

tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~tyrosinase-related+protein+1+(TYRP1)^#gephebase-summary-title)	Gephebase Gene	GP00002240	GepheID
Published	Entry Status	Martin	Main curator

Morphology (#gephebase-summary-title)	Trait Category
Coloration (coat) (#gephebase-summary-title)	Trait
Black coat (dominant WT allele)	Trait State in Taxon A
Lighter coat (recessive)	Trait State in Taxon B
Taxon A	Ancestral State
Domesticated (#gephebase-summary-title)	Taxonomic Status

GENOTYPIC CHANGE

Tyrp1	Generic Gene Name	P07147 (http://www.uniprot.org/uniprot/P07147)	UniProtKB Mus musculus
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1	Synonyms	ABG76825 (https://www.ncbi.nlm.nih.gov/nuccore/ABG76825)	GenebankID or UniProtKB
10090.ENSMUSP00000006151 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)	String		
Belongs to the tyrosinase family.	Sequence Similarities		
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	GO - Molecular Function		

(<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

Yes (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes~#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.570G>A p.Trp190ter

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	Trp	STP	190

Main Reference

A premature stop codon in the TYRP1 gene is associated with brown coat colour in the European rabbit (*Oryctolagus cuniculus*). (2014) (<https://pubmed.ncbi.nlm.nih.gov/24814776>)

Authors

Utzeri VJ; Ribani A; Fontanesi L

Abstract

Classical genetic studies in European rabbits (*Oryctolagus cuniculus*) suggested the presence of two alleles at the brown coat colour locus: a wild-type B allele that gives dense black pigment throughout the coat and a recessive b allele that in the homozygous condition (b/b genotype) produces brown rabbits that are unable to develop black pigmentation. In several other species, this locus is determined by mutations in the tyrosinase-related protein 1 (TYRP1) gene, encoding a melanocyte enzyme needed for the production of dark eumelanin. In this study, we investigated the rabbit TYRP1 gene as a strong candidate for the rabbit brown coat colour locus. A total of 3846 bp of the TYRP1 gene were sequenced in eight rabbits of different breeds and identified 23 single nucleotide polymorphisms (SNPs; 12 in intronic regions, five in exons and six in the 3'-untranslated region) and an insertion/deletion of 13 bp, in the 3'-untranslated region, organised in a few haplotypes. A mutation in exon 2 (g.41360196G>A) leads to a premature stop codon at position 190 of the deduced amino acid sequence (p.Trp190ter). Therefore, translation predicts a truncated TYRP1 protein lacking almost completely the tyrosinase domain. Genotyping 203 rabbits of 32 different breeds identified this mutation only in brown Havana rabbits. Its potential functional relevance in disrupting the TYRP1 protein and its presence only in brown animals strongly argue for this non-sense mutation being a causative mutation for the recessive b allele at the brown locus in *Oryctolagus cuniculus*.

Â© 2014 Stichting International Foundation for Animal Genetics.

Additional References

RELATED GEPHE

Related Genes

4 (Agouti (ASIP), MC1R, Melanophilin (MLPH), tyrosinase (TYR)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~9986~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001249/9986/>