

GEPHE SUMMARY

Gephebase Gene
SOX10

Entry Status
Published

GepheID
GP00001067

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (feathers)

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

Taxon A

Latin Name
Columba livia

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)

NCBI Taxonomy ID
8932

is Taxon A an Intraspecies?
No

Taxon B

Latin Name
Columba livia

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)

NCBI Taxonomy ID
8932

is Taxon B an Intraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Sox10

Synonyms
gt; Dom; Sox21; Sox-10

String
10090.ENSMUSP00000039466

Sequence Similarities
-

GO - Molecular Function
GO:0042802 : identical protein binding
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific
GO:0003700 : DNA-binding transcription factor activity
GO:0008134 : transcription factor binding
GO:0044212 : transcription regulatory region DNA binding
GO:0003677 : DNA binding
GO:0003682 : chromatin binding
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding

UniProtKB Mus musculus
Q04888

GenebankID or UniProtKB
XP_005514744

GO:0000980 : RNA polymerase II distal enhancer sequence-specific DNA binding
GO:0003705 : transcription factor activity, RNA polymerase II distal enhancer sequence-specific binding
GO:1990841 : promoter-specific chromatin binding

GO - Biological Process

GO:0007417 : central nervous system development
GO:0043066 : negative regulation of apoptotic process
GO:0007422 : peripheral nervous system development
GO:0045944 : positive regulation of transcription by RNA polymerase II
GO:0006355 : regulation of transcription, DNA-templated
GO:0030154 : cell differentiation
GO:0045892 : negative regulation of transcription, DNA-templated
GO:0001755 : neural crest cell migration
GO:0045893 : positive regulation of transcription, DNA-templated
GO:0090090 : negative regulation of canonical Wnt signaling pathway
GO:0001701 : in utero embryonic development
GO:0010628 : positive regulation of gene expression
GO:0030318 : melanocyte differentiation
GO:0048546 : digestive tract morphogenesis
GO:0002009 : morphogenesis of an epithelium
GO:0048484 : enteric nervous system development
GO:0048469 : cell maturation
GO:0032808 : lacrimal gland development
GO:0048589 : developmental growth
GO:0071393 : cellular response to progesterone stimulus
GO:0022010 : central nervous system myelination
GO:0061138 : morphogenesis of a branching epithelium
GO:0010626 : negative regulation of Schwann cell proliferation
GO:0014003 : oligodendrocyte development
GO:0048709 : oligodendrocyte differentiation
GO:0014015 : positive regulation of gliogenesis
GO:0031643 : positive regulation of myelination
GO:0002052 : positive regulation of neuroblast proliferation
GO:0006368 : transcription elongation from RNA polymerase II promoter

GO - Cellular Component

GO:0005737 : cytoplasm
GO:0005654 : nucleoplasm
GO:0005634 : nucleus
GO:0000785 : chromatin
GO:0031315 : extrinsic component of mitochondrial outer membrane

Presumptive Null

No

Molecular Type

Cis-regulatory

Aberration Type

Deletion

Deletion Size

1-10 kb

Molecular Details of the Mutation

melanocyte conserved enhancer deletion

Experimental Evidence

Linkage Mapping

Main Reference

Epistatic and combinatorial effects of pigmentary gene mutations in the domestic pigeon. (2014)

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 through 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

3 (MC1R, SLC45A2=MATP, tyrosinase-related protein 1 (TYRP1))

Related Haplotypes

1

EXTERNAL LINKS

COMMENTS

@Epistasis - Enhancer deletion - Two alleles