

GEPHE SUMMARY

|                                                                                                                                                                                                                                                                                                                        |                |            |              |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|------------|--------------|
|                                                                                                                                                                                                                                                                                                                        | Gephebase Gene |            | GepheID      |
| tyrosinase-related protein 1 (TYRP1) ( <a +tyrosinase-related+protein+1+(tyrp1)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+tyrosinase-related+protein+1+(TYRP1)^#gephebase-summary-title</a> ) |                | GP00001150 |              |
|                                                                                                                                                                                                                                                                                                                        | Entry Status   | Martin     | Main curator |
| Published                                                                                                                                                                                                                                                                                                              |                |            |              |

PHENOTYPIC CHANGE

|                                                                                                                                                                                                                                                                                                                                                                                     |                             |                                                                                                                                                                                                                                                                                                                                                                                     |                             |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------|
|                                                                                                                                                                                                                                                                                                                                                                                     | Trait Category              |                                                                                                                                                                                                                                                                                                                                                                                     |                             |
| Morphology ( <a +morphology^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology^#gephebase-summary-title</a> )                                                                                                                                            |                             | Trait                                                                                                                                                                                                                                                                                                                                                                               |                             |
| Coloration (feathers) ( <a +coloration+(feathers)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+(feathers)^#gephebase-summary-title</a> )                                                                                                                             |                             |                                                                                                                                                                                                                                                                                                                                                                                     |                             |
|                                                                                                                                                                                                                                                                                                                                                                                     | Trait State in Taxon A      |                                                                                                                                                                                                                                                                                                                                                                                     |                             |
| Columba livia - blue/black                                                                                                                                                                                                                                                                                                                                                          |                             | Trait State in Taxon B                                                                                                                                                                                                                                                                                                                                                              |                             |
| Columba livia - brown b2 allele                                                                                                                                                                                                                                                                                                                                                     |                             |                                                                                                                                                                                                                                                                                                                                                                                     |                             |
|                                                                                                                                                                                                                                                                                                                                                                                     | Ancestral State             |                                                                                                                                                                                                                                                                                                                                                                                     |                             |
| Taxon A                                                                                                                                                                                                                                                                                                                                                                             |                             | Taxonomic Status                                                                                                                                                                                                                                                                                                                                                                    |                             |
| Domesticated ( <a +domesticated^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated^#gephebase-summary-title</a> )                                                                                                                                  |                             |                                                                                                                                                                                                                                                                                                                                                                                     |                             |
|                                                                                                                                                                                                                                                                                                                                                                                     | Taxon A                     |                                                                                                                                                                                                                                                                                                                                                                                     | Taxon B                     |
|                                                                                                                                                                                                                                                                                                                                                                                     | Latin Name                  |                                                                                                                                                                                                                                                                                                                                                                                     | Latin Name                  |
| Columba livia ( <a +columba+livia^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Columba+livia^#gephebase-summary-title</a> )                                                                                                                           |                             | Columba livia ( <a +columba+livia^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Columba+livia^#gephebase-summary-title</a> )                                                                                                                           |                             |
|                                                                                                                                                                                                                                                                                                                                                                                     | Common Name                 |                                                                                                                                                                                                                                                                                                                                                                                     | Common Name                 |
| rock pigeon                                                                                                                                                                                                                                                                                                                                                                         |                             | rock pigeon                                                                                                                                                                                                                                                                                                                                                                         |                             |
|                                                                                                                                                                                                                                                                                                                                                                                     | Synonyms                    |                                                                                                                                                                                                                                                                                                                                                                                     | Synonyms                    |
| Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789                                                                                                                                                                                                                                                                    |                             | Columba livia domestica; rock pigeon; carrier pigeon; domestic pigeon; rock dove; Columba livia Gmelin, JF, 1789                                                                                                                                                                                                                                                                    |                             |
|                                                                                                                                                                                                                                                                                                                                                                                     | Rank                        |                                                                                                                                                                                                                                                                                                                                                                                     | Rank                        |
| species                                                                                                                                                                                                                                                                                                                                                                             |                             | 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 |                             | 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) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8931">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8931</a> )                                                                                                                                                                                                    |                             | Columba () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8931">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8931</a> )                                                                                                                                                                                                    |                             |
|                                                                                                                                                                                                                                                                                                                                                                                     | NCBI Taxonomy ID            |                                                                                                                                                                                                                                                                                                                                                                                     | NCBI Taxonomy ID            |
| 8932 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8932">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8932</a> )                                                                                                                                                                                                                          |                             | 8932 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8932">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8932</a> )                                                                                                                                                                                                                          |                             |
|                                                                                                                                                                                                                                                                                                                                                                                     | is Taxon A an Intraspecies? |                                                                                                                                                                                                                                                                                                                                                                                     | is Taxon B an Intraspecies? |
| No                                                                                                                                                                                                                                                                                                                                                                                  |                             | No                                                                                                                                                                                                                                                                                                                                                                                  |                             |

