

## GEPHE SUMMARY

	Gephebase Gene	GephelD
tyrosinase-related protein 1 (TYRP1) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^#gephebase-summary-title</a> )	GP00001156	Main curator
	Courtier	
	Entry Status	
Published		

## PHENOTYPIC CHANGE

	Trait Category
Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title</a> )	Trait
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#gephebase-summary-title</a> )	Trait State in Taxon A
dark	Trait State in Taxon B
light	Ancestral State
Taxon A	Taxonomic Status
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title</a> )	

Taxon A	Latin Name	Taxon B	Latin Name
Ovis aries ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title</a> )		Ovis aries ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title</a> )	
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) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935</a> )	Parent	Ovis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935</a> )	Parent
9940 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940</a> )	NCBI Taxonomy ID	9940 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940</a> )	NCBI Taxonomy ID
is Taxon A an Infraspecies?		is Taxon B an Infraspecies?	
No		No	

## GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Mus musculus
Tyrp1		P07147 ( <a href="http://www.uniprot.org/uniprot/P07147">http://www.uniprot.org/uniprot/P07147</a> )
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000006151 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151</a> )	String	ABG76825 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/ABG76825">https://www.ncbi.nlm.nih.gov/nuccore/ABG76825</a> )
Belongs to the tyrosinase family.	Sequence Similarities	
GO:0042803 : protein homodimerization activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042803">https://www.ebi.ac.uk/QuickGO/term/GO:0042803</a> )	GO - Molecular Function	
GO:0046982 : protein heterodimerization activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046982">https://www.ebi.ac.uk/QuickGO/term/GO:0046982</a> )		
GO:0046872 : metal ion binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046872">https://www.ebi.ac.uk/QuickGO/term/GO:0046872</a> )		

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=%22No%22#gephebase-summary-title>)

Molecular Type

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular+Type=%22Coding%22#gephebase-summary-title>)

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration+Type=%22SNP%22#gephebase-summary-title>)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c. 869G>T p.C290F ; Cys residue involved shows a high degree of evolutionary conservation; it is conserved not only across vertebrates but also in the two paralogues of TYRP1, DCT and TYR

Experimental Evidence

Linkage Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=%22Linkage+Mapping%22#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Cys	Phe	290

Main Reference

Compelling evidence that a single nucleotide substitution in TYRP1 is responsible for coat-colour polymorphism in a free-living population of Soay sheep. (2007)  
(<https://pubmed.ncbi.nlm.nih.gov/17254985/>)

Authors

Gratten J; Beraldi D; Lowder BV; McRae AF; Visscher PM; Pemberton JM; Slate J

Abstract

Identifying the genes that underlie phenotypic variation in natural populations is a central objective of evolutionary genetics. Here, we report the identification of the gene and causal mutation underlying coat colour variation in a free-living population of Soay sheep (*Ovis aries*). We targeted tyrosinase-related protein 1 (TYRP1), a positional candidate gene based on previous work that mapped the Coat colour locus to an approximately 15cM window on chromosome 2. We identified a non-synonymous substitution in exon IV that was perfectly associated with coat colour. This polymorphism is predicted to cause the loss of a cysteine residue that is highly evolutionarily conserved and likely to be of functional significance. We eliminated the possibility that this association is due to the presence of strong linkage disequilibrium with an unknown regulatory mutation by demonstrating that there is no difference in relative TYRP1 expression between colour morphs. Analysis of this putative causal mutation in a complex pedigree of more than 500 sheep revealed almost perfect co-segregation with coat colour (chi<sup>2</sup>-test, p<0.0001, LOD=110.20), and very tight linkage between Coat colour and TYRP1 (LOD=29.50).

Additional References

## RELATED GEPHE

Related Genes

2 (Agouti (ASIP), MC1R) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=%229940%22/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

3 ([https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=%22tyrosinase-related+protein+1+\(TYRP1\)%22/and+Taxon+ID=%229940%22/or+Gene+Gephebase=%22tyrosinase-related+protein+1+\(TYRP1\)%22/and+Taxon+ID=%229940%22#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=%22tyrosinase-related+protein+1+(TYRP1)%22/and+Taxon+ID=%229940%22/or+Gene+Gephebase=%22tyrosinase-related+protein+1+(TYRP1)%22/and+Taxon+ID=%229940%22#gephebase-summary-title))

## EXTERNAL LINKS

## COMMENTS

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