GEPHE SUMMARY

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

Entry Status
Published

GepheID
GP00001362

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
GSS

Synonyms
GSHS; HEL-S-64p; HEL-S-88n

String
9606.ENSP00000216951 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000216951)

Sequence Similarities
Belongs to the eukaryotic GSH synthase family.

GO - Molecular Function
GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)
GO:0042803 : protein homodimerization activity

UniProtKB Homo sapiens
P48637 (http://www.uniprot.org/uniprot/P48637)

GenebankID or UniProtKB
()

(https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0000287 : magnesium ion binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0000287)
GO:0043295 : glutathione binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043295)
GO:0004363 : glutathione synthase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004363)
GO:0016594 : glycine binding (https://www.ebi.ac.uk/QuickGO/term/GO:0016594)

GO - Biological Process

GO:0007399 : nervous system development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007399)
GO:0007568 : aging (https://www.ebi.ac.uk/QuickGO/term/GO:0007568)
GO:0043200 : response to amino acid
(https://www.ebi.ac.uk/QuickGO/term/GO:0043200)
GO:0006979 : response to oxidative stress
(https://www.ebi.ac.uk/QuickGO/term/GO:0006979)
GO:0046686 : response to cadmium ion
(https://www.ebi.ac.uk/QuickGO/term/GO:0046686)
GO:0031667 : response to nutrient levels
(https://www.ebi.ac.uk/QuickGO/term/GO:0031667)
GO:0006520 : cellular amino acid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006520)
GO:0006750 : glutathione biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006750)
GO:0034612 : response to tumor necrosis factor
(https://www.ebi.ac.uk/QuickGO/term/GO:0034612)
GO:0009410 : response to xenobiotic stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:0009410)

GO - Cellular Component

GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0070062 : extracellular exosome (https://www.ebi.ac.uk/QuickGO/term/GO:0070062)

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, EIF2S2, 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 EIF2S2) responsible for the genetic association with skin pigmentation