GEPHE SUMMARY

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

GepheID
GP00002310

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
WT

Trait State in Taxon B
Albino coat from various breeds sampled in Germany

Ancestral State
Taxon A

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

Taxon A

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

Common Name
domestic ferret

Synonyms
Mustela furo; domestic ferret; black ferret; ferret; Mustela putorius furo Linnaeus, 1758; Mustela putoris furo

Rank
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
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 Infraspecies?
No

Taxon B

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

Common Name
domestic ferret

Synonyms
Mustela furo; domestic ferret; black ferret; ferret; Mustela putorius furo Linnaeus, 1758; Mustela putoris furo

Rank
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
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 B an Infraspecies?
Yes

Taxon B Description
-

GENOTYPIC CHANGE

Generic Gene Name
Tyr

Synonyms
c; Oca1; skc35; albino

String
10090.ENSMUSP00000004770 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770)

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)

UniProtKB Mus musculus
P11344 (http://www.uniprot.org/uniprot/P11344)

GenebankID or UniProtKB
()

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)

Molecular Type

Coding (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)

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)

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

Blaszczyk 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