

Oca2 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~Oca2^#gephebase-summary-title)	Gephebase Gene	GP00000748	GepheID
Published	Entry Status	Martin	Main curator

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology~#gephebase-summary-title)			
	Trait		
Coloration (skin) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(skin)~#gephebase-summary-title)			
	Trait State in Taxon A		
Homo sapiens - Asia			
	Trait State in Taxon B		
Homo sapiens - Eastern Asia - Han Chinese with lighter skin			
	Ancestral State		
Taxon A			
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific~#gephebase-summary-title)	Taxonomic Status		
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; Hominae; 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; Hominae; 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 Infrasppecies?		is Taxon B an Infrasppecies?
No		No	

UniProtKB Mus musculus	Generic Gene Name	UniProtKB Mus musculus
Q62052 (http://www.uniprot.org/uniprot/Q62052)	Oca2	
GenebankID or UniProtKB	Synonyms	
AAH12097 (https://www.ncbi.nlm.nih.gov/nucore/AAH12097)	p: D7Nic1; p<cas>; D7H15S12; D7lcr28RN; P	
	String	
	10090.ENSMUSP00000032633 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000032633)	
	Sequence Similarities	
	Belongs to the CitM (TC 2.A.11) transporter family.	
	GO - Molecular Function	
	-	
	GO - Biological Process	
	GO:0055085 : transmembrane transport (https://www.ebi.ac.uk/QuickGO/term/GO:0055085)	

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:0048066 : developmental pigmentation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048066>)
GO:0030318 : melanocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)
GO:0007286 : spermatid development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007286>)

GO - Cellular Component

GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
GO:0010008 : endosome membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010008>)
GO:0005789 : endoplasmic reticulum membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005789>)
GO:0005765 : lysosomal membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005765>)
GO:0033162 : melanosome membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033162>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

His615Arg

Experimental Evidence

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

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

Main Reference

Association of the OCA2 polymorphism His615Arg with melanin content in east Asian populations: further evidence of convergent evolution of skin pigmentation. (2010)
(<https://pubmed.ncbi.nlm.nih.gov/20221248>)

Authors

Edwards M; Bigham A; Tan J; Li S; Gozdzik A; Ross K; Jin L; Parra EJ

Abstract

The last decade has witnessed important advances in our understanding of the genetics of pigmentation in European populations, but very little is known about the genes involved in skin pigmentation variation in East Asian populations. Here, we present the results of a study evaluating the association of 10 Single Nucleotide Polymorphisms (SNPs) located within 5 pigmentation candidate genes (OCA2, DCT, ADAM17, ADAMTS20, and TYRP1) with skin pigmentation measured quantitatively in a sample of individuals of East Asian ancestry living in Canada. We show that the non-synonymous polymorphism rs1800414 (His615Arg) located within the OCA2 gene is significantly associated with skin pigmentation in this sample. We replicated this result in an independent sample of Chinese individuals of Han ancestry. This polymorphism is characterized by a derived allele that is present at a high frequency in East Asian populations, but is absent in other population groups. In both samples, individuals with the derived G allele, which codes for the amino acid arginine, show lower melanin levels than those with the ancestral A allele, which codes for the amino acid histidine. An analysis of this non-synonymous polymorphism using several programs to predict potential functional effects provides additional support for the role of this SNP in skin pigmentation variation in East Asian populations. Our results are consistent with previous research indicating that evolution to lightly-pigmented skin occurred, at least in part, independently in Europe and East Asia.

Additional References

A Genetic Mechanism for Convergent Skin Lightening during Recent Human Evolution. (2016) (<https://pubmed.ncbi.nlm.nih.gov/26744415>)

RELATED GEPHE

Related Genes

14 (Agouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, Kit ligand, MC1R, MFSD12, 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

1 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Oca2~/and+Taxon ID=~9606~/or+Gene Gephebase=~Oca2~/and+Taxon ID=~9606~#gephebase-summary-title>)

EXTERNAL LINKS

