

GEPHE SUMMARY

	Gephebase Gene		GepheID
MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)		GP00000621	
	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
	Latin Name		Latin Name
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)	
	Common Name		Common Name
human		human	
	Synonyms		Synonyms
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	
	Rank		Rank
species		species	
	Lineage		Lineage
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	
	Parent		Parent
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)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
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)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
MC1R		Q01726 (http://www.uniprot.org/uniprot/Q01726)	
	Synonyms		GenebankID or UniProtKB
CMM5; MSH-R; SHEP2; MSHR		NP_002377 (https://www.ncbi.nlm.nih.gov/nuccore/NP_002377)	
	String		
9606.ENSPP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)			
	Sequence Similarities		
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>)

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

Nonsynonymous

Arg142His

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