

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^MC1R^#gephebase-summary-title)	Gephebase Gene	GP00002390	GepheID
Published	Entry Status	Courtier	Main curator

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	
	Latin Name		Latin Name
Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Gallus gallus^#gephebase-summary-title)	Gallus gallus	Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Gallus gallus^#gephebase-summary-title)	Gallus gallus
chicken	Common Name	chicken	Common Name
Gallus gallus domesticus; chicken; bantam; chickens	Synonyms	Gallus gallus domesticus; chicken; bantam; chickens	Synonyms
species	Rank	species	Rank
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	Lineage	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	Lineage
Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)	Parent	Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)	Parent
9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)	NCBI Taxonomy ID	9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	AGY49276 (https://www.ncbi.nlm.nih.gov/nuccore/AGY49276)	GenebankID or UniProtKB
9606.ENSPO0000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000451605)	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>)

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

the Arg213Cys mutation may be the cause of the loss or the decrease of function of the receptor to produce eumelanin

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	Arg	Cys	213

Main Reference

Association between polymorphism in the melanocortin 1 receptor gene and E locus plumage color phenotype. (2014) (<https://pubmed.ncbi.nlm.nih.gov/24795300>)

Authors

DÁvila SG; Gil MG; Resino-TalavÁin P; Campo JL

Abstract

The purpose of this study was to investigate the effect of the melanocortin 1 receptor (MC1R) gene on plumage color in chickens. The gene was sequenced in 77 males and 77 females from 13 Spanish breeds, carrying 6 different alleles in the E locus (E*E, E*R, E*WH, E*N, E*B, E*BC), a recessive wheaten (yellowish-white) tester line (E*Y), and a White Leghorn population (heterozygous E*E). A total of 11 significant SNP were detected. Nine of them were nonsynonymous (T212C, G274A, G376A, T398AC, G409A, A427G, C637T, A644C, and G646A, corresponding to amino acid changes Met72Thr, Glu92Lys, Val126Ile, Leu133GlnPro, Ala137Thr, Thr143Ala, Arg213Cys, His215Pro, and Val216Ile), and 2 were synonymous (C69T and C834T). With respect to the significant SNP, 7 had an allelic frequency of 0.5 or greater for some of the alleles at the E locus. These results indicated a significant correlation between MC1R polymorphism and the presence of different alleles at the E locus. All the populations carrying the E*E or E*R alleles, except the Birchen Leonesa, had the G274A polymorphism. Eleven haplotypes were made with 7 of the significant SNP. The distribution of these haplotypes in the different alleles of the E locus showed that each haplotype was predominantly associated to one allele. The number of haplotypes was greatest for the Black Menorca, Birchen Leonesa, and Blue Andaluza breeds, whereas the Quail Castellana and Red-barred Vasca breeds were monomorphic. Our results suggested that the Glu92Lys mutation may be responsible of the activation of the receptor for eumelanin production, being necessary but not sufficient to express the extended black phenotype. They also suggested that the Arg213Cys mutation may be the cause of the loss or the decrease of function of the receptor to produce eumelanin, and the Ala137Thr mutation may be a candidate to attenuate the Glu92Lys effect. The observed co-segregation of the E locus alleles and polymorphisms in MC1R confirms that the E locus is equivalent to MC1R.

Additional References

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>)

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