

## GEPHE SUMMARY

MC1R ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^MC1R^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^MC1R^#gephebase-summary-title</a> )	Gephebase Gene	GP00000588	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title</a> )	Trait Category		
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+coat^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+coat^#gephebase-summary-title</a> )	Trait		
Oryctolagus cuniculus	Trait State in Taxon A		
Oryctolagus cuniculus	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title</a> )	Taxonomic Status		
	Taxon A		Taxon B
Oryctolagus cuniculus ( <a href="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</a> )	Latin Name	Oryctolagus cuniculus ( <a href="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</a> )	Latin Name
rabbit	Common Name	rabbit	Common Name
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	Synonyms	Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	Synonyms
species	Rank	species	Rank
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	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	Lineage
Oryctolagus () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984</a> )	Parent	Oryctolagus () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984</a> )	Parent
9986 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986</a> )	NCBI Taxonomy ID	9986 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 ( <a href="http://www.uniprot.org/uniprot/Q01726">http://www.uniprot.org/uniprot/Q01726</a> )	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	CBJ17605 ( <a href="https://www.ncbi.nlm.nih.gov/nucleotide/17605">https://www.ncbi.nlm.nih.gov/nucleotide/17605</a> )	GenebankID or UniProtKB
9606.ENSP00000451605 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000451605">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000451605</a> )	String		
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities		
GO:0008528 : G protein-coupled peptide receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008528">https://www.ebi.ac.uk/QuickGO/term/GO:0008528</a> )	GO - Molecular Function		
GO:0004977 : melanocortin receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004977">https://www.ebi.ac.uk/QuickGO/term/GO:0004977</a> )			
GO:0004980 : melanocyte-stimulating hormone receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004980">https://www.ebi.ac.uk/QuickGO/term/GO:0004980</a> )			
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

1-9 bp

Deletion Size

6bp in-frame del of 94-95

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 (*Oryctolagus cuniculus*). (2006) (<https://pubmed.ncbi.nlm.nih.gov/16978179>)

Main Reference

Fontanesi L; Tazzoli M; Beretti F; Russo V

Authors

**Abstract**  
We sequenced almost the complete coding region of the MC1R gene in several domestic rabbits (*Oryctolagus cuniculus*) 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).

Additional References

RELATED GEPHE

Related Genes

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 Haplotypes

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>)

EXTERNAL LINKS

