

## GEPHE SUMMARY

**Gephebase Gene**  
BTN1A1

**Entry Status**  
Published

**GepheID**  
GP00002160

**Main curator**  
Martin

## PHENOTYPIC CHANGE

**Trait Category**  
Physiology

**Trait**  
Pathogen resistance (virus)

**Trait State in Taxon A**  
Sensitive to type C avian leukosis viruses

**Trait State in Taxon B**  
Resistant to type C avian leukosis viruses (autosomal recessive)

**Ancestral State**  
Unknown

**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?**  
No

## GENOTYPIC CHANGE

**Generic Gene Name**  
BTN1A1

**Synonyms**  
BT; BTN; BTN1

**String**  
9606.ENSP00000244513

**Sequence Similarities**  
Belongs to the immunoglobulin superfamily. BTN/MOG family.

**GO - Molecular Function**  
GO:0005102 : signaling receptor binding  
GO:0038023 : signaling receptor activity

**GO - Biological Process**  
GO:0050852 : T cell receptor signaling pathway  
GO:0050776 : regulation of immune response  
GO:0001817 : regulation of cytokine production

**GO - Cellular Component**  
GO:0005886 : plasma membrane  
GO:0005887 : integral component of plasma membrane

**UniProtKB Homo sapiens**  
Q13410

**GenebankID or UniProtKB**

GO:0005615 : extracellular space  
GO:0009897 : external side of plasma membrane

#### Presumptive Null

Yes

#### Molecular Type

Coding

#### Aberration Type

SNP

#### SNP Coding Change

Nonsense

#### Molecular Details of the Mutation

g.808011C>A c.165C>A p.C55\*

#### Experimental Evidence

Linkage Mapping

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Cys	STP	55

#### Main Reference

The receptor for the subgroup C avian sarcoma and leukosis viruses, Tvc, is related to mammalian butyrophilins, members of the immunoglobulin superfamily. (2005)

#### Authors

Elleder D; Stepanets V; Melder DC; Senigl F; Geryk J; Pajer P; Plach<sup>1</sup>/<sub>2</sub> J; Hejnar J; Svoboda J; Federspiel MJ

#### Abstract

The five highly related envelope subgroups of the avian sarcoma and leukosis viruses (ASLVs), subgroup A [ASLV(A)] to ASLV(E), are thought to have evolved from an ancestral envelope glycoprotein yet utilize different cellular proteins as receptors. Alleles encoding the subgroup A ASLV receptors (Tva), members of the low-density lipoprotein receptor family, and the subgroup B, D, and E ASLV receptors (Tvb), members of the tumor necrosis factor receptor family, have been identified and cloned. However, alleles encoding the subgroup C ASLV receptors (Tvc) have not been cloned. Previously, we established a genetic linkage between tvc and several other nearby genetic markers on chicken chromosome 28, including tva. In this study, we used this information to clone the tvc gene and identify the Tvc receptor. A bacterial artificial chromosome containing a portion of chicken chromosome 28 that conferred susceptibility to ASLV(C) infection was identified. The tvc gene was identified on this genomic DNA fragment and encodes a 488-amino-acid protein most closely related to mammalian butyrophilins, members of the immunoglobulin protein family. We subsequently cloned cDNAs encoding Tvc that confer susceptibility to infection by subgroup C viruses in chicken cells resistant to ASLV(C) infection and in mammalian cells that do not normally express functional ASLV receptors. In addition, normally susceptible chicken DT40 cells were resistant to ASLV(C) infection after both tvc alleles were disrupted by homologous recombination. Tvc binds the ASLV(C) envelope glycoproteins with low-nanomolar affinity, an affinity similar to that of binding of Tva and Tvb with their respective envelope glycoproteins. We have also identified a mutation in the tvc gene in line L15 chickens that explains why this line is resistant to ASLV(C) infection.

#### Additional References

## RELATED GEPHE

#### Related Genes

2 (MX1, Tva)

#### Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS