

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000587	GepheID
Published	Entry Status	Martin	Main curator

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+Trait=^Morphology^#gephebase-summary-title)			
	Trait		
Coloration (feathers) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration(feathers)^#gephebase-summary-title)			
	Trait State in Taxon A		
Numida meleagris			
	Trait State in Taxon B		
Numida meleagris			
	Ancestral State		
Taxon A			
	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=^Domesticated^#gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		Latin Name
Numida meleagris (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Numida meleagris^#gephebase-summary-title)		Numida meleagris (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Numida meleagris^#gephebase-summary-title)	
	Common Name		Common Name
helmeted guineafowl		helmeted guineafowl	
	Synonyms		Synonyms
helmeted guineafowl; Numida meleagris (Linnaeus, 1758)		helmeted guineafowl; Numida meleagris (Linnaeus, 1758)	
	Rank		Rank
species		species	
	Lineage		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; Numididae; Numida		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; Numididae; Numida	
	Parent		Parent
Numida () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8995)		Numida () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8995)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
8996 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8996)		8996 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8996)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

MC1R	Generic Gene Name	Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	ADC39974 (https://www.ncbi.nlm.nih.gov/nucore/ADC39974)	GenebankID or UniProtKB
9606.ENSPP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)	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 (<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)	
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)	Molecular Type
Deletion (<a +deletion^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Aberration Type=">https://www.gephebase.org/search-criteria?/and+Aberration Type="+Deletion^#gephebase-summary-title)	Aberration Type
1-9 bp	Deletion Size
Phe256del	Molecular Details of the Mutation
Candidate Gene (<a +candidate="" gene^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=">https://www.gephebase.org/search-criteria?/and+Experimental Evidence="+Candidate Gene^#gephebase-summary-title)	Experimental Evidence
Melanism in guinea fowl (Numida meleagris) is associated with a deletion of Phenylalanine-256 in the MC1R gene. (2010) (https://pubmed.ncbi.nlm.nih.gov/20477788)	Main Reference
Vidal O; Araguas RM; Fern��ndez E; Heras S; Sanz N; Pla C	Authors
We have characterized a deletion in the MC1R gene causing the loss of one amino acid (p.Phe256del), which is perfectly associated with melanism in guinea fowl (Numida meleagris). Co-segregation of the p.Phe256del with melanism was confirmed in 25 offspring born from a cross of two heterozygote birds; therefore we suggest that this mutation is responsible for the black phenotype. Interestingly, this is the first case of recessive melanism linked to MC1R.	Abstract
	Additional References

RELATED GEPHE

No matches found.	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA000374/8996/>

