

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000631	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)		Trait Category	
Coloration (skin) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(skin)^#gephebase-summary-title)		Trait	
Sus scrofa		Trait State in Taxon A	
Sus scrofa		Trait State in Taxon B	
Taxon A		Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Sus scrofa		Sus scrofa domesticus	
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Sus scrofa scrofa^#gephebase-summary-title)		(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Sus scrofa domesticus^#gephebase-summary-title)	
pig		domestic pig	
pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofus		Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig	
species		subspecies	
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		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	
Sus () - (Rank: genus)		Sus scrofa (pig) - (Rank: species)	
(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		9825	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9825)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
	Synonyms	ALU33721 (https://www.ncbi.nlm.nih.gov/nuccore/ALU33721)	GenebankID or UniProtKB
	String		
9606.ENSPP00000451605			
(http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)			
Belongs to the G-protein coupled receptor 1 family.		Sequence Similarities	
GO:0008528 : G protein-coupled peptide receptor activity		GO - Molecular Function	
(https://www.ebi.ac.uk/QuickGO/term/GO:0008528)			
GO:0004977 : melanocortin receptor activity			
(https://www.ebi.ac.uk/QuickGO/term/GO:0004977)			
GO:0004980 : melanocyte-stimulating hormone receptor activity			
(https://www.ebi.ac.uk/QuickGO/term/GO:0004980)			
GO:0031625 : ubiquitin protein ligase binding			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0031625>)

GO - Biological Process

GO:0007275 : multicellular organism development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007275>)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
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:0007186 : G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007186>)
GO:0051897 : positive regulation of protein kinase B signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051897>)
GO:0019233 : sensory perception of pain
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019233>)
GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007189>)
GO:0035556 : intracellular signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035556>)
GO:0007187 : G protein-coupled receptor signaling pathway, coupled to cyclic nucleotide second messenger (<https://www.ebi.ac.uk/QuickGO/term/GO:0007187>)
GO:0032720 : negative regulation of tumor necrosis factor production
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032720>)
GO:0010739 : positive regulation of protein kinase A signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010739>)
GO:0090037 : positive regulation of protein kinase C signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090037>)
GO:0009650 : UV protection (<https://www.ebi.ac.uk/QuickGO/term/GO:0009650>)
GO:0070914 : UV-damage excision repair
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070914>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005887 : integral component of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)

Yes (#gephebase-summary-title)	Presumptive Null
Coding (#gephebase-summary-title)	Molecular Type
Insertion (#gephebase-summary-title)	Aberration Type
1-9 bp	Insertion Size
2bp insertion at 23 generates early stop @ 56 & D"121"N ?	Molecular Details of the Mutation
Linkage Mapping (#gephebase-summary-title)	Experimental Evidence
A frameshift mutation in MC1R and a high frequency of somatic reversions cause black spotting in pigs. (2001) (https://pubmed.ncbi.nlm.nih.gov/11404341)	Main Reference
Kijas JM; Moller M; Plastow G; Andersson L	Authors
Black spotting on a red or white background in pigs is determined by the E(P) allele at the MC1R/Extension locus. A previous comparison of partial MC1R sequences revealed that E(P) shares a missense mutation (D121N) with the E(D2) allele for dominant black color. Sequence analysis of the entire coding region now reveals a second mutation in the form of a 2-bp insertion at codon 23 (nt67insCC). This mutation expands a tract of six C nucleotides to eight and introduces a premature stop codon at position 56. This frameshift mutation is expected to cause a recessive red color, which was in fact observed in some breeds with the E(P) allele present (Tamworth and Hereford). RT-PCR analyses were conducted using skin samples taken from both spotted and background areas of spotted pigs. The background red area had transcript only from the mutant nt67insCC MC1R allele, whereas the black spot also contained a transcript without the 2-bp insertion. This indicates that black spots are due to somatic reversion events that restore the frame and MC1R function. The phenotypic expression of the E(P) allele is highly variable and the associated coat color ranges from red, red with black spots, white with black spots, to almost completely solid black. In several breeds of pigs the phenotypic manifestation of this allele has been modified by selection for or against black spots.	Abstract
Detection of genetic diversity and selection at the coding region of the melanocortin receptor 1 (MC1R) gene in Tibetan pigs and Landrace pigs. (2016) (https://pubmed.ncbi.nlm.nih.gov/26431999)	Additional References

RELATED GEPHE

3 (Agouti, Kit (type III receptor protein-tyrosine kinase), tyrosinase-related protein 1 (TYRP1)) (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 Genes
6 (#gephebase-summary-title)	Related Haplotypes

EXTERNAL LINKS

