

GEPHE SUMMARY

<p>tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^tyrosinase-related+protein+1+(TYRP1)^#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>GP00002126</p> <p>Santos</p> <p>Entry Status</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)</p> <p>Coloration (feathers) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration(feathers)^#gephebase-summary-title)</p> <p>nonâ€chocolate plumage</p> <p>chocolate plumage</p> <p>Unknown</p> <p>Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus+gallus^#gephebase-summary-title)</p> <p>chicken</p> <p>Gallus gallus domesticus; chicken; bantam; chickens</p> <p>species</p> <p>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</p> <p>Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030)</p> <p>9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031)</p> <p>No</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Infrasppecies?</p>	<p>Taxon B</p> <p>Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Gallus+gallus^#gephebase-summary-title)</p> <p>chicken</p> <p>Gallus gallus domesticus; chicken; bantam; chickens</p> <p>species</p> <p>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</p> <p>Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030)</p> <p>9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031)</p> <p>No</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Infrasppecies?</p>
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GENOTYPIC CHANGE

<p>Tyrp1</p> <p>b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1</p> <p>10090.ENSMUSP00000006151 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)</p> <p>Belongs to the tyrosinase family.</p> <p>GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)</p> <p>GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)</p> <p>GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P07147 (http://www.uniprot.org/uniprot/P07147)</p> <p>0</p>	<p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
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GO:0004497 : monoxygenase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004497>)

GO - Biological Process

GO:0032438 : melanosome organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
GO:0048023 : positive regulation of melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)
GO:0006583 : melanin biosynthetic process from tyrosine
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006583>)
GO:0030318 : melanocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)
GO:0043438 : acetoacetic acid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043438>)
GO:0006582 : melanin metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006582>)

GO - Cellular Component

GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
GO:0030669 : clathrin-coated endocytic vesicle membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030669>)
GO:0010008 : endosome membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010008>)
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

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

g.30830367C>A c.640C>A p.His214Asn

Experimental Evidence

Linkage Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	His	Asn	214

Main Reference

A missense mutation in TYRP1 causes the chocolate plumage color in chicken and alters melanosome structure. (2019) (<https://pubmed.ncbi.nlm.nih.gov/30457703/>)

Authors

Li J; Bed'hom B; Marthey S; Valade M; Dureux A; Moroldo M; PÄ©choux C; Coville JL; Gourichon D; Vieaud A; Dorshorst B; Andersson L; Tixier-Boichard M

Abstract

The chocolate plumage color in chickens is due to a sex-linked recessive mutation, choc, which dilutes eumelanin pigmentation. Because TYRP1 is sex-linked in chickens, and TYRP1 mutations determine brown coat color in mammals, TYRP1 appeared as the obvious candidate gene for the choc mutation. By combining gene mapping with gene capture, a complete association was identified between the chocolate phenotype and a missense mutation leading to a His214Asn change in the ZnA zinc-binding domain of the protein. A diagnostic test confirmed complete association by screening 428 non-chocolate chickens of various origins. This is the first TYRP1 mutation described in the chicken. Electron microscopy analysis showed that melanosomes were more numerous in feather follicles of chocolate chickens but exhibited an abnormal structure characterized by a granular content and an irregular shape. A similar altered morphology was observed on melanosomes of another TYRP1 mutant in birds, the roux mutation of the quail.

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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 (TYR)) (<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

@parallelism <https://omia.org/OMIA001259/9031/>