

GEPHE SUMMARY

	Gephebase Gene		GepheID
MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)		GP00000633	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)			
	Trait		
Coloration (skin) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(skin)^#gephebase-summary-title)			
	Trait State in Taxon A		
Sus scrofa			
	Trait State in Taxon B		
Sus scrofa			
	Ancestral State		
Data not curated			
	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated^#gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		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)	
	Common Name		Common Name
pig		domestic pig	
	Synonyms		Synonyms
pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofus		Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig	
	Rank		Rank
species		subspecies	
	Lineage		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		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		Parent
Sus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9822)		Sus scrofa (pig) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9823 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823)		9825 (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

	Generic Gene Name		UniProtKB Homo sapiens
MC1R		Q01726 (http://www.uniprot.org/uniprot/Q01726)	
	Synonyms		GenebankID or UniProtKB
CMM5; MSH-R; SHEP2; MSHR		ALU33721 (https://www.ncbi.nlm.nih.gov/nuccore/ALU33721)	
	String		
9606.ENSP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000451605)			
	Sequence Similarities		
Belongs to the G-protein coupled receptor 1 family.			
	GO - Molecular Function		
GO:0008528 : G protein-coupled peptide receptor activity (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>)

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

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title>)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

D¹²¹N Actually 124

Experimental Evidence

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

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

Main Reference

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

Authors

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

Abstract

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.

Additional References

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>)

RELATED GEPHE

Related Genes

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>)

EXTERNAL LINKS

COMMENTS