

GEPHE SUMMARY

	Gephebase Gene	GephelD
tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria/?and+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^#gephebase-summary-title)	GP00001327	
	Entry Status	Main curator
Published	Prigent	

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		
Liangshan pig-black coat	Trait State in Taxon B		
Liangshan pig-blond coat	Ancestral State		
Taxon A	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria/?and+Taxonomic Status=^Domesticated^#gephebase-summary-title)			
Taxon A		Taxon B	
Sus scrofa	Latin Name	Sus scrofa domesticus	Latin Name
(https://www.gephebase.org/search-criteria/?and+Taxon and Synonyms=^Sus scrofa^#gephebase-summary-title)		(https://www.gephebase.org/search-criteria/?and+Taxon and Synonyms=^Sus scrofa domesticus^#gephebase-summary-title)	
pig	Common Name	domestic pig	Common Name
pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofus	Synonyms	Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig	Synonyms
species	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; Cetartiodactyla; Suina; Suidae; Sus	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; Suina; Suidae; Sus; Sus scrofa	Lineage
Sus () - (Rank: genus)	Parent	Sus scrofa (pig) - (Rank: species)	Parent
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9822)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)	
9823	NCBI Taxonomy ID	9825	NCBI Taxonomy ID
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9825)	
Yes	is Taxon A an Infraspecies?	Yes	is Taxon B an Infraspecies?
Liangshan pig-black coat	Taxon A Description	Liangshan pig-blond coat	Taxon B Description

GENOTYPIC CHANGE

	Generic Gene Name		
Tyrp1			UniProtKB Mus musculus
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1	Synonyms	P07147 (http://www.uniprot.org/uniprot/P07147)	GenebankID or UniProtKB
10090.ENSMUSP00000006151 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)	String	KT581974 (https://www.ncbi.nlm.nih.gov/nuccore/KT581974)	
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:0046872 : metal ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0046872>)

GO:0004497 : monooxygenase activity

(<https://www.ebi.ac.uk/QuickGO/term/GO:0004497>)

GO - Biological Process

GO:0032438 : melanosome organization

(<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)

GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)

GO:0048023 : positive regulation of melanin biosynthetic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)

GO:0006583 : melanin biosynthetic process from tyrosine

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006583>)

GO:0030318 : melanocyte differentiation

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)

GO:0043438 : acetoacetic acid metabolic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0043438>)

GO:0006582 : melanin metabolic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006582>)

GO - Cellular Component

GO:0016021 : integral component of membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

GO:0030669 : clathrin-coated endocytic vesicle membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030669>)

GO:0010008 : endosome membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0010008>)

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 ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^No))

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

1-9 bp

Molecular Details of the Mutation

g.17599_17604del 6bp deletion in exon 8 resulting in deletion of Met and Gly residues at positions 495 and 496 in TYRP1 protein ; c.1484_1489del6 p.Met495_Gly496del

Experimental Evidence

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

Main Reference

A 6-bp deletion in exon 8 and two mutations in introns of TYRP1 are associated with blond coat color in Liangshan pigs. (2016) (<https://pubmed.ncbi.nlm.nih.gov/26680103/>)

Authors

Wu X; Zhang Y; Shen L; Du J; Luo J; Liu C; Pu Q; Yang R; Li X; Bai L; Tang G; Zhang S; Zhu L

Abstract

Melanocortin receptor 1 (MC1R), Agouti signaling protein (ASIP), and Tyrosinase-related protein 1 (TYRP1) are reported critical genes that regulate pheomelanin and eumelanin synthesis in mammals. Liangshan pig is a special Chinese indigenous pig breed with two completely different coat colors, solid black and blond. In this study, we detected polymorphisms of the above three genes and assessed the relationships between the variations and coat color phenotypes in Liangshan pigs. The findings revealed that the blond phenotype of Liangshan pig was related to dominant mutations in TYRP1, but not related to mutations in MC1R or ASIP. We found three closely linked mutations in TYRP1, g.8406G>A in intron 4, g.11100A>G in intron 5, and g.17599_17604del in exon 8, that were completely associated with blond coat color in Liangshan pigs. Further analysis revealed that a 6-bp deletion mutation resulted in deletion of Met and Gly residues at positions 495 and 496 in TYRP1 protein, and altered the structure of transmembrane domain of TYRP1. Together, our findings indicated that these three mutations in TYRP1 cause the blond phenotype in Liangshan pigs.

Copyright © 2015 Elsevier B.V. All rights reserved.

Additional References

RELATED GEPHE

Related Genes

3 (Agouti, Kit (type III receptor protein-tyrosine kinase), MC1R) ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=^9823^/and+Trait=Coloration/or+Taxon+ID=^9825^/and+Trait=Coloration/and+groupHaplotypes=true))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

the deletion is associated in the same haplotype with 2 substitutions g.8406G>A and g.11100A>G in introns 4-5 ; <https://omia.org/OMIA001249/9823/>