GENOTYPIC CHANGE

|                                                                                                                                                                                                                                          |                         |                                                                                                                                    |                         |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------|------------------------------------------------------------------------------------------------------------------------------------|-------------------------|
|                                                                                                                                                                                                                                          | Generic Gene Name       |                                                                                                                                    | UniProtKB Mus musculus  |
| Tyrp1                                                                                                                                                                                                                                    |                         | P07147 ( <a href="http://www.uniprot.org/uniprot/P07147">http://www.uniprot.org/uniprot/P07147</a> )                               |                         |
|                                                                                                                                                                                                                                          | Synonyms                |                                                                                                                                    | GenebankID or UniProtKB |
| b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1                                                                                                                                                                                           |                         | NP_001302454 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/NP_001302454">https://www.ncbi.nlm.nih.gov/nuccore/NP_001302454</a> ) |                         |
|                                                                                                                                                                                                                                          | String                  |                                                                                                                                    |                         |
| 10090.ENSMUSP00000006151 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151</a> ) |                         |                                                                                                                                    |                         |
|                                                                                                                                                                                                                                          | Sequence Similarities   |                                                                                                                                    |                         |
| Belongs to the tyrosinase family.                                                                                                                                                                                                        |                         |                                                                                                                                    |                         |
|                                                                                                                                                                                                                                          | GO - Molecular Function |                                                                                                                                    |                         |
| GO:0042803 : protein homodimerization activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042803">https://www.ebi.ac.uk/QuickGO/term/GO:0042803</a> )                                                                             |                         |                                                                                                                                    |                         |
| GO:0046982 : protein heterodimerization activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046982">https://www.ebi.ac.uk/QuickGO/term/GO:0046982</a> )                                                                           |                         |                                                                                                                                    |                         |

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                  |
| Yes ( <a href="https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title</a> )                                               |                                   |
|                                                                                                                                                                                                                                                                 | Molecular Type                    |
| Coding ( <a href="https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title</a> )                                          |                                   |
|                                                                                                                                                                                                                                                                 | Aberration Type                   |
| SNP ( <a href="https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP^#gephebase-summary-title</a> )                                                 |                                   |
|                                                                                                                                                                                                                                                                 | SNP Coding Change                 |
| Nonsense                                                                                                                                                                                                                                                        |                                   |
|                                                                                                                                                                                                                                                                 | Molecular Details of the Mutation |
| Arg72*                                                                                                                                                                                                                                                          |                                   |
|                                                                                                                                                                                                                                                                 | Experimental Evidence             |
| Linkage Mapping ( <a href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping^#gephebase-summary-title</a> ) |                                   |

|            | Taxon A | Taxon B | Position |
|------------|---------|---------|----------|
| Codon      | -       | -       | -        |
| Amino-acid | Arg     | STP     | 72       |

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               |                |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|
|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | Main Reference |
| Epistatic and combinatorial effects of pigmentary gene mutations in the domestic pigeon. (2014) ( <a href="https://pubmed.ncbi.nlm.nih.gov/24508169">https://pubmed.ncbi.nlm.nih.gov/24508169</a> )                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           |                |
|                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | 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 ( <i>Columba livia</i> ) 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 <i>Tyrp1</i> , <i>Sox10</i> , and <i>Slc45a2</i> 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 <i>Tyrp1</i> and <i>Sox10</i> , 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

|                                                                                                                                                                                                                                                                                                                                                                                                                                                                               |                    |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------|
|                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | Related Genes      |
| 6 (MC1R, Mlana, ndp (norrin), slc2a11b, SLC45A2=MATP, SOX10) ( <a href="https://www.gephebase.org/search-criteria?/or+Taxon ID=~8932^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title">https://www.gephebase.org/search-criteria?/or+Taxon ID=~8932^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title</a> )                                                                                                                        |                    |
|                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | Related Haplotypes |
| 2 ( <a href="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^#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^#gephebase-summary-title</a> ) |                    |

EXTERNAL LINKS

COMMENTS

@Epistasis Multiple alleles @AllelicSeries