

## GEPHE SUMMARY

**Gephebase Gene**  
Mlana

**Entry Status**  
Published

**GepheID**  
GP00002342

**Main curator**  
Martin

## PHENOTYPIC CHANGE

**Trait Category**  
Morphology

**Trait**  
Coloration (feathers ; white-spotting)

**Trait State in Taxon A**  
WT

**Trait State in Taxon B**  
Almond phenotype in Stripper breed and other depigmentation traits in other breeds(allelic series)

**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**  
MLANA

**Synonyms**  
MART1; MART-1

**String**  
9606.ENSP00000370886

**Sequence Similarities**  
-

**GO - Molecular Function**  
-

**GO - Biological Process**  
-

**GO - Cellular Component**  
GO:0005887 : integral component of plasma membrane  
GO:0005794 : Golgi apparatus  
GO:0005789 : endoplasmic reticulum membrane

**UniProtKB Homo sapiens**  
Q16655

**GenebankID or UniProtKB**

GO:0042470 : melanosome  
GO:0005802 : trans-Golgi network

#### Presumptive Null

No

#### Molecular Type

Gene Amplification

#### Aberration Type

Complex Change

#### Molecular Details of the Mutation

CNV with 7 copies of the outer 77-kb segment and 14 copies of the inner 25-kb segment in the genomes of female (ZStW) Almond pigeons

#### Experimental Evidence

Association Mapping

#### Main Reference

A copy number variant is associated with a spectrum of pigmentation patterns in the rock pigeon (*Columba livia*). (2020)

#### Authors

Bruders R; Van Hollebeke H; Osborne EJ; Kronenberg Z; Maclary E; Yandell M; Shapiro MD

#### Abstract

Rock pigeons (*Columba livia*) display an extraordinary array of pigment pattern variation. One such pattern, Almond, is characterized by a variegated patchwork of plumage colors that are distributed in an apparently random manner. Almond is a sex-linked, semi-dominant trait controlled by the classical Stipper (St) locus. Heterozygous males (ZStZ+ sex chromosomes) and hemizygous Almond females (ZStW) are favored by breeders for their attractive plumage. In contrast, homozygous Almond males (ZStZSt) develop severe eye defects and often lack plumage pigmentation, suggesting that higher dosage of the mutant allele is deleterious. To determine the molecular basis of Almond, we compared the genomes of Almond pigeons to non-Almond pigeons and identified a candidate St locus on the Z chromosome. We found a copy number variant (CNV) within the differentiated region that captures complete or partial coding sequences of four genes, including the melanosome maturation gene *Mlana*. We did not find fixed coding changes in genes within the CNV, but all genes are misexpressed in regenerating feather bud collar cells of Almond birds. Notably, six other alleles at the St locus are associated with depigmentation phenotypes, and all exhibit expansion of the same CNV. Structural variation at St is linked to diversity in plumage pigmentation and gene expression, and thus provides a potential mode of rapid phenotypic evolution in pigeons.

#### Additional References

## RELATED GEPHE

#### Related Genes

5 (MC1R, ndp (norrin), SLC45A2=MATP, SOX10, tyrosinase-related protein 1 (TYRP1))

#### Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

@AllelicSeries @CNV @HeterozygoteAdvantage ; homozygous males show severe eye defects