

GEPHE SUMMARY

MFSD12 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~MFSD12^#gephebase-summary-title)	Gephebase Gene	GP00002245	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology^#gephebase-summary-title)	Trait Category		
Coloration (skin) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+skin^#gephebase-summary-title)	Trait		
variation in melanization of skin melanocyte across East Asian and Native American populations	Trait State in Taxon A		
variation in melanization of skin melanocyte across East Asian and Native American populations	Trait State in Taxon B		
Taxon A	Ancestral State		
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Intraspecific^#gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
	Latin Name		Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Homo+sapiens^#gephebase-summary-title)	Latin Name	Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Homo+sapiens^#gephebase-summary-title)	Latin Name
human	Common Name	human	Common Name
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	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	Synonyms
species	Rank	species	Rank
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	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	Lineage
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)	Parent	Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)	Parent
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)	NCBI Taxonomy ID	9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

MFSD12	Generic Gene Name	Q6NUT3 (http://www.uniprot.org/uniprot/Q6NUT3)	UniProtKB Homo sapiens
PP3501; C19orf28	Synonyms		GenebankID or UniProtKB
9606.ENSPP00000347583 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000347583)	String		
Belongs to the major facilitator superfamily.	Sequence Similarities		
GO:0015293 : symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015293)	GO - Molecular Function		
	GO - Biological Process		

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 ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes))

Molecular Type

Coding ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding))

Aberration Type

SNP ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=~SNP))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Y182H

Experimental Evidence

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

	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; Chacón-Duque JC; Hurtado M; Villegas V; Granja V; Acuña-Alonzo V; Jaramillo C; Arias W; Lozano RB; Everardo P; Gómez-Valdés J; Villamil-Ramírez H; Silva de Cerqueira CC; Hunemeier T; Ramallo V; Schuler-Faccini L; Salzano FM; Gonzalez-José 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#gpepbase-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#gpepbase-summary-title>)

EXTERNAL LINKS

COMMENTS

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

