

GEPHE SUMMARY

	Gephebase Gene		GepheID
tyrosinase (TYR) (<a +tyrosinase+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+GeneGephebase=">https://www.gephebase.org/search-criteria?/and+GeneGephebase="+tyrosinase+"#gephebase-summary-title)	GP00001144		
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+TraitCategory=">https://www.gephebase.org/search-criteria?/and+TraitCategory="+Morphology+"#gephebase-summary-title)	Trait		
Coloration (eyes; hair; skin) (<a +coloration+(eyes;+hair;+skin)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+(eyes;+hair;+skin)+"#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 (<a +intraspecific+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=">https://www.gephebase.org/search-criteria?/and+TaxonomicStatus="+Intraspecific+"#gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens+"#gephebase-summary-title)	Common Name		Common Name
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	Rank		Rank
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	Parent		Parent
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)	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No			No

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Tyr	Synonyms		GenebankID or UniProtKB
c; Oca1; skc35; albino	String		AAB60319 (https://www.ncbi.nlm.nih.gov/nuccore/AAB60319)
10090.ENSMUSP00000004770 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770)	Sequence Similarities		
Belongs to the tyrosinase family.	GO - Molecular Function		
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)			
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)			

GO:0005507 : copper ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005507>)
GO:0004503 : monophenol monooxygenase activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0004503>)

GO - Biological Process

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:0008283 : cell proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0008283>)
GO:0033280 : response to vitamin D (<https://www.ebi.ac.uk/QuickGO/term/GO:0033280>)
GO:0051591 : response to cAMP (<https://www.ebi.ac.uk/QuickGO/term/GO:0051591>)
GO:0009411 : response to UV (<https://www.ebi.ac.uk/QuickGO/term/GO:0009411>)
GO:0048538 : thymus development (<https://www.ebi.ac.uk/QuickGO/term/GO:0048538>)

GO - Cellular Component

GO:0016021 : integral component of membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0043231 : intracellular membrane-bounded organelle (<https://www.ebi.ac.uk/QuickGO/term/GO:0043231>)
GO:0048471 : perinuclear region of cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0048471>)
GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)
GO:0033162 : melanosome membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0033162>)

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-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

R402Q

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; Sigurgeirsson 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.

Two newly identified genetic determinants of pigmentation in Europeans. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18488028>)
ASIP and TYR pigmentation variants associate with cutaneous melanoma and basal cell carcinoma. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18488027>)

RELATED GEPHE

14 (Agouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, Kit ligand, MC1R, MFSD12, Oca2, OPRM1, SLC24A5 (NCKX5), SLC45A2=MATP, TPCN2, tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~9606~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

1 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~tyrosinase \(TYR\)~/and+Taxon ID=~9606~/or+Gene Gephebase=~tyrosinase \(TYR\)~/and+Taxon ID=~9606~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~tyrosinase (TYR)~/and+Taxon ID=~9606~/or+Gene Gephebase=~tyrosinase (TYR)~/and+Taxon ID=~9606~#gephebase-summary-title))

EXTERNAL LINKS

