GEPHE SUMMARY

Gephebase Gene
tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^#gephebase-summary-title)

Entry Status
Published

GepheID
GP00001157

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
Tyrp1

Synonyms
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1

String
10090.ENSMUSP00000006151 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)

Sequence Similarities
Belongs to the tyrosinase family.

GO - Molecular Function
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0046982 : protein heterodimerization activity

UniProtKB Mus musculus
P07147 (http://www.uniprot.org/uniprot/P07147)

GenebankID or UniProtKB
AAC15468 (https://www.ncbi.nlm.nih.gov/nuccore/AAC15468)

(https://www.ebi.ac.uk/QuickGO/term/GO:0046982)
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0004497 : monooxygenase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004497)

GO - Biological Process

GO:0032438 : melanosome organization
(https://www.ebi.ac.uk/QuickGO/term/GO:0032438)
GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0048023 : positive regulation of melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0048023)
GO:0006583 : melanin biosynthetic process from tyrosine
(https://www.ebi.ac.uk/QuickGO/term/GO:0006583)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0043438 : acetoacetic acid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043438)
GO:0006582 : melanin metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006582)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0030669 : clathrin-coated endocytic vesicle membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0030669)
GO:0010008 : endosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0010008)
GO:0042470 : melanosome (https://www.ebi.ac.uk/QuickGO/term/GO:0042470)
GO:0033162 : melanosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0033162)

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

Sequences associated with human iris pigmentation. (2003) (https://pubmed.ncbi.nlm.nih.gov/14704187)

Authors

Frudakis T; Thomas M; Gaskin Z; Venkateswarlu K; Chandra KS; Ginjupalli S; Gunturi S; Natrajan S; Ponnuswamy VK; Ponnuswamy KN

Abstract

To determine whether and how common polymorphisms are associated with natural distributions of iris colors, we surveyed 851 individuals of mainly European descent at 335 SNP loci in 13 pigmentation genes and 419 other SNPs distributed throughout the genome and known or thought to be informative for certain elements of population structure. We identified numerous SNPs, haplotypes, and diplotypes (diploid pairs of haplotypes) within the OCA2, MYO5A, TYRP1, AIM, DCT, and TYR genes and the CYP1A2-15q22-ter, CYP1B1-2p21, CYP2C8-10q23, CYP2C9-10q24, and MAOA-Xp11.4 regions as significantly associated with iris colors. Half of the associated SNPs were located on chromosome 15, which corresponds with results that others have previously obtained from linkage analysis. We identified 5 additional genes (ASIP, MC1R, POMC, and SILV) and one additional region (GSTT2-22q11.23) with haplotype and/or diplotypes, but not individual SNP alleles associated with iris colors. For most of the genes, multilocus gene-wise genotype sequences were more strongly associated with iris colors than were haplotypes or SNP alleles. Diplotypes for these genes explain 15% of iris color variation. Apart from representing the first comprehensive candidate gene study for variable iris pigmentation and constituting a first step toward developing a classification model for the inference of iris color from DNA, our results suggest that cryptic population structure might serve as a leverage tool for complex trait gene mapping if genomes are screened with the appropriate ancestry informative markers.

Additional References

Two newly identified genetic determinants of pigmentation in Europeans. (2008) (https://pubmed.ncbi.nlm.nih.gov/18488028)

RELATED GEPHE

Related Genes

14 (Agouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, Kit ligand, MC1R, MFSD12, Oca2, OPRM1, SLC24A5 (NCKX5), SLC45A2=MATP, TPCN2, tyrosinase (TYR))
(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=^tyrosinase-related protein 1 (TYRP1)^/and+Taxon ID=^9606^/or+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^/and+Taxon ID=^9606^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS