

GEPHE SUMMARY

	Gephebase Gene	GephelD
tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria/?and+Gene+Gephebase=%tyrosinase-related+protein+1+(TYRP1)%#gephebase-summary-title)	GP00001149	Main curator
	Entry Status	
Published	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 - brown b1 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	Taxon B	Latin Name
Columba livia (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Columba+livia%#gephebase-summary-title)		Columba livia (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Columba+livia%#gephebase-summary-title)	
rock pigeon	Common Name	rock pigeon	Common Name
Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789	Synonyms	Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789	Synonyms
species	Rank	species	Rank
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	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	Lineage
Columba () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8931)	Parent	Columba () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8931)	Parent
8932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8932)	NCBI Taxonomy ID	8932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8932)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

Tyrp1	Generic Gene Name	UniProtKB Mus musculus
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000006151 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)	String	NP_001302454 (https://www.ncbi.nlm.nih.gov/nuccore/NP_001302454)
Belongs to the tyrosinase family.	Sequence Similarities	
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)	GO - Molecular Function	
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)		

GO:0046872 : metal ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0046872>)

GO:0004497 : monooxygenase activity

(<https://www.ebi.ac.uk/QuickGO/term/GO:0004497>)

GO - Biological Process

GO:0032438 : melanosome organization

(<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)

GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)

GO:0048023 : positive regulation of melanin biosynthetic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)

GO:0006583 : melanin biosynthetic process from tyrosine

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006583>)

GO:0030318 : melanocyte differentiation

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)

GO:0043438 : acetoacetic acid metabolic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0043438>)

GO:0006582 : melanin metabolic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006582>)

GO - Cellular Component

GO:0016021 : integral component of membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

GO:0030669 : clathrin-coated endocytic vesicle membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030669>)

GO:0010008 : endosome membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0010008>)

GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)

GO:0033162 : melanosome membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0033162>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Ala23Pro

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ala	Pro	23

Main Reference

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

Authors

Domany 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

6 (MC1R, Mlana, ndp (norrin), slc2a11b, SLC45A2=MATP, SOX10) ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+TaxonID=^8932#/and+Trait=Coloration/and+groupHaplotypes=true))

Related Haplotypes

2 ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=^tyrosinase-related+protein+1+(TYRP1)^/and+Taxon+ID=^8932#/or+Gene+Gephebase=^tyrosinase-related+protein+1+(TYRP1)^/and+Taxon+ID=^8932))

EXTERNAL LINKS

COMMENTS

@Epistasis Multiple alleles @AllelicSeries