GEPHE SUMMARY

Gephebase Gene
tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^#gephebase-summary-title)

GephebaseID
GP00001327

Entry Status
Published

Main curator
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

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

Common Name
pig

Synonyms
pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofus

Rank
species

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

Parent
Sus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9822)

NCBI Taxonomy ID
9823 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)

is Taxon A an Infraspecies?
Yes

Taxon A Description
Liangshan pig-black coat

Taxon B

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

Common Name
domestic pig

Synonyms
Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig

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; Cetartiodactyla; Suina; Suidae; Sus; Sus scrofa

Parent
Sus scrofa (pig) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)

NCBI Taxonomy ID
9825 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9825)

is Taxon B an Infraspecies?
Yes

Taxon B Description
Liangshan pig-blond coat

GENOTYPIC CHANGE

Generic Gene Name
Tyrp1

Synonyms
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1

String
10090.ENSMUSP00000006151 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)

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
P07147 (http://www.uniprot.org/uniprot/P07147)

GenebankID or UniProtKB
KT581974 (https://www.ncbi.nlm.nih.gov/nuccore/KT581974)

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 (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

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

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.

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Additional References

RELATED GEPHE

Related Genes

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

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/