GEPHE SUMMARY

Gephebase Gene

CYP19A1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^CYP19A1^#gephebase-summary-title)

GepheID

GP00002050

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

Trait State in Taxon A

wild-type

Trait State in Taxon B

Henny feathering - dominant mutation that transforms male-specific plumage to female-like plumage

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?

No

GENOTYPIC CHANGE

Generic Gene Name

CYP19A1

Synonyms

ARO; ARO1; CPV1; CYAR; CYP19; CYPXIX; P-450AROM

String

9606.ENSP00000379683 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000379683)

Sequence Similarities

Belongs to the cytochrome P450 family.

GO - Molecular Function

GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)
GO:0005506 : iron ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005506)
GO:0070330 : aromatase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0070330)
GO:0009055 : electron transfer activity (https://www.ebi.ac.uk/QuickGO/term/GO:0009055)

UniProtKB Homo sapiens

P11511 (http://www.uniprot.org/uniprot/P11511)

GenebankID or UniProtKB

()

GO:0016712 : oxidoreductase activity, acting on paired donors, with incorporation or reduction of molecular oxygen, reduced flavin or flavoprotein as one donor, and incorporation of one atom of oxygen (https://www.ebi.ac.uk/QuickGO/term/GO:0016712)
GO:0008395 : steroid hydroxylase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008395)
GO:0019825 : oxygen binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019825)

GO - Biological Process

GO:0060736 : prostate gland growth (https://www.ebi.ac.uk/QuickGO/term/GO:0060736)
GO:0016125 : sterol metabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0016125)
GO:0008585 : female gonad development (https://www.ebi.ac.uk/QuickGO/term/GO:0008585)
GO:0030879 : mammary gland development (https://www.ebi.ac.uk/QuickGO/term/GO:0030879)
GO:0006694 : steroid biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006694)
GO:0030540 : female genitalia development (https://www.ebi.ac.uk/QuickGO/term/GO:0030540)
GO:0006710 : androgen catabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006710)
GO:0006703 : estrogen biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006703)
GO:0002677 : negative regulation of chronic inflammatory response (https://www.ebi.ac.uk/QuickGO/term/GO:0002677)
GO:0010760 : negative regulation of macrophage chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0010760)
GO:2000866 : positive regulation of estradiol secretion (https://www.ebi.ac.uk/QuickGO/term/GO:2000866)
GO:0061370 : testosterone biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0061370)
GO:0060065 : uterus development (https://www.ebi.ac.uk/QuickGO/term/GO:0060065)

GO - Cellular Component

GO:0016021 : integral component of membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0016020 : membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)
GO:0005783 : endoplasmic reticulum (https://www.ebi.ac.uk/QuickGO/term/GO:0005783)
GO:0005789 : endoplasmic reticulum membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005789)

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

7524â€‰bp insertion at the 5′end of CYP19A1 of an intact endogenous retrovirus (99% sequence identity to the avian leukosis virus ev-1 and ev-21 strains suggesting a recent integration) - The ERV 3′LTR contains a powerful transcriptional enhancer and core promoter with TATA box.

Experimental Evidence

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

Main Reference

Characterization of the endogenous retrovirus insertion in CYP19A1 associated with henny feathering in chicken. (2019) (https://pubmed.ncbi.nlm.nih.gov/31467598)

Authors

Li J; Davis BW; Jern P; Dorshorst BJ; Siegel PB; Andersson L

Abstract

Henny feathering in chickens is determined by a dominant mutation that transforms male-specific plumage to female-like plumage. Previous studies indicated that this phenotype is caused by ectopic expression in skin of CYP19A1 encoding aromatase that converts androgens to estrogen and thereby inhibits the development of male-specific plumage. A long terminal repeat (LTR) from an uncharacterized endogenous retrovirus (ERV) insertion was found in an isoform of the CYP19A1 transcript from henny feathering chicken. However, the complete sequence and the genomic position of the insertion were not determined.

We used publicly available whole genome sequence data to determine the flanking sequences of the ERV, and then PCR amplified the entire insertion and sequenced it using Nanopore long reads and Sanger sequencing. The 7524â€‰bp insertion contains an intact endogenous retrovirus that was not found in chickens representing 31 different breeds not showing henny feathering or in samples of the ancestral red junglefowl. The sequence shows over 99% sequence identity to the avian leukosis virus ev-1 and ev-21 strains, suggesting a recent integration. The ERV 3′LTR, containing a powerful transcriptional enhancer and core promoter with TATA box together with binding sites for EFIII and Ig/EBP inside the CYP19A1 5′ untranslated region, was detected partially in an aromatase transcript, which present a plausible explanation for ectopic expression of aromatase in non-ovarian tissues underlying the henny feathering phenotype.

We demonstrate that the henny feathering allele harbors an insertion of an intact avian leukosis virus at the 5′end of CYP19A1. The presence of this ERV showed complete concordance with the henny feathering phenotype both within a pedigree segregating for this phenotype and across breeds.

Additional References

RELATED GEPHE

Related Genes

14 (ABCA1, Agouti (ASIP), CDKN2A, EDN3, Endothelin receptor B2, GRAMD3, MC1R, Melanophilin (MLPH), PMEL17, SLC45A2=MATP, SLCO1B3, 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

No matches found.

EXTERNAL LINKS

COMMENTS

@TE - ectopic expression of aromatase in non-ovarian tissues underlying the henny feathering phenotype https://omia.org/OMIA000452/9031/