

GEPHE SUMMARY

SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="SLC45A2=MATP"#gephebase-summary-title)	Gephebase Gene	GP00001062	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category="Morphology"#gephebase-summary-title)	Trait Category		
Coloration (feathers) (https://www.gephebase.org/search-criteria?/and+Trait ="Coloration (feathers)"#gephebase-summary-title)	Trait		
Columba livia - blue/black	Trait State in Taxon A		
Columba livia - recessive dilute (Dun)	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status="Domesticated"#gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Columba livia (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Columba livia"#gephebase-summary-title)	Latin Name	Columba livia (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Columba livia"#gephebase-summary-title)	Latin Name
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 Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

SLC45A2	Generic Gene Name	Q9UMX9 (http://www.uniprot.org/uniprot/Q9UMX9)	UniProtKB Homo sapiens
1A1; AIM1; MATP; OCA4; SHEP5	Synonyms	XP_005506068 (https://www.ncbi.nlm.nih.gov/nuccore/XP_005506068)	GenebankID or UniProtKB
9606.ENSP00000296589 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000296589)	String		
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.	Sequence Similarities		
GO:0008506 : sucrose:proton symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008506)	GO - Molecular Function		
GO:0042438 : melanin biosynthetic process	GO - Biological Process		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
 GO:0048066 : developmental pigmentation
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0048066>)
 GO:0007601 : visual perception (<https://www.ebi.ac.uk/QuickGO/term/GO:0007601>)
 GO:0050896 : response to stimulus (<https://www.ebi.ac.uk/QuickGO/term/GO:0050896>)
 GO:0015770 : sucrose transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015770>)
 GO - Cellular Component

GO:0016021 : integral component of membrane
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
 GO:0033162 : melanosome membrane
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0033162>)

No ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=)) Presumptive Null
 Coding ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=)) Molecular Type
 SNP ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=)) Aberration Type
 Nonsynonymous SNP Coding Change
 His341Arg Molecular Details of the Mutation
 Linkage Mapping ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=)) Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

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

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

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 Abstract

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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RELATED GEPHE

5 (MC1R, Mlana, ndp (norrin), SOX10, tyrosinase-related protein 1 (TYRP1)) ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=)) Related Genes

No matches found. Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Epistasis

