

GEPHE SUMMARY

tyrosinase (TYR) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^tyrosinase+(TYR)^#gephebase-summary-title)	Gephebase Gene	GP00002375	GepheID
Published	Entry Status	Santos	Main curator

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		
WT - brown coat	Trait State in Taxon A		
White coat - dilution phenotype	Trait State in Taxon B		
Taxon A	Ancestral State		
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title)	Taxonomic Status		

Taxon A		Taxon B	
	Latin Name		Latin Name
Cervus elaphus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Cervus+elaphus^#gephebase-summary-title)	Cervus elaphus	Cervus elaphus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Cervus+elaphus^#gephebase-summary-title)	Cervus elaphus
red deer	Common Name	red deer	Common Name
red deer; Cervus elaphus Linnaeus 1758; Cervus elaphus Linnaeus, 1758	Synonyms	red deer; Cervus elaphus Linnaeus 1758; Cervus elaphus 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; Cervidae; Cervinae; Cervus	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; Cervidae; Cervinae; Cervus	Lineage
Cervus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9859)	Parent	Cervus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9859)	Parent
9860 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9860)	NCBI Taxonomy ID	9860 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9860)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

Tyr	Generic Gene Name	P11344 (http://www.uniprot.org/uniprot/P11344)	UniProtKB Mus musculus
c; Oca1; skc35; albino	Synonyms		GenebankID or UniProtKB
10090.ENSMUSP00000004770 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770)	String		
Belongs to the tyrosinase family.	Sequence Similarities		
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)	GO - Molecular Function		
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)			
GO:0005507 : copper ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005507)			
GO:0004503 : monophenol monooxygenase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004503)			

- GO:0042438 : melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
- GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
- GO:0008283 : cell proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0008283>)
- GO:0033280 : response to vitamin D (<https://www.ebi.ac.uk/QuickGO/term/GO:0033280>)
- GO:0051591 : response to cAMP (<https://www.ebi.ac.uk/QuickGO/term/GO:0051591>)
- GO:0009411 : response to UV (<https://www.ebi.ac.uk/QuickGO/term/GO:0009411>)
- GO:0048538 : thymus development (<https://www.ebi.ac.uk/QuickGO/term/GO:0048538>)

GO - Cellular Component

- GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
- GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
- GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
- GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
- GO:0043231 : intracellular membrane-bounded organelle
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043231>)
- GO:0048471 : perinuclear region of cytoplasm
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048471>)
- 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="^No^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type="^Coding^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=))

Aberration Type

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type="^SNP^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

"a non-synonymous mutation with exchange of a glycine residue at position 291 of the tyrosinase protein by arginine was identified as the cause of dilution of the coat colour"

The mutation is on nucleotide 871 (Gâ€‰%>â€‰%A) but the exact codons are not specified in the paper.

Experimental Evidence

Candidate Gene ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence="^Candidate Gene^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=))

	Taxon A	Taxon B	Position
Codon	-	-	871
Amino-acid	Gly	Arg	291

Main Reference

A genome-wide scan study identifies a single nucleotide substitution in the tyrosinase gene associated with white coat colour in a red deer (*Cervus elaphus*) population. (2020)
(<https://pubmed.ncbi.nlm.nih.gov/32041521>)

Authors

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Abstract

Red deer with very pale coat colour are observed sporadically. In the red deer (*Cervus elaphus*) population of Reinhardswald in Germany, about 5% of animals have a white coat colour that is not associated with albinism. In order to facilitate the conservation of the animals, it should be determined whether and to what extent brown animals carry the white gene. For this purpose, samples of one white hind and her brown calf were available for whole genome sequencing to identify the single nucleotide polymorphism(s) responsible for the white phenotype. Subsequently, samples from 194 brown and 11 white animals were genotyped.

Based on a list of colour genes of the International Federation of Pigment Cell Societies, a non-synonymous mutation with exchange of a glycine residue at position 291 of the tyrosinase protein by arginine was identified as the cause of dilution of the coat colour. A gene test led to exactly matching genotypes in all examined animals. The study showed that 14% of the brown animals carry the white gene. This provides a simple and reliable way of conservation for the white animals. However, results could not be transferred to another, unrelated red deer population with white animals. Although no brown animals with a white tyrosinase genotype were detected, the cause for the white colouring in this population was different.

A gene test for the conservation of white red deer is available for the population of the Reinhardswald. While mutations in the tyrosinase are commonly associated with oculocutaneous albinism type 1, the amino acid exchange at position 291 was found to be associated with coat colour dilution in *Cervus elaphus*.

Additional References

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS