GEPHE SUMMARY

Gephebase Gene
SLCO1B3 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SLCO1B3^#gephebase-summary-title)

GepheID
GP00001064

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Coloration (blue eggs) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (blue eggs)^#gephebase-summary-title)

Trait State in Taxon A
Gallus gallus

Trait State in Taxon B
Gallus gallus - American blue-shelled breed

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 - American blue-shelled breed

GENOTYPIC CHANGE

Generic Gene Name
SLCO1B3

Synonyms
LST3; HBLRR; LST-2; OATP8; OATP-8; OATP1B3; SLC21A8; LST-3TM13; LST2

String
9606.ENSP00000261196 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000261196)

Sequence Similarities
Belongs to the organo anion transporter (TC 2.A.60) family.

GO - Molecular Function
GO:0008514 : organic anion transmembrane transporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008514)
GO:0015125 : bile acid transmembrane transporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015125)

UniProtKB Homo sapiens
Q9NPD5 (http://www.uniprot.org/uniprot/Q9NPD5)

GenebankID or UniProtKB
AER35427 (https://www.ncbi.nlm.nih.gov/nuccore/AER35427)

GO:0015347 : sodium-independent organic anion transmembrane transporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015347)

GO - Biological Process

GO:0015711 : organic anion transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015711)
GO:0015721 : bile acid and bile salt transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015721)
GO:0043252 : sodium-independent organic anion transport (https://www.ebi.ac.uk/QuickGO/term/GO:0043252)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005887 : integral component of plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0016323 : basolateral plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016323)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Insertion Size

1-10 kb

Molecular Details of the Mutation

TE (EAV-HP) promoter insertion resulting in uterus expression

Experimental Evidence

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

Main Reference

An EAV-HP insertion in 5′ Flanking region of SLCO1B3 causes blue eggshell in the chicken. (2013) (https://pubmed.ncbi.nlm.nih.gov/23359636)

Authors

Wang Z; Qu L; Yao J; Yang X; Li G; Zhang Y; Li J; Wang X; Bai J; Xu G; Deng X; Yang N; Wu C

Abstract

The genetic determination of eggshell coloration has not been determined in birds. Here we report that the blue eggshell is caused by an EAV-HP insertion that promotes the expression of SLCO1B3 gene in the uterus (shell gland) of the oviduct in chicken. In this study, the genetic map location of the blue eggshell gene was refined by linkage analysis in an F(2) chicken population, and four candidate genes within the refined interval were subsequently tested for their expression levels in the shell gland of the uterus from blue-shelled and non-blue-shelled hens. SLCO1B3 gene was found to be the only one expressed in the uterus of blue-shelled hens but not in that of non-blue-shelled hens. Results from a pyrosequencing analysis showed that only the allele of SLCO1B3 from blue-shelled chickens was expressed in the uterus of heterozygous hens (O*LC/O*N). SLCO1B3 gene belongs to the organic anion transporting polypeptide (OATP) family; and the OATPs, functioning as membrane transporters, have been reported for the transportation of amphipathic organic compounds, including bile salt in mammals. We subsequently resequenced the whole genomic region of SLCO1B3 and discovered an EAV-HP insertion in the 5′ flanking region of SLCO1B3. The EAV-HP insertion was found closely associated with blue eggshell phenotype following complete Mendelian segregation. In situ hybridization also demonstrated that the blue eggshell is associated with ectopic expression of SLCO1B3 in shell glands of uterus. Our finding strongly suggests that the EAV-HP insertion is the causative mutation for the blue eggshell phenotype. The insertion was also found in another Chinese blue-shelled breed and an American blue-shelled breed. In addition, we found that the insertion site in the blue-shelled chickens from Araucana is different from that in Chinese breeds, which implied independent integration events in the blue-shelled chickens from the two continents, providing a parallel evolutionary example at the molecular level.

Additional References

RELATED GEPHE

Related Genes

14 (ABCA1, Agouti (ASIP), CDKN2A, CYP19A1, EDN3, Endothelin receptor B2, GRAMD3, MC1R, Melanophilin (MLPH), PMEL17, SLC45A2=MATP, 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

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^SLCO1B3^/and+Taxon ID=^9031^/or+Gene Gephebase=^SLCO1B3^/and+Taxon ID=^9031^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

2 independent insertions