

Gephebase Gene		GepheID
tyrosinase-related protein 1 (TYRP1) (<a +tyrosinase-related+protein+1+(tyrp1)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+tyrosinase-related+protein+1+(TYRP1)+"#gephebase-summary-title)	GP00002239	Main curator
Published	Entry Status	

Trait Category		Trait	
Morphology (#gephebase-summary-title)			
Coloration (coat) (#gephebase-summary-title)			
	Trait State in Taxon A		
darker fiber		Trait State in Taxon B	
lighter fiber in Valais Red			
	Ancestral State		
Taxon A			
	Taxonomic Status		
Domesticated (#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
<i>Ovis aries</i> (#gephebase-summary-title)		<i>Ovis aries</i> (#gephebase-summary-title)	
	Common Name		Common Name
sheep		sheep	
	Synonyms		Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758		Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	
	Rank		Rank
species		species	
	Lineage		Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis	
	Parent		Parent
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)		Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)		9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

Generic Gene Name	UniProtKB Mus musculus
Tyrp1	P07147 (http://www.uniprot.org/uniprot/P07147)
Synonyms	GenebankID or UniProtKB
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1	ABG76825 (https://www.ncbi.nlm.nih.gov/nuccore/ABG76825)
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>)

Yes (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes~#gephebase-summary-title>)

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title>)

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

Nonsense

c.1066C>T p.(Arg356*)

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

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	STP	356

Main Reference

Identification of two TYRP1 loss-of-function alleles in Valais Red sheep. (2019) (<https://pubmed.ncbi.nlm.nih.gov/31571241>)

Authors

Paris JM; Letko A; HÄßler IM; Ammann P; Flury C; DrÄ¶gemÄ¼ller C

Abstract

The Valais Red sheep breed is a local breed of the Swiss canton Valais. Although the breed is characterised by its brown colour, black animals occasionally occur and the objective of this study was to identify the causative genetic variants responsible for the obvious difference. A GWAS using high-density SNP data to compare 51 brown and 38 black sheep showed a strong signal on chromosome 2 at the TYRP1 locus. Haplotype analyses revealed three different brown-associated alleles. The WGS of three sheep revealed four protein-changing variants within the TYRP1 gene. Three of these variants were associated with the recessively inherited brown coat colour. This includes the known missense variant TYRP1:c.869G>T designated as b and two novel loss-of-function variants. We propose to designate the frame-shift variant TYRP1:c.86_87delGA as b and the nonsense variant TYRP1:c.1066C>T as b . Interestingly, the b allele occurs only in local breeds of Switzerland whereas the b allele seems to be more widespread across Europe.

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Additional References

RELATED GEPHE

Related Genes

2 (Agouti (ASIP), MC1R) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~9940~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

3 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~tyrosinase-related protein 1 \(TYRP1\)~/and+Taxon ID=~9940~/or+Gene Gephebase=~tyrosinase-related protein 1 \(TYRP1\)~/and+Taxon ID=~9940~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~tyrosinase-related protein 1 (TYRP1)~/and+Taxon ID=~9940~/or+Gene Gephebase=~tyrosinase-related protein 1 (TYRP1)~/and+Taxon ID=~9940~#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

@AllelicSeries <https://omia.org/OMIA001249/9940/>