GEPHE SUMMARY

Gephebase Gene
tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^#gephebase-summary-title)

GepheID
GP00002239

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^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 (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title)

Common Name
sheep

Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

Rank
species

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

Parent
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)

NCBI Taxonomy ID
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title)

Common Name
sheep

Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

Rank
species

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

Parent
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)

NCBI Taxonomy ID
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Tyrp1

Synonyms
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1

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)

UniProtKB Mus musculus
P07147 (http://www.uniprot.org/uniprot/P07147)

GenebankID or UniProtKB
ABG76825 (https://www.ncbi.nlm.nih.gov/nuccore/ABG76825)

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

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	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; HÃ¤fliger IM; Ammann P; Flury C; DrÃ¶gemÃ¼ller 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/