi.ac.uk/QuickGO/term/GO:0000785)
GO:0031315 : extrinsic component of mitochondrial outer membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0031315)

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

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

Deletion Size

1-10 kb

Molecular Details of the Mutation

melanocyte conserved enhancer deletion

Experimental Evidence

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

Main Reference

Epistatic and combinatorial effects of pigmentary gene mutations in the domestic pigeon. (2014) (https://pubmed.ncbi.nlm.nih.gov/24508169)

Authors

Domyan ET; Guernsey MW; Kronenberg Z; Krishnan S; Boissy RE; Vickrey AI; Rodgers C; Cassidy P; Leachman SA; Fondon JW; Yandell M; Shapiro MD

Abstract

Understanding the molecular basis of phenotypic diversity is a critical challenge in biology, yet we know little about the mechanistic effects of different mutations and epistatic relationships among loci that contribute to complex traits. Pigmentation genetics offers a powerful model for identifying mutations underlying diversity and for determining how additional complexity emerges from interactions among loci. Centuries of artificial selection in domestic rock pigeons (Columba livia) have cultivated tremendous variation in plumage pigmentation through the combined effects of dozens of loci. The dominance and epistatic hierarchies of key loci governing this diversity are known through classical genetic studies, but their molecular identities and the mechanisms of their genetic interactions remain unknown. Here we identify protein-coding and cis-regulatory mutations in Tyrp1, Sox10, and Slc45a2 that underlie classical color phenotypes of pigeons and present a mechanistic explanation of their dominance and epistatic relationships. We also find unanticipated allelic heterogeneity at Tyrp1 and Sox10, indicating that color variants evolved repeatedly though mutations in the same genes. These results demonstrate how a spectrum of coding and regulatory mutations in a small number of genes can interact to generate substantial phenotypic diversity in a classic Darwinian model of evolution.

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Additional References

RELATED GEPHE

Related Genes

6 (MC1R, Mlana, ndp (norrin), slc2a11b, SLC45A2=MATP, tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^8932^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^SOX10^/and+Taxon ID=^8932^/or+Gene Gephebase=^SOX10^/and+Taxon ID=^8932^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@Epistasis - Enhancer deletion - Two alleles @AllelicSeries