

GEPHE SUMMARY

tyrosinase (TYR) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase= [^] tyrosinase (TYR) [^] #gephebase-summary-title)	Gephebase Gene	GP00002310	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) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=<sup>^</sup>Coloration">https://www.gephebase.org/search-criteria?/and+Trait=[^]Coloration (coat) [^] #gephebase-summary-title)	Trait		
WT	Trait State in Taxon A		
Albino coat from various breeds sampled in Germany	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		Taxon B
	Latin Name		Latin Name
Mustela putorius furo (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=<sup>^</sup>Mustela+putorius+furo<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=[^]Mustela+putorius+furo[^]#gephebase-summary-title)	Common Name	Mustela putorius furo (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=<sup>^</sup>Mustela+putorius+furo<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=[^]Mustela+putorius+furo[^]#gephebase-summary-title)	Common Name
domestic ferret	Synonyms	domestic ferret	Synonyms
Mustela furo; domestic ferret; black ferret; ferret; Mustela putorius furo Linnaeus, 1758; Mustela putorius furo	Rank	Mustela furo; domestic ferret; black ferret; ferret; Mustela putorius furo Linnaeus, 1758; Mustela putorius furo	Rank
subspecies	Lineage	subspecies	Lineage
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; Mustela; Mustela putorius	Parent	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; Mustela; Mustela putorius	Parent
Mustela putorius (European polecat) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9668)	NCBI Taxonomy ID	Mustela putorius (European polecat) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9668)	NCBI Taxonomy ID
9669 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9669)	is Taxon A an Intraspecies?	9669 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9669)	is Taxon B an Intraspecies?
No		Yes	Taxon B Description
		-	

GENOTYPIC CHANGE

Tyr	Generic Gene Name	P11344 (http://www.uniprot.org/uniprot/P11344)	UniProtKB Mus musculus
c; Oca1; skc35; albino	Synonyms	0	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 - Biological Process

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=~No~#gephebase-summary-title))

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=~Coding~#gephebase-summary-title))

Aberration Type

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

Deletion Size

-

Molecular Details of the Mutation

Deletion of exon 4 as detected by Southern Blot

Experimental Evidence

Linkage Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Linkage+Mapping~#gephebase-summary-title))

Main Reference

Identification of a tyrosinase (TYR) exon 4 deletion in albino ferrets (*Mustela putorius furo*). (2007) (<https://pubmed.ncbi.nlm.nih.gov/17655555>)

Authors

Blaszczuk WM; Distler C; Dekomien G; Arning L; Hoffmann KP; Epplen JT

Abstract

Albinism is due to a lack of pigmentation in hair, skin and eye, and has been shown to occur in several animal species. Mutations of the tyrosinase (TYR) gene account for albinism in domestic cats, rabbits, cattle, mice and rats. In this study, we demonstrate that a TYR mutation accounts for albinism in the ferret (*Mustela putorius furo*). The coding sequence of the five exons of TYR was determined in genomic DNA from wild-type pigmented 'sable' coloured and albino ferrets. It was not possible to amplify TYR exon 4 in albino ferrets originating from different breeds.

The deletion of exon 4 in albino ferrets was confirmed by Southern blot hybridization of genomic DNA from albino and pigmented ferrets. This is the first report of a deletion of a TYR exon in a non-human mammal.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA000202/9669/> @Parallelism

