

GEPHE SUMMARY

	Gephebase Gene		GepheID
SLC45A2=MATP (<a +slc45a2='MATP`"#gephebase-summary-title"' href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+SLC45A2=MATP`"#gephebase-summary-title)		GP00001058	
	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)		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)			
	Trait State in Taxon A		
Coturnix japonica		Trait State in Taxon B	
Coturnix japonica - imperfect albinism			
	Ancestral State		
Data not curated		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)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Coturnix japonica (<a +coturnix+japonica`"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Coturnix+japonica`"#gephebase-summary-title)		Coturnix japonica (<a +coturnix+japonica`"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Coturnix+japonica`"#gephebase-summary-title)	
	Common Name		Common Name
Japanese quail		Japanese quail	
	Synonyms		Synonyms
Coturnix coturnix Japonicus; Coturnix coturnix japonica; Coturnix coturnix japonica; Coturnix japonica japonica; Japanese quail; Coturnix japonica Temminck & Schlegel, 1849		Coturnix coturnix Japonicus; Coturnix coturnix japonica; Coturnix coturnix japonica; Coturnix japonica japonica; Japanese quail; Coturnix japonica Temminck & Schlegel, 1849	
	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; Galloanserae; Galliformes; Phasianidae; Perdicinae; Coturnix		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; Perdicinae; Coturnix	
	Parent		Parent
Coturnix () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9090)		Coturnix () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9090)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
93934 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 93934)		93934 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 93934)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		Yes	
			Taxon B Description
		Coturnix japonica - imperfect albinism	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
SLC45A2		Q9UMX9 (http://www.uniprot.org/uniprot/Q9UMX9)	
	Synonyms		GenebankID or UniProtKB
1A1; AIM1; MATP; OCA4; SHEP5		XP_015703941 (https://www.ncbi.nlm.nih.gov/nucore/XP_015703941)	
	String		
9606.ENSPO00000296589 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO00000296589)			
	Sequence Similarities		
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.			
	GO - Molecular Function		
GO:0008506 : sucrose:proton symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008506)			

GO:0042438 : melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
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>)

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
SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title)	Aberration Type
-	SNP Coding Change
G->T transversion at the splice acceptor site just preceding exon 4; causes in-frame skipping of exon 4	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title)	Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Mutations in SLC45A2 cause plumage color variation in chicken and Japanese quail. (2007) (https://pubmed.ncbi.nlm.nih.gov/17151254)	Main Reference
Gunnarsson U; Hellstr�m AR; Tixier-Boichard M; Minvielle F; Bed�hom B; Ito S; Jensen P; Rattink A; Vereijken A; Andersson L	Authors
S*S (Silver), S*N (wild type/gold), and S*AL (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*A) 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*AL 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*A 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*C) 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.	Abstract
	Additional References

RELATED GEPHE

6 (Agouti, Endothelin receptor B, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~93934~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~SLC45A2=MATP~/and+Taxon ID=~93934~/or+Gene Gephebase=~SLC45A2=MATP~/and+Taxon ID=~93934~#gephebase-summary-title)	Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Splicing <https://omia.org/OMIA000370/93934/>

