

GEPHE SUMMARY

SLCO1B3 (https://www.gephebase.org/search-criteria/?and+Gene Gephebase=%SLCO1B3%#gephebase-summary-title)	Gephebase Gene	GP00001065	GephelD
Published	Entry Status	Courtier	Main curator

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 - Chinese 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	Taxon B	Latin Name
Gallus gallus (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Gallus+gallus%#gephebase-summary-title)		Gallus gallus (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Gallus+gallus%#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
	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; Phasianinae; Gallus		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	
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
No	is Taxon A an Infraspecies?	Yes	is Taxon B an Infraspecies?
			Taxon B Description
			Gallus gallus - Chinese blue-shelled breed

GENOTYPIC CHANGE

SLCO1B3	Generic Gene Name	UniProtKB Homo sapiens
LST3; HBLRR; LST-2; OATP8; OATP-8; OATP1B3; SLC21A8; LST-3TM13; LST2	Synonyms	GenebankID or UniProtKB
9606.ENSP00000261196 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000261196)	String	AER35427 (https://www.ncbi.nlm.nih.gov/nuccore/AER35427)
Belongs to the organo anion transporter (TC 2.A.60) family.	Sequence Similarities	
GO:0008514 : organic anion transmembrane transporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008514)	GO - Molecular Function	
GO:0015125 : bile acid transmembrane transporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015125)		

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=%27No%27#gephebase-summary-title>)

Molecular Type

Cis-regulatory (<https://www.gephebase.org/search-criteria/?and+Molecular+Type=%27Cis-regulatory%27#gephebase-summary-title>)

Aberration Type

Insertion (<https://www.gephebase.org/search-criteria/?and+Aberration+Type=%27Insertion%27#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=%27Linkage+Mapping%27#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 amphiphatic 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=%279031%27+and+Trait=Coloration+and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

1 (<https://www.gephebase.org/search-criteria/?or+Gene+Gephebase=%27SLCO1B3%27+and+Taxon+ID=%279031%27+or+Gene+Gephebase=%27SLCO1B3%27+and+Taxon+ID=%279031%27#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

2 independent insertions