

GEPHE SUMMARY

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)	GP00002238	Main curator
Published	Entry Status	Martin

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title)		Trait Category	
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat~#gephebase-summary-title)		Trait	
darker fiber	Trait State in Taxon A		
lighter fiber in Valais Red	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Ovis+aries~#gephebase-summary-title)	Latin Name	Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Ovis+aries~#gephebase-summary-title)	Latin Name
sheep	Common Name	sheep	Common Name
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	Synonyms	Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	Synonyms
species	Rank	species	Rank
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	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	Lineage
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)	Parent	Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)	Parent
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)	NCBI Taxonomy ID	9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

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 - Molecular Function		

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
Yes (#gephebase-summary-title)	
	Molecular Type
Coding (#gephebase-summary-title)	
	Aberration Type
Deletion (#gephebase-summary-title)	
	Deletion Size
1-9 bp	
	Molecular Details of the Mutation
c.86_87delGA p.(Glu29ValfsTer5)	
	Experimental Evidence
Linkage Mapping (#gephebase-summary-title)	
	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; Haffliger IM; Ammann P; Flury C; Dragmaller 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)	

EXTERNAL LINKS

COMMENTS

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

