

## GEPHE SUMMARY

SLC45A2=MATP ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase= <sup>^</sup> SLC45A2=MATP <sup>^</sup> #gephebase-summary-title)	Gephebase Gene	GP00002294	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category= <sup>^</sup> Morphology <sup>^</sup> #gephebase-summary-title)	Trait Category		
Coloration (feathers) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> = <sup>^</sup> Coloration (feathers) <sup>^</sup> #gephebase-summary-title)	Trait		
WT feather	Trait State in Taxon A		
Silver feather color	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status= <sup>^</sup> Domesticated <sup>^</sup> #gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Gallus gallus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms= <sup>^</sup> Gallus gallus <sup>^</sup> #gephebase-summary-title)	Latin Name	Gallus gallus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms= <sup>^</sup> Gallus gallus <sup>^</sup> #gephebase-summary-title)	Latin Name
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) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030</a> )	Parent	Gallus () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030</a> )	Parent
9031 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031</a> )	NCBI Taxonomy ID	9031 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

SLC45A2	Generic Gene Name	Q9UMX9 ( <a href="http://www.uniprot.org/uniprot/Q9UMX9">http://www.uniprot.org/uniprot/Q9UMX9</a> )	UniProtKB Homo sapiens
1A1; AIM1; MATP; OCA4; SHEP5	Synonyms	XP_015703941 ( <a href="https://www.ncbi.nlm.nih.gov/nucore/XP_015703941">https://www.ncbi.nlm.nih.gov/nucore/XP_015703941</a> )	GenebankID or UniProtKB
9606.ENSPP0000296589 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP0000296589">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP0000296589</a> )	String		
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.	Sequence Similarities		
GO:0008506 : sucrose:proton symporter activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008506">https://www.ebi.ac.uk/QuickGO/term/GO:0008506</a> )	GO - Molecular Function		
GO:0042438 : melanin biosynthetic process ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042438">https://www.ebi.ac.uk/QuickGO/term/GO:0042438</a> )	GO - Biological Process		

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

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)

Molecular Type

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

Aberration Type

SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)

SNP Coding Change

Nonsense

Molecular Details of the Mutation

c.1039C>A p.L347M

Experimental Evidence

Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Leu	Met	347

Main Reference

Mutations in SLC45A2 cause plumage color variation in chicken and Japanese quail. (2007) (https://pubmed.ncbi.nlm.nih.gov/17151254)

Authors

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Abstract

S<sup>S</sup> (Silver), S<sup>N</sup> (wild type/gold), and S<sup>AL</sup> (sex-linked imperfect albinism) form a series of alleles at the S (Silver) locus on chicken (*Gallus gallus*) chromosome Z. Similarly, sex-linked imperfect albinism (AL<sup>A</sup>) is the bottom recessive allele at the orthologous AL locus in Japanese quail (*Coturnix japonica*). The solute carrier family 45, member 2, protein (SLC45A2), previously denoted membrane-associated transporter protein (MATP), has an important role in vesicle sorting in the melanocytes. Here we report five SLC45A2 mutations. The 106delT mutation in the chicken S<sup>AL</sup> allele results in a frameshift and a premature stop codon and the corresponding mRNA appears to be degraded by nonsense-mediated mRNA decay. A splice-site mutation in the Japanese quail AL<sup>A</sup> allele causes in-frame skipping of exon 4. Two independent missense mutations (Tyr277Cys and Leu347Met) were associated with the Silver allele in chicken. The functional significance of the former mutation, associated only with Silver in White Leghorn, is unclear. Ala72Asp was associated with the cinnamon allele (AL<sup>C</sup>) in the Japanese quail. The most interesting feature concerning the SLC45A2 variants documented in this study is the specific inhibition of expression of red pheomelanin in Silver chickens. This phenotypic effect cannot be explained on the basis of the current, incomplete, understanding of SLC45A2 function. It is an enigma why recessive null mutations at this locus cause an almost complete absence of both eumelanin and pheomelanin whereas some missense mutations are dominant and cause a specific inhibition of pheomelanin production.

Additional References

## RELATED GEPHE

Related Genes

13 (ABCA1, Agouti (ASIP), CDKN2A, CYP19A1, EDN3, Endothelin receptor B2, MC1R, Melanophilin (MLPH), PMEL17, 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 Haplotypes

2 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^SLC45A2=MATP^/and+Taxon ID=^9031^/or+Gene Gephebase=^SLC45A2=MATP^/and+Taxon ID=^9031^#gephebase-summary-title)

## EXTERNAL LINKS

## COMMENTS

@AllelicSeries https://omia.org/OMIA000915/9031/

