GEPHE SUMMARY

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

GepheID
GP00001059

Entry Status
Published

Main curator
Martin

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

Trait State in Taxon B
Gallus gallus - imperfect albinism

Ancestral State
Taxon A

Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Gallus gallus^#gephebase-summary-title)

Common Name
chicken

Synonyms
Gallus gallus domesticus; chicken; bantam; chickens

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; Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus

Parent
Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)

NCBI Taxonomy ID
9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Gallus gallus^#gephebase-summary-title)

Common Name
chicken

Synonyms
Gallus gallus domesticus; chicken; bantam; chickens

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; Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus

Parent
Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)

NCBI Taxonomy ID
9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)

is Taxon B an Infraspecies?
Yes

Taxon B Description
Gallus gallus - imperfect albinism

GENOTYPIC CHANGE

Generic Gene Name
SLC45A2

Synonyms
1A1; AIM1; MATP; OCA4; SHEP5

String
9606.ENSP00000296589 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000296589)

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 - Biological Process

UniProtKB Homo sapiens
Q9UMX9 (http://www.uniprot.org/uniprot/Q9UMX9)

GenebankID or UniProtKB
XP_015703941 (https://www.ncbi.nlm.nih.gov/nuccore/XP_015703941)

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)

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

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

1bp deletion at codon 36; resulting in frameshift and a premature stop codon in exon 1; the corresponding mRNA appears to be degraded by Nonsense-mediated mRNA decay.

Experimental Evidence

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

Main Reference

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

Authors

Gunnarsson U; HellstrÃ¶m AR; Tixier-Boichard M; Minvielle F; Bed'hom B; Ito S; Jensen P; Rattink A; Vereijken A; Andersson L

Abstract

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.

Additional References

RELATED GEPHE

Related Genes

14 (ABCA1, Agouti (ASIP), CDKN2A, CYP19A1, EDN3, Endothelin receptor B2, GRAMD3, 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/