

PMEL17 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~PMEL17^#gephebase-summary-title)	Gephebase Gene	GP00000913	GepheID
Published	Entry Status	Martin	Main curator

Taxon A		Taxon B	
Taxon A	Latin Name	Taxon B	Latin Name
Gallus gallus (<a +gallus+gallus`"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">#gephebase-summary-title)	Gallus gallus (<a +gallus+gallus`"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">#gephebase-summary-title)	Gallus gallus (<a +gallus+gallus`"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">#gephebase-summary-title)	Gallus gallus (<a +gallus+gallus`"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">#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
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	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	Lineage
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 Intraspecies?		is Taxon B an Intraspecies?	
Yes		Yes	
Gallus gallus - wild pit gamecock	Taxon A Description	Gallus gallus - Dominant white pit gamecock	Taxon B Description

PMEL	Generic Gene Name	UniProtKB Gallus gallus
	Synonyms	Q98917 (http://www.uniprot.org/uniprot/Q98917)
SILV; MMP115; PMEL17	String	GenebankID or UniProtKB
9031.ENS GALP00000023540 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9031.ENS GALP00000023540)	Sequence Similarities	
Belongs to the PMEL/NMB family.	GO - Molecular Function	
-	GO - Biological Process	
GO:0032438 : melanosome organization (https://www.ebi.ac.uk/QuickGO/term/GO:0032438)		

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>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)	Presumptive Null
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)	Molecular Type
Insertion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Insertion^#gephebase-summary-title)	Aberration Type
1-9 bp	Insertion Size
insertion of amino acid 723-725 due to a 9bp-insert in exon 10	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title)	Experimental Evidence
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)	Main Reference
Kerje S; Sharma P; Gunnarsson U; Kim H; Bagchi S; Fredriksson R; SchÃ¼tz K; Jensen P; von Heijne G; Okimoto R; Andersson L	Authors

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

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=Coloration/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
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)	Related Haplotypes

EXTERNAL LINKS

COMMENTS

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