

## GEPHE SUMMARY

tyrosinase (TYR) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase="tyrosinase (TYR)"#gephebase-summary-title)	Gephebase Gene	GP00002314	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category="Morphology"#gephebase-summary-title)	Trait Category		
Coloration (skin) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> ="Coloration (skin)"#gephebase-summary-title)	Trait		
WT	Trait State in Taxon A		
White whale (single wild individual)	Trait State in Taxon B		
Taxon A	Ancestral State		
Intraspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status="Intraspecific"#gephebase-summary-title)	Taxonomic Status		

Taxon A	Latin Name	Taxon B	Latin Name
Megaptera novaeangliae ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms="Megaptera novaeangliae"#gephebase-summary-title)		Megaptera novaeangliae ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms="Megaptera novaeangliae"#gephebase-summary-title)	
humpback whale	Common Name	humpback whale	Common Name
humpback whale; Megaptera novaeangliae	Synonyms	humpback whale; Megaptera novaeangliae	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; Cetartiodactyla; Cetacea; Mysticeti; Balaenopteridae; Megaptera	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Cetacea; Mysticeti; Balaenopteridae; Megaptera	Lineage
Megaptera () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9772">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9772</a> )	Parent	Megaptera () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9772">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9772</a> )	Parent
9773 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9773">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9773</a> )	NCBI Taxonomy ID	9773 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9773">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9773</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

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

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

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

1-9 bp Deletion Size

(264 del C) at codon 88 Molecular Details of the Mutation

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

Variation in the tyrosinase gene associated with a white humpback whale (*Megaptera novaeangliae*). (2012 Jan-Feb) (https://pubmed.ncbi.nlm.nih.gov/22140253) Main Reference

Polanowski AM; Robinson-Laverick SM; Paton D; Jarman SN Authors

Tyrosinase-negative oculocutaneous albinism (OCA1A) is characterized by lifelong white hair and skin, a phenotype that has been described in most mammalian species worldwide. Tyrosinase is the key enzyme in melanin biosynthesis, and mutations in the tyrosinase gene result in OCA1A. We examined sequence variation at exon 1 of the tyrosinase gene in 66 humpback whale samples collected from the east coast of Australia, including an anomalously white humpback whale known as "Migaloo." We identified 3 novel variants, including a cytosine deletion that results in a premature stop codon in exon 1. The deletion truncates the tyrosinase protein including the putative catalytic domains that are essential for tyrosinase enzymatic activity. Migaloo was homozygous for this deletion, suggesting that the albino phenotype is a consequence of inactive tyrosinase caused by the frameshift in the tyrosinase gene. Abstract

Additional References

RELATED GEPHE

No matches found. Related Genes

No matches found. Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Parallelism https://omia.org/OMIA000202/9773/