

GEPHE SUMMARY

tyrosinase (TYR) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase= [^] tyrosinase (TYR) [^] #gephebase-summary-title)	Gephebase Gene	GP00002374	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; albinism) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=<sup>^</sup>Coloration (coat; albinism)<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=[^]Coloration (coat; albinism)[^]#gephebase-summary-title)	Trait		
wildtype coat	Trait State in Taxon A		
white coat	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	
Nyctereutes procyonoides viverrinus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Nyctereutes procyonoides viverrinus<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Nyctereutes procyonoides viverrinus[^]#gephebase-summary-title)	Latin Name	Nyctereutes procyonoides viverrinus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Nyctereutes procyonoides viverrinus<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Nyctereutes procyonoides viverrinus[^]#gephebase-summary-title)	Latin Name
-	Common Name	-	Common Name
Nyctereutes procyonoides subsp. viverrinus	Synonyms	Nyctereutes procyonoides subsp. viverrinus	Synonyms
subspecies	Rank	subspecies	Rank
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; Canidae; Nyctereutes; Nyctereutes procyonoides	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; Canidae; Nyctereutes; Nyctereutes procyonoides	Lineage
Nyctereutes procyonoides (raccoon dog) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 34880)	Parent	Nyctereutes procyonoides (raccoon dog) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 34880)	Parent
476259 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 476259)	NCBI Taxonomy ID	476259 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 476259)	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	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: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

Yes ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes))

Molecular Type

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

Aberration Type

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

Deletion Size

10-100 kb

Molecular Details of the Mutation

“We examined the structure and nucleotide sequence of TYR in an albino tanuki and found that the third exon was removed due to a deletion of approximately 11 kb. In addition, two nonsynonymous nucleotide substitutions were found in the fifth exon. These mutations are possible causes of the albino phenotype; however, the order of occurrence is unclear. Even if the 11-kb deletion was not the first of these mutations, it is considered to cause a total loss of the tyrosinase function because the third exon carries codons for one of the two copper-binding sites of tyrosinase and these sites are essential for the enzyme function. Intriguingly, the deletion was not a simple removal of an 11-kb segment: an internal portion was retained as a segment in the reverse orientation. We propose possible formation processes for this mutation that involve multiple DNA scission events, or an inversion followed by a deletion.”

Experimental Evidence

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

Main Reference

Complex intragene deletion leads to oculocutaneous albinism in tanuki (Japanese raccoon dog). (2020) (<https://pubmed.ncbi.nlm.nih.gov/32783776>)

Authors

Mae Y; Nagara K; Miyazaki M; Katsura Y; Enomoto Y; Koga A

Abstract

Tanuki (*Nyctereutes procyonoides viverrinus*), or Japanese raccoon dog, is a canine native to Japan. Tanuki with complete oculocutaneous albinism are relatively frequent in mountainous areas of mainland Japan. Tyrosinase, which is encoded by the TYR gene, is an enzyme essential for the biosynthesis of melanin pigment. We examined the structure and nucleotide sequence of TYR in an albino tanuki and found that the third exon was removed due to a deletion of approximately 11 kb. In addition, two nonsynonymous nucleotide substitutions were found in the fifth exon. These mutations are possible causes of the albino phenotype; however, the order of occurrence is unclear. Even if the 11-kb deletion was not the first of these mutations, it is considered to cause a total loss of the tyrosinase function because the third exon carries codons for one of the two copper-binding sites of tyrosinase and these sites are essential for the enzyme function. Intriguingly, the deletion was not a simple removal of an 11-kb segment: an internal portion was retained as a segment in the reverse orientation. We propose possible formation processes for this mutation that involve multiple DNA scission events, or an inversion followed by a deletion.

Additional References

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

