

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000619	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 (red hair) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration (red hair)^#gephebase-summary-title)		Trait	
Homo sapiens		Trait State in Taxon A	
Homo sapiens		Trait State in Taxon B	
Data not curated		Ancestral State	
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Homo sapiens^#gephebase-summary-title)		Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Homo sapiens^#gephebase-summary-title)	
human		human	
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens		human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens	
species		species	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo	
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)		Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)		9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	
No		No	

GENOTYPIC CHANGE

MC1R	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	GenebankID or UniProtKB
	String	
9606.ENSPO0000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000451605)		
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)		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0031625>)

(<https://www.ebi.ac.uk/QuickGO/term/GO:0070914>)

(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)

No (<https://www.qephebase.org/search-criteria/?and+Presumptive+Null=%5E%5CNo%5E%5C#qephebase-summary-title>)

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular+Type=^Coding^#gephebase-summary-title>)

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration+Type=^SNP^#gephebase-summary-title>)

Nonsynonymous

Asp84Glu

Association Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Association+Mapping^#gephebase-summary-title>)

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

Genetic determinants of hair, eye and skin pigmentation in Europeans. (2007) (<https://pubmed.ncbi.nlm.nih.gov/17952075>)

Sulem P; Gudbjartsson DF; Stacey SN; Helgason A; Rafnar T; Magnusson KP; Manolescu A; Karason A; Palsson A; Thorleifsson G; Jakobsdottir M; Steinberg S; Pállsson S; Jonasson F; Siqureirsson B; Thorisdottir K; Ragnarsson R; Benediktsdottir KR; Aben KK; Kiemeny LA; Olafsson JH; Gulcher J; Kong A; Thorsteinsdottir U; Stefansson K

Hair, skin and eye colors are highly heritable and visible traits in humans. We carried out a genome-wide association scan for variants associated with hair and eye pigmentation, skin sensitivity to sun and freckling among 2,986 Icelanders. We then tested the most closely associated SNPs from six regions--four not previously implicated in the normal variation of human pigmentation--and replicated their association in a second sample of 2,718 Icelanders and a sample of 1,214 Dutch. The SNPs from all six regions met the criteria for genome-wide significance. A variant in *SLC24A4* is associated with eye and hair color, a variant near *KITLG* is associated with hair color, two coding variants in *TYR* are associated with eye color and freckles, and a variant on 6p25.3 is associated with freckles. The fifth region provided refinements to a previously reported association in *OCA2*, and the sixth encompasses previously described variants in *MC1R*.

A genome-wide association study identifies novel alleles associated with hair color and skin pigmentation. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18483556>)

14 (Aqouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, Kit ligand, MFSD12, Oca2, OPRM1, SLC24A5 (NCKX5), SLC45A2=MATP, TPCN2, tyrosinase (TYR),

tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

12 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^MC1R^/and+Taxon ID=^9606^/or+Gene Gephebase=^MC1R^/and+Taxon ID=^9606^#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS