

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000626	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

		Trait Category	
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)		Trait	
Coloration (red hair) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration (red hair)^#gephebase-summary-title)		Trait State in Taxon A	
Homo sapiens		Trait State in Taxon B	
Homo sapiens		Ancestral State	
Data not curated		Taxonomic Status	
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific^#gephebase-summary-title)			
Taxon A		Taxon B	
Homo sapiens		Homo sapiens	
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Homo sapiens^#gephebase-summary-title)		(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)		Homo () - (Rank: genus)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	
9606		9606	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	
No		No	

GENOTYPIC CHANGE

		Generic Gene Name	
MC1R		Q01726 (http://www.uniprot.org/uniprot/Q01726)	
CMM5; MSH-R; SHEP2; MSHR		NP_002377 (https://www.ncbi.nlm.nih.gov/nuccore/NP_002377)	
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 - Molecular Function	
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](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

Asp294His

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>)
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>)

Related Haplotypes

EXTERNAL LINKS

COMMENTS