

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