

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^MC1R^#gephebase-summary-title)	Gephebase Gene	GP00001337	GepheID
Published	Entry Status	Prigent	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)	Trait Category		
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+coat^#gephebase-summary-title)	Trait		
Hawaiian feral pig	Trait State in Taxon A		
Hawaiian feral pig-black	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Sus scrofa (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sus+scrofa^#gephebase-summary-title)	Latin Name	Sus scrofa domesticus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sus+scrofa+domesticus^#gephebase-summary-title)	Latin Name
pig	Common Name	domestic pig	Common Name
pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofaus	Synonyms	Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig	Synonyms
species	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
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
Yes		Yes	
Hawaiian feral pig	Taxon A Description	Hawaiian feral pig-black	Taxon B Description

GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	0	GenebankID or UniProtKB
9606.ENSP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000451605)	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>)

Presumptive Null

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))

Molecular Type

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))

Aberration 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))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.G>A p.Asp124Asn

Experimental Evidence

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))

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

Main Reference

Strong signatures of selection in the domestic pig genome. (2012) (<https://pubmed.ncbi.nlm.nih.gov/23151514>)

Authors

Rubin CJ; Megens HJ; Martinez Barrio A; Maqbool K; Sayyab S; Schwochow D; Wang C; Carlborg Å-; Jern P; JÄrgensen CB; Archibald AL; Fredholm M; Groenen MA; Andersson L

Abstract

Domestication of wild boar (*Sus scrofa*) and subsequent selection have resulted in dramatic phenotypic changes in domestic pigs for a number of traits, including behavior, body composition, reproduction, and coat color. Here we have used whole-genome resequencing to reveal some of the loci that underlie phenotypic evolution in European domestic pigs. Selective sweep analyses revealed strong signatures of selection at three loci harboring quantitative trait loci that explain a considerable part of one of the most characteristic morphological changes in the domestic pig—the elongation of the back and an increased number of vertebrae. The three loci were associated with the NR6A1, PLAG1, and LCORL genes. The latter two have repeatedly been associated with loci controlling stature in other domestic animals and in humans. Most European domestic pigs are homozygous for the same haplotype at these three loci. We found an excess of derived nonsynonymous substitutions in domestic pigs, most likely reflecting both positive selection and relaxed purifying selection after domestication. Our analysis of structural variation revealed four duplications at the KIT locus that were exclusively present in white or white-spotted pigs, carrying the Dominant white, Patch, or Belt alleles. This discovery illustrates how structural changes have contributed to rapid phenotypic evolution in domestic animals and how alleles in domestic animals may evolve by the accumulation of multiple causative mutations as a response to strong directional selection.

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>)

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))

6 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^MC1R^/and+Taxon ID=^9823^/or+Gene Gephebase=^MC1R^/and+Taxon ID=^9825^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=^MC1R^/and+Taxon+ID=^9823^/or+Gene+Gephebase=^MC1R^/and+Taxon+ID=^9825^#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

possible @Introgression - the same mutation is known in European domestic pigs but happens independently in Hawaii