

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000605	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)		Trait Category	
Coloration (feathers; eyes) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration (feathers; eyes)^#gephebase-summary-title)		Trait	
Gallus gallus		Trait State in Taxon A	
Gallus gallus		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		Taxon B	
Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Gallus gallus^#gephebase-summary-title)		Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Gallus gallus^#gephebase-summary-title)	
chicken		chicken	
Gallus gallus domesticus; chicken; bantam; chickens		Gallus gallus domesticus; chicken; bantam; chickens	
species		species	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus	
Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)		Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)	
9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)		9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)	
No		No	

GENOTYPIC CHANGE

MC1R	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	Q01726 (http://www.uniprot.org/uniprot/Q01726)
	String	AGY49276 (https://www.ncbi.nlm.nih.gov/nuccore/AGY49276)
CMM5; MSH-R; SHEP2; MSHR		
9606.ENSPP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)		
Belongs to the G-protein coupled receptor 1 family.		
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 (<a +no^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Presumptive Null=">https://www.gephebase.org/search-criteria?/and+Presumptive Null="+No^#gephebase-summary-title)	
	Molecular Type
Coding (<a +coding^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Molecular Type=">https://www.gephebase.org/search-criteria?/and+Molecular Type="+Coding^#gephebase-summary-title)	
	Aberration Type
SNP (<a +snp^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Aberration Type=">https://www.gephebase.org/search-criteria?/and+Aberration Type="+SNP^#gephebase-summary-title)	
	SNP Coding Change
Nonsynonymous	
	Molecular Details of the Mutation
E92K; constitutive activation	
	Experimental Evidence
Linkage Mapping (<a +linkage="" href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=" mapping^#gephebase-summary-title"="">https://www.gephebase.org/search-criteria?/and+Experimental Evidence="+Linkage Mapping^#gephebase-summary-title)	

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

	Main Reference
A possible involvement of melanocortin 1-receptor in regulating feather color pigmentation in the chicken. (1996) (https://pubmed.ncbi.nlm.nih.gov/8764834)	
	Authors
Takeuchi S; Suzuki H; Yabuuchi M; Takahashi S	
	Abstract
The Extended black (E) locus on chromosome 1 acts within the melanocyte to regulate feather color pigmentation in the chicken. Several alleles exhibiting different pigmentation have been described and their phenotypes are similar to those of the murine extension locus which encodes melanocortin 1-receptor (MC1-R), the receptor for alpha-melanocyte-stimulating hormone (alpha-MSH). To investigate whether the MC1-R gene is responsible for E-locus function, we examined the structure of MC1-R in E-locus mutants by RFLP analysis and genomic DNA sequencing. In the most recessive allele (ey), which exhibits a uniformly red-yellow pigmentation, MC1-R was found to contain amino acid substitutions possibly causing functional deficiency. On the other hand, in the most dominant allele (E), which confers a uniformly black pigmentation, MC1-R possessed mutation responsible for a constitutively active MC1-R and resultant black coat color in mice. Our finding that the structure of MC1-R was affected by individual E-locus alleles strongly suggests that MC1-R is associated with the E-locus. Furthermore, since the same mutation of MC1-R was found in mice and chickens that exhibit the same pigmentation, it is possible that the regulatory mechanism of MC1-R function is shared in chickens and mammals.	
	Additional References
Melanocortin 1-receptor (MC1R) mutations are associated with plumage colour in chicken. (2003) (https://pubmed.ncbi.nlm.nih.gov/12873211) Association between polymorphism in the melanocortin 1 receptor gene and E locus plumage color phenotype. (2014) (https://pubmed.ncbi.nlm.nih.gov/24795300)	

RELATED GEPHE

	Related Genes
14 (ABCA1, Agouti (ASIP), CDKN2A, CYP19A1, EDN3, Endothelin receptor B2, GRAMD3, Melanophilin (MLPH), PMEL17, SLC45A2=MATP, SLCO1B3, SOX10, tyrosinase (TYR),	

tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9031^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

5 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^MC1R^/and+Taxon ID=^9031^/or+Gene Gephebase=^MC1R^/and+Taxon ID=^9031^#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS