

GEPHE SUMMARY

	Gephebase Gene	GephelD
SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+Gene Gephebase= [~] SLC45A2=MATP ^{#gephebase-summary-title})	GP00001062	Main curator
	Entry Status	Martin
Published		

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 dilute (Dun)	Ancestral State	
Taxon A	Taxonomic Status	
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status= [~] Domesticated ^{#gephebase-summary-title})		
Taxon A		Taxon B
	Latin Name	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	Common Name
Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789	Synonyms	Synonyms
	Rank	Rank
species	Lineage	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	Parent
Columba () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8931)		
	NCBI Taxonomy ID	NCBI Taxonomy ID
8932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8932)		
	is Taxon A an Infraspecies?	is Taxon B an Infraspecies?
No	No	

GENOTYPIC CHANGE

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

Presumptive Null

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=%27No%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive%20Null=%27No%27#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=%27Coding%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular%20Type=%27Coding%27#gephebase-summary-title))

Aberration Type

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type=%27SNP%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration%20Type=%27SNP%27#gephebase-summary-title))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

His341Arg

Experimental Evidence

Linkage Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=%27Linkage Mapping%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental%20Evidence=%27Linkage%20Mapping%27#gephebase-summary-title))

	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

Domany 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 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

6 (MC1R, Mlana, ndp (norrin), slc2a11b, SOX10, tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+TaxonID=%278932%27/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Genes

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Epistasis

