

GEPHE SUMMARY

tyrosinase (TYR) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^tyrosinase+(TYR)^#gephebase-summary-title)	Gephebase Gene	GP00002319	GepheID
Published	Entry Status	Martin	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	Trait State in Taxon A
Complete albinism	Trait State in Taxon B
Taxon A	Ancestral State
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated^#gephebase-summary-title)	Taxonomic Status

Taxon A	Latin Name	Taxon B	Latin Name
Neovison vison (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Neovison+vison^#gephebase-summary-title)	Neovison vison (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Neovison+vison^#gephebase-summary-title)	Neovison vison (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Neovison+vison^#gephebase-summary-title)	Neovison vison (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Neovison+vison^#gephebase-summary-title)
American mink	American mink	American mink	American mink
Mustela vison; American mink; mink; Mustela vison species	Mustela vison; American mink; mink; Mustela vison species	Mustela vison; American mink; mink; Mustela vison species	Mustela vison; American mink; mink; Mustela vison species
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Mustelidae; Mustelinae; Neovison	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Mustelidae; Mustelinae; Neovison	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Mustelidae; Mustelinae; Neovison	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Mustelidae; Mustelinae; Neovison
Neovison () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452645)	Neovison () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452645)	Neovison () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452645)	Neovison () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452645)
452646 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452646)	452646 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452646)	452646 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452646)	452646 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=452646)
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	0	
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)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null="No"#gephebase-summary-title) Presumptive Null
 Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type="Coding"#gephebase-summary-title) Molecular Type
 SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type="SNP"#gephebase-summary-title) Aberration Type
 Nonsense SNP Coding Change
 c.138T>A p.C46* Molecular Details of the Mutation
 Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence="Candidate Gene"#gephebase-summary-title) Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Cys	STP	46

Albinism in the American mink (*Neovison vison*) is associated with a tyrosinase nonsense mutation. (2008) (https://pubmed.ncbi.nlm.nih.gov/18822100) Main Reference
 Anistoroaei R; Fredholm M; Christensen K; Leeb T Authors
 Albino phenotypes are documented in various species including the American mink. In other species the albino phenotypes are associated with tyrosinase (TYR) gene mutations; therefore TYR was considered the candidate gene for albinism in mink. Four microsatellite markers were chosen in the predicted region of the TYR gene. Genotypes at the markers Mvi6025 and Mvi6034 were found to be associated with the albino phenotype within an extended half-sib family. A BAC clone containing Mvi6034 was mapped to chromosome 7q1.1-q1.3 by fluorescent in situ hybridization. Subsequent analysis of genomic TYR sequences from wild-type and albino mink samples identified a nonsense mutation in exon 1, which converts a TGT codon encoding cysteine to a TGA stop codon (c.138T>A, p.C46X; EU627590). The mutation truncates more than 90% of the normal gene product including the putative catalytic domains. The results indicate that the nonsense mutation is responsible for the albino phenotype in the American mink. Abstract

Additional References

RELATED GEPHE

2 (Melanophilin (MLPH), tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID="452646"/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title) Related Genes
 1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase="tyrosinase (TYR)"/and+Taxon ID="452646"/or+Gene Gephebase="tyrosinase (TYR)"/and+Taxon ID="452646"#gephebase-summary-title) Related Haplotypes

EXTERNAL LINKS

COMMENTS

