

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000634	GepheID
Published	Entry Status	Martin	Main curator

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		
Data not curated	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=^Domesticated^#gephebase-summary-title)	Taxonomic Status		

Taxon A	Latin Name	Taxon B	Latin Name
Sus scrofa (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sus+scrofa^#gephebase-summary-title)		Sus scrofa domesticus (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 scrofa species	Synonyms	Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig	Synonyms
	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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9822)	Parent	Sus scrofa (pig) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9825)	Parent
9823 (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)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	ALU33721 (https://www.ncbi.nlm.nih.gov/nucore/ALU33721)	GenebankID or UniProtKB
9606.ENSPP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)	String		
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities		
GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)	GO - Molecular Function		
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>)

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

Nonsynonymous SNP Coding Change

A¹⁶¹V actually 164 Molecular Details of the Mutation

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Melanocortin receptor 1 (MC1R) mutations and coat color in pigs. (1998) (<https://pubmed.ncbi.nlm.nih.gov/9799269>) Main Reference

Kijas JM; Wales R; Trmsten A; Chardon P; Moller M; Andersson L Authors

The melanocortin receptor 1 (MC1R) plays a central role in regulation of eumelanin (black/brown) and phaeomelanin (red/yellow) synthesis within the mammalian melanocyte and is encoded by the classical Extension (E) coat color locus. Sequence analysis of MC1R from seven porcine breeds revealed a total of four allelic variants corresponding to five different E alleles. The European wild boar possessed a unique MC1R allele that we believe is required for the expression of a wild-type coat color. Two different MC1R alleles were associated with the dominant black color in pigs. MC1R^{*2} was found in European Large Black and Chinese Meishan pigs and exhibited two missense mutations compared with the wild-type sequence. Comparative data strongly suggest that one of these, L99P, may form a constitutively active receptor. MC1R^{*3} was associated with the black color in the Hampshire breed and involved a single missense mutation D121N. This same MC1R variant was also associated with EP, which results in black spots on a white or red background. Two different missense mutations were identified in recessive red (e/e) animals. One of these, A240T, occurs at a highly conserved position, making it a strong candidate for disruption of receptor function. 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](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

EXTERNAL LINKS

COMMENTS