GEPHE SUMMARY

Gephebase Gene

OPRM1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^OPRM1^#gephebase-summary-title)

GepheID

GP00001536

Entry Status

Published

Main curator

Prigent

PHENOTYPIC CHANGE

Trait Category

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^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

Human Europeans with lighter skin pigmentation

Trait State in Taxon B

Human Native Americans with darker skin pigmentation

Ancestral State

Taxon A

Taxonomic Status

Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name

Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name

human

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

species

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

Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID

9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon A an Infraspecies?

Yes

Taxon A Description

Human Europeans with lighter skin pigmentation

Taxon B

Latin Name

Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name

human

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

species

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

Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID

9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Human Native Americans with darker skin pigmentation

GENOTYPIC CHANGE

Generic Gene Name

OPRM1

Synonyms

MOP; MOR; LMOR; MOR1; OPRM; M-OR-1

String

9606.ENSP00000394624 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000394624)

Sequence Similarities

Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function

GO:0004930 : G protein-coupled receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004930)

UniProtKB Homo sapiens

P35372 (http://www.uniprot.org/uniprot/P35372)

GenebankID or UniProtKB

()

GO:0031681 : G-protein beta-subunit binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0031681)
GO:0019904 : protein domain specific binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0019904)
GO:0001965 : G-protein alpha-subunit binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0001965)
GO:0005245 : voltage-gated calcium channel activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0005245)
GO:0004979 : beta-endorphin receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004979)
GO:0031005 : filamin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031005)
GO:0038047 : morphine receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0038047)
GO:0042923 : neuropeptide binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042923)
GO:0008022 : protein C-terminus binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0008022)

GO - Biological Process

GO:0048149 : behavioral response to ethanol
(https://www.ebi.ac.uk/QuickGO/term/GO:0048149)
GO:0007186 : G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007186)
GO:0070374 : positive regulation of ERK1 and ERK2 cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0070374)
GO:0008285 : negative regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0008285)
GO:0045019 : negative regulation of nitric oxide biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0045019)
GO:0007204 : positive regulation of cytosolic calcium ion concentration
(https://www.ebi.ac.uk/QuickGO/term/GO:0007204)
GO:0051930 : regulation of sensory perception of pain
(https://www.ebi.ac.uk/QuickGO/term/GO:0051930)
GO:0032496 : response to lipopolysaccharide
(https://www.ebi.ac.uk/QuickGO/term/GO:0032496)
GO:0019221 : cytokine-mediated signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0019221)
GO:0060079 : excitatory postsynaptic potential
(https://www.ebi.ac.uk/QuickGO/term/GO:0060079)
GO:0007626 : locomotory behavior (https://www.ebi.ac.uk/QuickGO/term/GO:0007626)
GO:0007200 : phospholipase C-activating G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007200)
GO:0019233 : sensory perception of pain
(https://www.ebi.ac.uk/QuickGO/term/GO:0019233)
GO:0042060 : wound healing (https://www.ebi.ac.uk/QuickGO/term/GO:0042060)
GO:0032100 : positive regulation of appetite
(https://www.ebi.ac.uk/QuickGO/term/GO:0032100)
GO:0032094 : response to food (https://www.ebi.ac.uk/QuickGO/term/GO:0032094)
GO:0043950 : positive regulation of cAMP-mediated signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0043950)
GO:0045429 : positive regulation of nitric oxide biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0045429)
GO:0042220 : response to cocaine (https://www.ebi.ac.uk/QuickGO/term/GO:0042220)
GO:0009314 : response to radiation (https://www.ebi.ac.uk/QuickGO/term/GO:0009314)
GO:0007187 : G protein-coupled receptor signaling pathway, coupled to cyclic nucleotide
second messenger (https://www.ebi.ac.uk/QuickGO/term/GO:0007187)
GO:0007218 : neuropeptide signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007218)
GO:0043951 : negative regulation of cAMP-mediated signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0043951)
GO:0002438 : acute inflammatory response to antigenic stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:0002438)
GO:0007191 : adenylate cyclase-activating dopamine receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007191)
GO:0031635 : adenylate cyclase-inhibiting opioid receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0031635)
GO:0042755 : eating behavior (https://www.ebi.ac.uk/QuickGO/term/GO:0042755)
GO:0044849 : estrous cycle (https://www.ebi.ac.uk/QuickGO/term/GO:0044849)
GO:0007194 : negative regulation of adenylate cyclase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0007194)
GO:0106072 : negative regulation of adenylate cyclase-activating G protein-coupled
receptor signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0106072)
GO:0051481 : negative regulation of cytosolic calcium ion concentration
(https://www.ebi.ac.uk/QuickGO/term/GO:0051481)
GO:0061358 : negative regulation of Wnt protein secretion
(https://www.ebi.ac.uk/QuickGO/term/GO:0061358)
GO:0038003 : opioid receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0038003)
GO:0050769 : positive regulation of neurogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0050769)

