

GEPHE SUMMARY

Gephebase Gene
SLCO1B3

Entry Status
Published

GepheID
GP00001064

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (blue eggs)

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

Taxon A

Latin Name
Gallus gallus

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)

NCBI Taxonomy ID
9031

is Taxon A an Intraspecies?
No

Taxon B

Latin Name
Gallus gallus

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)

NCBI Taxonomy ID
9031

is Taxon B an Intraspecies?
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

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

GO - Molecular Function
GO:0008514 : organic anion transmembrane transporter activity
GO:0015125 : bile acid transmembrane transporter activity
GO:0015347 : sodium-independent organic anion transmembrane transporter activity

GO - Biological Process
GO:0015711 : organic anion transport
GO:0015721 : bile acid and bile salt transport
GO:0043252 : sodium-independent organic anion transport

UniProtKB Homo sapiens
Q9NPD5

GenebankID or UniProtKB
AER35427

GO - Cellular Component

GO:0005886 : plasma membrane

GO:0005887 : integral component of plasma membrane

GO:0016323 : basolateral plasma membrane

Presumptive Null

No

Molecular Type

Cis-regulatory

Aberration Type

Insertion

Insertion Size

1-10 kb

Molecular Details of the Mutation

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

Experimental Evidence

Linkage Mapping

Main Reference

An EAV-HP insertion in 5' Flanking region of *SLCO1B3* causes blue eggshell in the chicken. (2013)

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

12 (*ABCA1*, *Agouti* (*ASIP*), *CDKN2A*, *CYP19A1*, *EDN3*, *Endothelin receptor B2*, *MC1R*, *Melanophilin* (*MLPH*), *PMEL17*, *SLC45A2=MATP*, *SOX10*, *tyrosinase-related protein 1* (*TYRP1*))

Related Haplotypes

1

EXTERNAL LINKS

COMMENTS

2 independent insertions