GEPHE SUMMARY

Gephebase Gene
SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SLC45A2=MATP^#gephebase-summary-title)

GepheID
GP00001061

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 (eyes; hair; skin) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (eyes; hair; skin)^#gephebase-summary-title)

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Homo sapiens

Ancestral State
Data not curated

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
SLC45A2

Synonyms
1A1; AIM1; MATP; OCA4; SHEP5

String
9606.ENSP00000296589 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000296589)

Sequence Similarities
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.

GO - Molecular Function
GO:0008506 : sucrose:proton symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008506)

GO - Biological Process

UniProtKB Homo sapiens
Q9UMX9 (http://www.uniprot.org/uniprot/Q9UMX9)

GenebankID or UniProtKB
AF172849 (https://www.ncbi.nlm.nih.gov/nuccore/AF172849)

GO:0042438 : melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
GO:0048066 : developmental pigmentation
(https://www.ebi.ac.uk/QuickGO/term/GO:0048066)
GO:0007601 : visual perception (https://www.ebi.ac.uk/QuickGO/term/GO:0007601)
GO:0050896 : response to stimulus (https://www.ebi.ac.uk/QuickGO/term/GO:0050896)
GO:0015770 : sucrose transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015770)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0033162 : melanosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0033162)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#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

Phe374Leu

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	-	-	-

Main Reference

Single nucleotide polymorphisms in the MATP gene are associated with normal human pigmentation variation. (2005) (https://pubmed.ncbi.nlm.nih.gov/15714523)

Authors

Graf J; Hodgson R; van Daal A

Abstract

Human physical pigmentation is determined by the type and amount of melanin and the process of pigmentation production probably involves more than 100 genes. A failure to synthesize melanin results in oculocutaneous albinism (OCA). A recently identified form of OCA results from mutations in the Membrane Associated Transporter Protein (MATP) gene. The role of MATP in human pigmentation is not clear. We investigated the role of two nonpathogenic nonsynonymous single nucleotide polymorphisms (SNPs) in the MATP gene to determine if they are associated with normal human skin, hair, and eye color variation. A total of 608 individuals from four different population groups (456 Caucasians, 31 Asians, 70 African-Americans, and 51 Australian Aborigines) were genotyped for c.814G>A (p.Glu272Lys) and c.1122C>G (p.Phe374Leu). Results indicate that the allele frequencies of both polymorphisms are significantly different between population groups. The two alleles, 374Leu and 272Lys, are significantly associated with dark hair, skin, and eye color in Caucasians. The odds ratios (ORs) of the LeuLeu genotype for black hair and olive skin are 25.63 and 28.65, respectively, and for the LysLys genotype are 43.23 and 8.27, respectively. The OR for eye color is lower at 3.48 for the LeuLeu and 6.57 for LysLys genotypes. This is the first report of this highly significant association of MATP polymorphisms with normal human pigmentation variation.

Additional References

A genome-wide association study identifies novel alleles associated with hair color and skin pigmentation. (2008) (https://pubmed.ncbi.nlm.nih.gov/18483556)
OPRM1 and EGFR contribute to skin pigmentation differences between Indigenous Americans and Europeans. (2012) (https://pubmed.ncbi.nlm.nih.gov/22198722)
Analysis of cultured human melanocytes based on polymorphisms within the SLC45A2/MATP, SLC24A5/NCKX5, and OCA2/P loci. (2009) (https://pubmed.ncbi.nlm.nih.gov/18650849)

RELATED GEPHE

Related Genes

14 (Agouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, Kit ligand, MC1R, MFSD12, Oca2, OPRM1, SLC24A5 (NCKX5), 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