

## GEPHE SUMMARY

**Gephebase Gene**  
tyrosinase-related protein 1 (TYRP1)

**Entry Status**  
Published

**GepheID**  
GP00001327

**Main curator**  
Prigent

## PHENOTYPIC CHANGE

**Trait Category**  
Morphology

**Trait**  
Coloration (coat)

**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

### Taxon A

**Latin Name**  
*Sus scrofa*

**Common Name**  
pig

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

**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)

**NCBI Taxonomy ID**  
9823

**is Taxon A an Intraspecies?**  
Yes

**Taxon A Description**  
Liangshan pig-black coat

### Taxon B

**Latin Name**  
*Sus scrofa domesticus*

**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)

**NCBI Taxonomy ID**  
9825

**is Taxon B an Intraspecies?**  
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

**Sequence Similarities**  
Belongs to the tyrosinase family.

**GO - Molecular Function**  
GO:0042803 : protein homodimerization activity  
GO:0046982 : protein heterodimerization activity  
GO:0046872 : metal ion binding  
GO:0004497 : monooxygenase activity

**GO - Biological Process**  
GO:0032438 : melanosome organization  
GO:0043473 : pigmentation  
GO:0048023 : positive regulation of melanin biosynthetic process

**UniProtKB** *Mus musculus*  
P07147

**GenebankID or UniProtKB**  
KT581974

GO:0006583 : melanin biosynthetic process from tyrosine

GO:0030318 : melanocyte differentiation

GO:0043438 : acetoacetic acid metabolic process

GO:0006582 : melanin metabolic process

#### GO - Cellular Component

GO:0016021 : integral component of membrane

GO:0030669 : clathrin-coated endocytic vesicle membrane

GO:0010008 : endosome membrane

GO:0042470 : melanosome

GO:0033162 : melanosome membrane

#### Presumptive Null

No

#### Molecular Type

Coding

#### 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

#### 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)

##### 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)

##### 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