

tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~tyrosinase-related+protein+1+(TYRP1)^#gephebase-summary-title)	Gephebase Gene	GP00002235	GepheID
Published	Entry Status	Martin	Main curator

	Trait Category
Morphology (#https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology)	
	Trait
Coloration (coat) (#https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat)	
	Trait State in Taxon A
Canis familiaris - various breeds	
	Trait State in Taxon B
Canis familiaris - Lancashire heeler	
	Ancestral State
Taxon A	
	Taxonomic Status
Domesticated (#https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated)	

GENOTYPIC CHANGE

Tyrp1	Generic Gene Name	UniProtKB Mus musculus
	Synonyms	GenebankID or UniProtKB
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1	String	
10090.ENSMUSP00000006151 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)	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:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)		

GO:0004497 : monooxygenase 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

Nonsense

Molecular Details of the Mutation

c.1025T>G p.Phe342Cys

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Phe	Cys	342

Main Reference

A novel TYRP1 variant is associated with liver and tan coat colour in Lancashire Heelers. (2019) (<https://pubmed.ncbi.nlm.nih.gov/31468558>)

Authors

Wright HE; Schofield E; Mellersh CS; Burmeister LM

Abstract

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Additional References

RELATED GEPHE

Related Genes

13 (Agouti (ASIP), GPR22, MFSD12, PMEL17, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PSMB7, beta-defensin 103 (CBD103), RALY (hnRNP associated with lethal yellow)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~9612~/and+Trait=Coloration/or+Taxon ID=~9615~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

4 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~tyrosinase-related protein 1 \(TYRP1\)~/and+Taxon ID=~9612~/or+Gene Gephebase=~tyrosinase-related protein 1 \(TYRP1\)~/and+Taxon ID=~9615~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~tyrosinase-related protein 1 (TYRP1)~/and+Taxon ID=~9612~/or+Gene Gephebase=~tyrosinase-related protein 1 (TYRP1)~/and+Taxon ID=~9615~#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

