

GEPHE SUMMARY

Gephebase Gene
CDKN2A

Entry Status
Published

GepheID
GP00000178

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (feathers ; sex-linked dilution)

Trait State in Taxon A
Gallus gallus

Trait State in Taxon B
B0/B2 White Leghorn males with Sex-linked dilution allele

Ancestral State
Taxon A

Taxonomic Status
Domesticated

	Taxon A	Taxon B
Latin Name	<i>Gallus gallus</i>	<i>Gallus gallus</i>
Common Name	chicken	chicken
Synonyms	Gallus gallus domesticus; chicken; bantam; chickens	Gallus gallus domesticus; chicken; bantam; chickens
Rank	species	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	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)	Gallus () - (Rank: genus)
NCBI Taxonomy ID	9031	9031
is Taxon A an Intraspecies?	No	No

GENOTYPIC CHANGE

Generic Gene Name
CDKN2A

Synonyms
ARF; MLM; P14; P16; P19; CMM2; INK4; MTS1; TP16; CDK4; CDKN2; INK4A; MTS-1; P14ARF; P19ARF; P16INK4; P16INK4A; P16-INK4A

String
9606.ENSP00000394932

Sequence Similarities
Belongs to the CDKN2 cyclin-dependent kinase inhibitor family.

GO - Molecular Function
GO:0003723 : RNA binding
GO:0019901 : protein kinase binding
GO:0004861 : cyclin-dependent protein serine/threonine kinase inhibitor activity
GO:0051059 : NF-kappaB binding

GO - Biological Process
GO:0045892 : negative regulation of transcription, DNA-templated
GO:0008285 : negative regulation of cell proliferation
GO:0007050 : cell cycle arrest

UniProtKB Homo sapiens
P42771

GenebankID or UniProtKB
NP_001308484

GO:0090398 : cellular senescence
 GO:0000082 : G1/S transition of mitotic cell cycle
 GO:0030308 : negative regulation of cell growth
 GO:0001953 : negative regulation of cell-matrix adhesion
 GO:0045736 : negative regulation of cyclin-dependent protein serine/threonine kinase activity
 GO:0032088 : negative regulation of NF-kappaB transcription factor activity
 GO:0042326 : negative regulation of phosphorylation
 GO:2000774 : positive regulation of cellular senescence
 GO:2000111 : positive regulation of macrophage apoptotic process
 GO:0034393 : positive regulation of smooth muscle cell apoptotic process
 GO:0007265 : Ras protein signal transduction
 GO:0090399 : replicative senescence
 GO:0035986 : senescence-associated heterochromatin focus assembly

GO - Cellular Component

GO:0005737 : cytoplasm
 GO:0005829 : cytosol
 GO:0005634 : nucleus
 GO:0035985 : senescence-associated heterochromatin focus

Mutation #1

Presumptive Null

No

Molecular Type

Cis-regulatory

Aberration Type

Complex Change

Molecular Details of the Mutation

12kb haplotype including 2 non-coding mutations

Experimental Evidence

Linkage Mapping

Main Reference

The evolution of Sex-linked barring alleles in chickens involves both regulatory and coding changes in CDKN2A. (2017)

Authors

Schwochow Thalman D; Ring H; Sundström E; Cao X; Larsson M; Kerje S; Hågglund A; Fogelholm J; Wright D; Jemth P; Hallböök F; Bed'Hom B; Dorshorst B; Tixier-Boichard M; Andersson L

Abstract

Sex-linked barring is a fascinating plumage pattern in chickens recently shown to be associated with two non-coding and two missense mutations affecting the ARF transcript at the CDKN2A tumor suppressor locus. It however remained a mystery whether all four mutations are indeed causative and how they contribute to the barring phenotype. Here, we show that Sex-linked barring is genetically heterogeneous, and that the mutations form three functionally different variant alleles. The B0 allele carries only the two non-coding changes and is associated with the most dilute barring pattern, whereas the B1 and B2 alleles carry both the two non-coding changes and one each of the two missense mutations causing the Sex-linked barring and Sex-linked dilution phenotypes, respectively. The data are consistent with evolution of alleles where the non-coding changes occurred first followed by the two missense mutations that resulted in a phenotype more appealing to humans. We show that one or both of the non-coding changes are cis-regulatory mutations causing a higher CDKN2A expression, whereas the missense mutations reduce the ability of ARF to interact with MDM2. Caspase assays for all genotypes revealed no apoptotic events and our results are consistent with a recent study indicating that the loss of melanocyte progenitors in Sex-linked barring in chicken is caused by premature differentiation and not apoptosis. Our results show that CDKN2A is a major locus driving the differentiation of avian melanocytes in a temporal and spatial manner.

Additional References

Sex-linked barring in chickens is controlled by the CDKN2A /B tumour suppressor locus. (2010)

Mutation #2

Presumptive Null

No

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

g.78636802 c.28C>T p.R10C

Experimental Evidence

Linkage Mapping

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	Cys	10

Main Reference

The evolution of Sex-linked barring alleles in chickens involves both regulatory and coding changes in CDKN2A. (2017)

Authors

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Additional References

[Sex-linked barring in chickens is controlled by the CDKN2A /B tumour suppressor locus. \(2010\)](#)

RELATED GEPHE

Related Genes

12 (ABCA1, Agouti (ASIP), CYP19A1, EDN3, Endothelin receptor B2, MC1R, Melanophilin (MLPH), PMEL17, SLC45A2=MATP, SLC01B3, SOX10, tyrosinase-related protein 1 (TYRP1))

Related Haplotypes

1

EXTERNAL LINKS

COMMENTS

@SexualTrait @ComplexAllele @AllelicSeries <https://omia.org/OMIA000102/9031/>