

GEPHE SUMMARY

tyrosinase (TYR) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+tyrosinase+(TYR)^#gephebase-summary-title)	Gephebase Gene	GP00002308	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category+Morphology^#gephebase-summary-title)	Trait Category		
Coloration (feathers) (https://www.gephebase.org/search-criteria?/and+Trait+Coloration+(feathers)^#gephebase-summary-title)	Trait		
WT	Trait State in Taxon A		
Recessive white from various meat breeds	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated^#gephebase-summary-title)	Taxonomic Status		
	Taxon A	Taxon B	
Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Gallus+gallus^#gephebase-summary-title)	Latin Name	Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Gallus+gallus^#gephebase-summary-title)	Latin Name
chicken	Common Name	chicken	Common Name
Gallus gallus domesticus; chicken; bantam; chickens	Synonyms	Gallus gallus domesticus; chicken; bantam; chickens	Synonyms
species	Rank	species	Rank
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	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	Lineage
Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030)	Parent	Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030)	Parent
9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031)	NCBI Taxonomy ID	9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

Tyr	Generic Gene Name	P11344 (http://www.uniprot.org/uniprot/P11344)	UniProtKB Mus musculus
c; Oca1; skc35; albino	Synonyms	()	GenebankID or UniProtKB
10090.ENSMUSP00000004770 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770)	String		
Belongs to the tyrosinase family.	Sequence Similarities		
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)	GO - Molecular Function		
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			

(<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 ([#ghepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null+No))

Molecular Type

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

Aberration Type

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

c.817_822del6 p.237-238delDW

Experimental Evidence

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

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

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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, SLC01B3, SOX10, tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID+9031+and+Trait+Coloration+and+group+Haplotypes=true#ghepbase-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#ghepbase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase+tyrosinase+(TYR)+and+Taxon+ID+9031+or+Gene+Gephebase+tyrosinase+(TYR)+and+Taxon+ID+9031#ghepbase-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)

