

## 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> )	GP00002237	Main curator
	Entry Status	Martin
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
darker Merino fiber	Trait State in Taxon B
lighter Awassi fiber	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> )	Common 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> )	Common Name
sheep	Synonyms	sheep	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	
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.2240C>G p.A746V

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	Ala	Val	746

Main Reference

QTL and association analysis for skin and fibre pigmentation in sheep provides evidence of a major causative mutation and epistatic effects. (2013)  
(<https://pubmed.ncbi.nlm.nih.gov/23451726>)

Authors

Raadsma HW; Jonas E; Fleet MR; Fullard K; Gongora J; Cavanagh CR; Tammen I; Thomson PC

Abstract

The pursuits of white features and white fleeces free of pigmented fibre have been important selection objectives for many sheep breeds. The cause and inheritance of non-white colour patterns in sheep has been studied since the early 19th century. Discovery of genetic causes, especially those which predispose pigmentation in white sheep, may lead to more accurate selection tools for improved apparel wool. This article describes an extended QTL study for 13 skin and fibre pigmentation traits in sheep. A total of 19 highly significant, 10 significant and seven suggestive QTL were identified in a QTL mapping experiment using an Awassi- Merino- Merino backcross sheep population. All QTL on chromosome 2 exceeded a LOD score of greater than 4 (range 4.4-30.1), giving very strong support for a major gene for pigmentation on this chromosome. Evidence of epistatic interactions was found for QTL for four traits on chromosomes 2 and 19. The ovine TYRP1 gene on OAR 2 was sequenced as a strong positional candidate gene. A highly significant association ( $P < 0.01$ ) of grandparental haplotypes across nine segregating SNP/microsatellite markers including one non-synonymous SNP with pigmentation traits could be shown. Up to 47% of the observed variation in pigmentation was accounted for by models using TYRP1 haplotypes and 83% for models with interactions between two QTL probabilities, offering scope for marker-assisted selection for these traits.

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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/>