GEPHE SUMMARY

Gephebase Gene
MFSD12 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MFSD12^#gephebase-summary-title)

GepheID
GP00002245

Entry Status
Published

Main curator
Martin

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
variation in melanization of skin melanocyte across East Asian and Native American populations

Trait State in Taxon B
variation in melanization of skin melanocyte across East Asian and Native American populations

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?
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
MFSD12

Synonyms
PP3501; C19orf28

String
9606.ENSP00000347583 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000347583)

Sequence Similarities
Belongs to the major facilitator superfamily.

GO - Molecular Function
GO:0015293 : symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015293)

GO - Biological Process

UniProtKB Homo sapiens
Q6NUT3 (http://www.uniprot.org/uniprot/Q6NUT3)

GenebankID or UniProtKB
()

GO:0048022 : negative regulation of melanin biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0048022)
GO:0008643 : carbohydrate transport (https://www.ebi.ac.uk/QuickGO/term/GO:0008643)
GO:0071702 : organic substance transport (https://www.ebi.ac.uk/QuickGO/term/GO:0071702)

GO - Cellular Component

GO:0005887 : integral component of plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0005765 : lysosomal membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005765)
GO:0005770 : late endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005770)

Presumptive Null

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#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

Y182H

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	Tyr	His	182

Main Reference

A GWAS in Latin Americans highlights the convergent evolution of lighter skin pigmentation in Eurasia. (2019) (https://pubmed.ncbi.nlm.nih.gov/30664655)

Authors

Adhikari K; Mendoza-Revilla J; Sohail A; Fuentes-Guajardo M; Lampert J; ChacÃ³n-Duque JC; Hurtado M; Villegas V; Granja V; AcuÃ±a-Alonzo V; Jaramillo C; Arias W; Lozano RB; Everardo P; GÃ³mez-ValdÃ©s J; Villamil-RamÃrez H; Silva de Cerqueira CC; Hunemeier T; Ramallo V; Schuler-Faccini L; Salzano FM; Gonzalez-JosÃ© R; Bortolini MC; Canizales-Quinteros S; Gallo C; Poletti G; Bedoya G; Rothhammer F; Tobin DJ; Fumagalli M; Balding D; Ruiz-Linares A

Abstract

We report a genome-wide association scan in >6,000 Latin Americans for pigmentation of skin and eyes. We found eighteen signals of association at twelve genomic regions. These include one novel locus for skin pigmentation (in 10q26) and three novel loci for eye pigmentation (in 1q32, 20q13 and 22q12). We demonstrate the presence of multiple independent signals of association in the 11q14 and 15q13 regions (comprising the GRM5/TYR and HERC2/OCA2 genes, respectively) and several epistatic interactions among independently associated alleles. Strongest association with skin pigmentation at 19p13 was observed for an Y182H missense variant (common only in East Asians and Native Americans) in MFSD12, a gene recently associated with skin pigmentation in Africans. We show that the frequency of the derived allele at Y182H is significantly correlated with lower solar radiation intensity in East Asia and infer that MFSD12 was under selection in East Asians, probably after their split from Europeans.

Additional References

Loci associated with skin pigmentation identified in African populations. (2017) (https://pubmed.ncbi.nlm.nih.gov/29025994)

RELATED GEPHE

Related Genes

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

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^MFSD12^/and+Taxon ID=^9606^/or+Gene Gephebase=^MFSD12^/and+Taxon ID=^9606^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@Parallelism @Fitness @AllelicSeries ; functional evidence for the function of this gene in skin melanocytes is presented in the references.