GO:0080135 : regulation of cellular response to stress
(https://www.ebi.ac.uk/QuickGO/term/GO:0080135)
GO:2000310 : regulation of NMDA receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:2000310)
GO:0070848 : response to growth factor
(https://www.ebi.ac.uk/QuickGO/term/GO:0070848)
GO:0007600 : sensory perception (https://www.ebi.ac.uk/QuickGO/term/GO:0007600)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005925 : focal adhesion (https://www.ebi.ac.uk/QuickGO/term/GO:0005925)
GO:0045121 : membrane raft (https://www.ebi.ac.uk/QuickGO/term/GO:0045121)
GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0005794 : Golgi apparatus (https://www.ebi.ac.uk/QuickGO/term/GO:0005794)
GO:0005623 : cell (https://www.ebi.ac.uk/QuickGO/term/GO:0005623)
GO:0030425 : dendrite (https://www.ebi.ac.uk/QuickGO/term/GO:0030425)
GO:0005783 : endoplasmic reticulum
(https://www.ebi.ac.uk/QuickGO/term/GO:0005783)
GO:0005768 : endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005768)
GO:0030424 : axon (https://www.ebi.ac.uk/QuickGO/term/GO:0030424)
GO:0042383 : sarcolemma (https://www.ebi.ac.uk/QuickGO/term/GO:0042383)
GO:0043204 : perikaryon (https://www.ebi.ac.uk/QuickGO/term/GO:0043204)
GO:0032839 : dendrite cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0032839)
GO:0032590 : dendrite membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0032590)
GO:0099055 : integral component of postsynaptic membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0099055)
GO:0099056 : integral component of presynaptic membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0099056)
GO:0097444 : spine apparatus (https://www.ebi.ac.uk/QuickGO/term/GO:0097444)

Presumptive Null

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

C>T associated SNP

Experimental Evidence

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

Main Reference

OPRM1 and EGFR contribute to skin pigmentation differences between Indigenous Americans and Europeans. (2012) (https://pubmed.ncbi.nlm.nih.gov/22198722)

Authors

Quillen EE; Bauchet M; Bigham AW; Delgado-Burbano ME; Faust FX; Klimentidis YC; Mao X; Stoneking M; Shriver MD

Abstract

Contemporary variation in skin pigmentation is the result of hundreds of thousands years of human evolution in new and changing environments. Previous studies have identified several genes involved in skin pigmentation differences among African, Asian, and European populations. However, none have examined skin pigmentation variation among Indigenous American populations, creating a critical gap in our understanding of skin pigmentation variation. This study investigates signatures of selection at 76 pigmentation candidate genes that may contribute to skin pigmentation differences between Indigenous Americans and Europeans. Analysis was performed on two samples of Indigenous Americans genotyped on genome-wide SNP arrays. Using four tests for natural selection--locus-specific branch length (LSBL), ratio of heterozygosities (lnRH), Tajima's D difference, and extended haplotype homozygosity (EHH)--we identified 14 selection-nominated candidate genes (SNCGs). SNPs in each of the SNCGs were tested for association with skin pigmentation in 515 admixed Indigenous American and European individuals from regions of the Americas with high ground-level ultraviolet radiation. In addition to SLC24A5 and SLC45A2, genes previously associated with European/non-European differences in skin pigmentation, OPRM1 and EGFR were associated with variation in skin pigmentation in New World populations for the first time.

Additional References

RELATED GEPHE

Related Genes

14 (Agouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, Kit ligand, MC1R, MFSD12, Oca2, 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

No matches found.

EXTERNAL LINKS

COMMENTS