GEPHE SUMMARY

Gephebase Gene

EIF2S2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^EIF2S2^#gephebase-summary-title)

GepheID

GP00001359

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-European dark pigmentation

Trait State in Taxon B

Human- European light pigmentation

Ancestral State

Unknown

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?

No

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?

No

GENOTYPIC CHANGE

Generic Gene Name

EIF2S2

Synonyms

EIF2; EIF2B; PPP1R67; EIF2beta; eIF-2-beta

String

9606.ENSP00000364119 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000364119)

Sequence Similarities

Belongs to the eIF-2-beta/eIF-5 family.

GO - Molecular Function

GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0003723 : RNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003723)
GO:0003743 : translation initiation factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003743)

UniProtKB Homo sapiens

P20042 (http://www.uniprot.org/uniprot/P20042)

GenebankID or UniProtKB

()

GO:0008135 : translation factor activity, RNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0008135)

GO - Biological Process

GO:0055085 : transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0055085)
GO:0008584 : male gonad development
(https://www.ebi.ac.uk/QuickGO/term/GO:0008584)
GO:0001701 : in utero embryonic development
(https://www.ebi.ac.uk/QuickGO/term/GO:0001701)
GO:0002176 : male germ cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0002176)
GO:0006413 : translational initiation (https://www.ebi.ac.uk/QuickGO/term/GO:0006413)

GO - Cellular Component

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:0005850 : eukaryotic translation initiation factor 2 complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0005850)

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

unknown

Experimental Evidence

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

Main Reference

Genetics of skin color variation in Europeans: genome-wide association studies with functional follow-up. (2015) (https://pubmed.ncbi.nlm.nih.gov/25963972)

Authors

Liu F; Visser M; Duffy DL; Hysi PG; Jacobs LC; Lao O; Zhong K; Walsh S; Chaitanya L; Wollstein A; Zhu G; Montgomery GW; Henders AK; Mangino M; Glass D; Bataille V; Sturm RA; Rivadeneira F; Hofman A; van IJcken WF; Uitterlinden AG; Palstra RJ; Spector TD; Martin NG; Nijsten TE; Kayser M

Abstract

In the International Visible Trait Genetics (VisiGen) Consortium, we investigated the genetics of human skin color by combining a series of genome-wide association studies (GWAS) in a total of 17,262 Europeans with functional follow-up of discovered loci. Our GWAS provide the first genome-wide significant evidence for chromosome 20q11.22 harboring the ASIP gene being explicitly associated with skin color in Europeans. In addition, genomic loci at 5p13.2 (SLC45A2), 6p25.3 (IRF4), 15q13.1 (HERC2/OCA2), and 16q24.3 (MC1R) were confirmed to be involved in skin coloration in Europeans. In follow-up gene expression and regulation studies of 22 genes in 20q11.22, we highlighted two novel genes EIF2S2 and GSS, serving as competing functional candidates in this region and providing future research lines. A genetically inferred skin color score obtained from the 9 top-associated SNPs from 9 genes in 940 worldwide samples (HGDP-CEPH) showed a clear gradual pattern in Western Eurasians similar to the distribution of physical skin color, suggesting the used 9 SNPs as suitable markers for DNA prediction of skin color in Europeans and neighboring populations, relevant in future forensic and anthropological investigations.

Additional References

RELATED GEPHE

Related Genes

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

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

the most likely candidate (with GSS) responsible for the genetic association with skin pigmentation