

GEPHE SUMMARY

tyrosinase (TYR) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^tyrosinase+(TYR)^#gephebase-summary-title)	Gephebase Gene	GP00002316	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
Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Oryctolagus+cuniculus^#gephebase-summary-title)	Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Oryctolagus+cuniculus^#gephebase-summary-title)	Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Oryctolagus+cuniculus^#gephebase-summary-title)	Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Oryctolagus+cuniculus^#gephebase-summary-title)
rabbit	Common Name	rabbit	Common Name
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	Synonyms	Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	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; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagus	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagus	Lineage
Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984)	Parent	Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984)	Parent
9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986)	NCBI Taxonomy ID	9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986)	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		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 - 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>)

- 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
- Nonsynonymous SNP Coding Change
- c.1118C>A p.T373K (also causes albinism in humans) 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	Tyr	Lys	373

- Tyrosinase gene variants in different rabbit strains. (2000) (<https://pubmed.ncbi.nlm.nih.gov/10920244>) Main Reference
- Aigner B; Besenfelder U; MÄ¼ller M; Brem G Authors
- Abstract
- Additional References

RELATED GEPHE

- 4 (Agouti (ASIP), MC1R, Melanophilin (MLPH), tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9986^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>) Related Genes
- No matches found. Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Parallelism <https://omia.org/OMIA000202/9986/>

