GEPHE SUMMARY

Gephebase Gene
tyrosinase-related protein 1 (TYRP1)

GephelD
GP00001156

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

Trait State in Taxon A
dark

Trait State in Taxon B
light

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Ovis aries

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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

Parent
Ovis () - (Rank: genus)

NCBI Taxonomy ID
9940

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Ovis aries

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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

Parent
Ovis () - (Rank: genus)

NCBI Taxonomy 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

Sequence Similarities
Belongs to the tyrosinase family.

GO - Molecular Function
GO:0042803 : protein homodimerization activity
GO:0046982 : protein heterodimerization activity
GO:0046872 : metal ion binding
GO:0004497 : monooxygenase activity

GO - Biological Process
GO:0032438 : melanosome organization
GO:0043473 : pigmentation
GO:0048023 : positive regulation of melanin biosynthetic process
GO:0006583 : melanin biosynthetic process from tyrosine

UniProtKB Mus musculus
P07147

GenebankID or UniProtKB
ABG76825

GO:0030318 : melanocyte differentiation
GO:0043438 : acetoacetic acid metabolic process
GO:0006582 : melanin metabolic process

GO - Cellular Component
GO:0016021 : integral component of membrane
GO:0030669 : clathrin-coated endocytic vesicle membrane
GO:0010008 : endosome membrane
GO:0042470 : melanosome
GO:0033162 : melanosome membrane

Presumptive Null
No

Molecular Type
Coding

Aberration Type
SNP

SNP Coding Change
Nonsynonymous

Molecular Details of the Mutation
C290F - G869T - The 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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

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)

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 (chi2-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)
Related Haplotypes
No matches found.

COMMENTS