

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000589	GepheID
	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 (coat) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(coat)^#gephebase-summary-title)		Trait State in Taxon A	
Oryctolagus cuniculus		Trait State in Taxon B	
Oryctolagus cuniculus		Ancestral State	
Taxon A		Taxonomic Status	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated^#gephebase-summary-title)			
Taxon A		Taxon B	
Oryctolagus cuniculus		Oryctolagus cuniculus	
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Oryctolagus cuniculus^#gephebase-summary-title)		(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Oryctolagus cuniculus^#gephebase-summary-title)	
Common Name		Common Name	
rabbit		rabbit	
Synonyms		Synonyms	
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits		Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	
Rank		Rank	
species		species	
Lineage		Lineage	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagus		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagus	
Parent		Parent	
Oryctolagus () - (Rank: genus)		Oryctolagus () - (Rank: genus)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9986		9986	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

MC1R	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	Q01726 (http://www.uniprot.org/uniprot/Q01726)
	String	GenebankID or UniProtKB
CMM5; MSH-R; SHEP2; MSHR		CBJ17605 (https://www.ncbi.nlm.nih.gov/nuccore/CBJ17605)
9606.ENSPP00000451605		
(http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)		
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>)

No (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)	Molecular Type
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion~#gephebase-summary-title)	Aberration Type
10-99 bp	Deletion Size
30bp in-frame del of 102-111	Molecular Details of the Mutation
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title)	Experimental Evidence
Mutations in the melanocortin 1 receptor (MC1R) gene are associated with coat colours in the domestic rabbit (<i>Oryctolagus cuniculus</i>). (2006) (https://pubmed.ncbi.nlm.nih.gov/16978179)	Main Reference
Fontanesi L; Tazzoli M; Beretti F; Russo V	Authors
We sequenced almost the complete coding region of the MC1R gene in several domestic rabbits (<i>Oryctolagus cuniculus</i>) and identified four alleles: two wild-type alleles differing by two synonymous single nucleotide polymorphisms (c.333A>G;c.555T>C), one allele with a 30-nucleotide in-frame deletion (c.304_333del30) and one allele with a 6-nucleotide in-frame deletion (c.280_285del6). A polymerase chain reaction-based protocol was used to distinguish the wild-type alleles from the other two alleles in 263 rabbits belonging to 37 breeds or strains. All red/fawn/yellow rabbits were homozygous for the c.304_333del30 allele. This allele represents the recessive e allele at the extension locus identified through pioneering genetic studies in this species. All Californian, Checkered, Giant White and New Zealand White rabbits were homozygous for allele c.280_285del6, which was also observed in the heterozygous condition in a few other breeds. Black coat colour is part of the standard colour in Californian and Checkered breeds, in contrast to the two albino breeds, Giant White and New Zealand White. Following the nomenclature established for the rabbit extension locus, the c.280_285del6 allele, which is dominant over c.304_333del30, may be allele E(D) or allele E(S).	Abstract
	Additional References

RELATED GEPHE

4 (Agouti (ASIP), Melanophilin (MLPH), tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~9986~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
2 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~MC1R~/and+Taxon ID=~9986~/or+Gene Gephebase=~MC1R~/and+Taxon ID=~9986~#gephebase-summary-title)	Related Haplotypes

EXTERNAL LINKS

