GEPHE SUMMARY

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

GepheID
GP00002308

Entry Status
Published

Main curator
Martin

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
WT

Trait State in Taxon B
Recessive white from various meat breeds

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
Tyr

Synonyms
c; Oca1; skc35; albino

String
10090.ENSMUSP00000004770 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770)

Sequence Similarities
Belongs to the tyrosinase family.

GO - Molecular Function
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)
GO:0005507 : copper ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005507)
GO:0004503 : monophenol monooxygenase activity

UniProtKB Mus musculus
P11344 (http://www.uniprot.org/uniprot/P11344)

GenebankID or UniProtKB
()

(https://www.ebi.ac.uk/QuickGO/term/GO:0004503)

GO - Biological Process

GO:0042438 : melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0008283 : cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008283)
GO:0033280 : response to vitamin D (https://www.ebi.ac.uk/QuickGO/term/GO:0033280)
GO:0051591 : response to cAMP (https://www.ebi.ac.uk/QuickGO/term/GO:0051591)
GO:0009411 : response to UV (https://www.ebi.ac.uk/QuickGO/term/GO:0009411)
GO:0048538 : thymus development (https://www.ebi.ac.uk/QuickGO/term/GO:0048538)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0043231 : intracellular membrane-bounded organelle
(https://www.ebi.ac.uk/QuickGO/term/GO:0043231)
GO:0048471 : perinuclear region of cytoplasm
(https://www.ebi.ac.uk/QuickGO/term/GO:0048471)
GO:0042470 : melanosome (https://www.ebi.ac.uk/QuickGO/term/GO:0042470)
GO:0033162 : melanosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0033162)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)

Aberration Type

Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title)

Deletion Size

1-9 bp

Molecular Details of the Mutation

c.817_822del6 p.237-238delDW

Experimental Evidence

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

Main Reference

Autosomal albino chicken mutation (ca/ca) deletes hexanucleotide (-deltaGACTGG817) at a copper-binding site of the tyrosinase gene. (2000) (https://pubmed.ncbi.nlm.nih.gov/10685888)

Authors

Tobita-Teramoto T; Jang GY; Kino K; Salter DW; Brumbaugh J; Akiyama T

Abstract

We compared tyrosinase cDNA sequences from a line of autosomal albino and Black Silky chickens isolated from cultured melanocytes by reverse transcription-polymerase chain reaction (RT-PCR). Both sources produce a single DNA fragment of predicted normal tyrosinase size. Direct sequencing of the PCR product showed three mutated sites in the tyrosinase gene of the albino chicken. Two silent point mutations and a deletion of six nucleotides (-deltaGACTGG) at 817 bp in the tyrosinase cDNA sequence were observed when compared with the White Leghorn and Black Silky cDNA sequences. The deduced albino chicken tyrosinase protein lacks two amino acids, aspartic acid and tryptophan. The position of these amino acids is consistent with one of the potential copper-binding sites that should be indispensable for function of the enzyme. We speculate that the six-base deletion is responsible for the inactive tyrosinase in this line of albino chickens.

Additional References

RELATED GEPHE

Related Genes

14 (ABCA1, Agouti (ASIP), CDKN2A, CYP19A1, EDN3, Endothelin receptor B2, GRAMD3, MC1R, Melanophilin (MLPH), PMEL17, SLC45A2=MATP, SLCO1B3, SOX10, 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

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^tyrosinase (TYR)^/and+Taxon ID=^9031^/or+Gene Gephebase=^tyrosinase (TYR)^/and+Taxon ID=^9031^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@Parallelism https://omia.org/OMIA000369/9031/ ; Recessive white is selected in the meat industry (as well as a dominant white)