

## GEPHE SUMMARY

tyrosinase (TYR) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase="tyrosinase (TYR)"#gephebase-summary-title)	Gephebase Gene	GP00002309	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category="Morphology"#gephebase-summary-title)	Trait Category		
Coloration (feathers) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> ="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 ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status="Domesticated"#gephebase-summary-title)	Taxonomic Status		
	Taxon A	Taxon B	
Gallus gallus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms="Gallus gallus"#gephebase-summary-title)	Latin Name	Gallus gallus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> 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) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030</a> )	Parent	Gallus () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030</a> )	Parent
9031 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031</a> )	NCBI Taxonomy ID	9031 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

Tyr	Generic Gene Name	P11344 ( <a href="http://www.uniprot.org/uniprot/P11344">http://www.uniprot.org/uniprot/P11344</a> )	UniProtKB Mus musculus
c; Oca1; skc35; albino	Synonyms	()	GenebankID or UniProtKB
10090.ENSMUSP00000004770 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770</a> )	String		
Belongs to the tyrosinase family.	Sequence Similarities		
GO:0042803 : protein homodimerization activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042803">https://www.ebi.ac.uk/QuickGO/term/GO:0042803</a> )	GO - Molecular Function		
GO:0046982 : protein heterodimerization activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046982">https://www.ebi.ac.uk/QuickGO/term/GO:0046982</a> )			
GO:0005507 : copper ion binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005507">https://www.ebi.ac.uk/QuickGO/term/GO:0005507</a> )			
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 ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No^#gephebase-summary-title](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](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding^#gephebase-summary-title))

Aberration Type

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

Insertion Size

1-10 kb

Molecular Details of the Mutation

insertion of a complete avian retroviral sequence in intron 4 causes aberrant transcripts lacking exon 5

Experimental Evidence

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

Main Reference

Complete association between a retroviral insertion in the tyrosinase gene and the recessive white mutation in chickens. (2006) (<https://pubmed.ncbi.nlm.nih.gov/16457736>)

Authors

Chang CM; Coville JL; Coquerelle G; Gourichon D; Oulmouden A; Tixier-Boichard M

Abstract

In chickens, three mutant alleles have been reported at the C locus, including the albino mutation, and the recessive white mutation, which is characterized by white plumage and pigmented eyes. The albino mutation was found to be a 6 bp deletion in the tyrosinase (TYR) gene. The present work describes an approach to identify the structural rearrangement in the TYR gene associated with the recessive white mutation.

Molecular analysis of the chicken TYR gene has revealed a major structural difference (Restriction Fragment Length Polymorphism, RFLP) in the genomic DNA of the recessive white chicken. A major size difference of 7.7 kb was found in intron 4 of the TYR gene by long-range PCR. Molecular cloning and sequencing results showed the insertion of a complete avian retroviral sequence of the Avian Leukosis Virus (ALV) family. Several aberrant transcripts of the tyrosinase gene were found in 10 week old recessive white chickens but not in the homozygous wild type colored chicken. We established a rapid genotyping diagnostic test based on the discovery of this retroviral insertion. It shows that all homozygous carriers of this insertion had a white plumage in various chicken strains. Furthermore, it was possible to distinguish heterozygous carriers from homozygous normal chickens in a segregating line.

In this study, we conclude that the insertion of a complete avian retroviral sequence in intron 4 of the tyrosinase gene is diagnostic of the recessive white mutation in chickens. This insertion causes aberrant transcripts lacking exon 5, and we propose that this insertion is the causal mutation for the recessive white allele in the chicken.

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+groupHaplotypes=true#gephebase-summary-title](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](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)

