

GEPHE SUMMARY

| | | | |
|---|----------------|------------|--------------|
| PME17 (https://www.gephebase.org/search-criteria/?and+Gene+Gephebase=%PME17%#gephebase-summary-title) | Gephebase Gene | GP00000913 | GephelD |
| | Entry Status | Martin | Main curator |
| 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 | | |
| Gallus gallus - wild pit gamecock | Trait State in Taxon B | | |
| Gallus gallus - Dominant white pit gamecock | 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 |
| Gallus gallus (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Gallus+gallus%#gephebase-summary-title) | | Gallus gallus (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Gallus+gallus%#gephebase-summary-title) | |
| chicken | Common Name | chicken | Common Name |
| Gallus gallus domesticus; chicken; bantam; chickens | Synonyms | Gallus gallus domesticus; chicken; bantam; chickens | Synonyms |
| species | Rank | species | Rank |
| | 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; Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus | | 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; Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus | |
| Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030) | Parent | Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030) | Parent |
| 9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031) | NCBI Taxonomy ID | 9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031) | NCBI Taxonomy ID |
| is Taxon A an Infraspecies? | | is Taxon B an Infraspecies? | |
| Yes | Taxon A Description | Yes | Taxon B Description |
| Gallus gallus - wild pit gamecock | | Gallus gallus - Dominant white pit gamecock | |

GENOTYPIC CHANGE

| | | |
|--|-------------------------|--|
| PME17 | Generic Gene Name | UniProtKB Gallus gallus |
| SILV; MMP15; PMEL17 | Synonyms | GenebankID or UniProtKB |
| 9031.ENSGALP00000023540 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9031.ENSGALP00000023540) | String | AY636128 (https://www.ncbi.nlm.nih.gov/nuccore/AY636128) |
| Belongs to the PMEL/NMB family. | Sequence Similarities | |
| - | GO - Molecular Function | |
| GO:0032438 : melanosome organization (https://www.ebi.ac.uk/QuickGO/term/GO:0032438) | GO - Biological Process | |

GO - Cellular Component

GO:0005887 : integral component of plasma membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)GO:0034493 : melanosome lumen (<https://www.ebi.ac.uk/QuickGO/term/GO:0034493>)

Presumptive Null

No (<https://www.gephebase.org/search-criteria/?and+Presumptive+Null=%No%#gephebase-summary-title>)

Molecular Type

Coding (<https://www.gephebase.org/search-criteria/?and+Molecular+Type=%Coding%#gephebase-summary-title>)

Aberration Type

Insertion (<https://www.gephebase.org/search-criteria/?and+Aberration+Type=%Insertion%#gephebase-summary-title>)

Insertion Size

1-9 bp

Molecular Details of the Mutation

insertion of amino acid 723-725 due to a 9bp-insert in exon 10

Experimental Evidence

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

Main Reference

The Dominant white, Dun and Smoky color variants in chicken are associated with insertion/deletion polymorphisms in the PMEL17 gene. (2004) (<https://pubmed.ncbi.nlm.nih.gov/15579702>)

Authors

Kerje S; Sharma P; Gunnarsson U; Kim H; Bagchi S; Fredriksson R; Schäffer K; Jensen P; von Heijne G; Okimoto R; Andersson L

Abstract

Dominant white, Dun, and Smoky are alleles at the Dominant white locus, which is one of the major loci affecting plumage color in the domestic chicken. Both Dominant white and Dun inhibit the expression of black eumelanin. Smoky arose in a White Leghorn homozygous for Dominant white and partially restores pigmentation. PMEL17 encodes a melanocyte-specific protein and was identified as a positional candidate gene due to its role in the development of eumelanosomes. Linkage analysis of PMEL17 and Dominant white using a red jungle fowl/White Leghorn intercross revealed no recombination between these loci. Sequence analysis showed that the Dominant white allele was exclusively associated with a 9-bp insertion in exon 10, leading to an insertion of three amino acids in the PMEL17 transmembrane region. Similarly, a deletion of five amino acids in the transmembrane region occurs in the protein encoded by Dun. The Smoky allele shared the 9-bp insertion in exon 10 with Dominant white, as expected from its origin, but also had a deletion of 12 nucleotides in exon 6, eliminating four amino acids from the mature protein. These mutations are, together with the recessive silver mutation in the mouse, the only PMEL17 mutations with phenotypic effects that have been described so far in any species.

Additional References

RELATED GEPHE

Related Genes

14 (ABCA1, Agouti (ASIP), CDKN2A, CYP19A1, EDN3, Endothelin receptor B2, GRAMD3, MC1R, Melanophilin (MLPH), SLC45A2-MATP, SLCO1B3, SOX10, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria/?or+Taxon+ID=%9031%and+Trait=Colorationand+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

2 (<https://www.gephebase.org/search-criteria/?or+Gene+Gephebase=%PMEL17%and+Taxon+ID=%9031%or+Gene+Gephebase=%PMEL17%and+Taxon+ID=%9031%#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

@Dominance @AllelicSeries @Parallelism <https://omia.org/OMIA000373/9031/>