GEPHE SUMMARY

Gephebase Gene
tyrosinase-related protein 1 (TYRP1)

GephelD
GP00001157

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (eyes; hair; skin)

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Homo sapiens

Ancestral State
Data not curated

Taxonomic Status
Intraspecific

Taxon A

Latin Name
Homo sapiens

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)

NCBI Taxonomy ID
9606

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Homo sapiens

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)

NCBI Taxonomy 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

Sequence Similarities
Belongs to the tyrosinase family.

GO - Molecular Function
GO:0042803 : protein homodimerization activity
GO:0046982 : protein heterodimerization activity
GO:0046872 : metal ion binding
GO:0004497 : monooxygenase activity

GO - Biological Process
GO:0032438 : melanosome organization
GO:0043473 : pigmentation

UniProtKB Mus musculus
P07147

GenebankID or UniProtKB
AAC15468

GO:0048023 : positive regulation of melanin biosynthetic process
GO:0006583 : melanin biosynthetic process from tyrosine
GO:0030318 : melanocyte differentiation
GO:0043438 : acetoacetic acid metabolic process
GO:0006582 : melanin metabolic process

GO - Cellular Component
GO:0016021 : integral component of membrane
GO:0030669 : clathrin-coated endocytic vesicle membrane
GO:0010008 : endosome membrane
GO:0042470 : melanosome
GO:0033162 : melanosome membrane

Presumptive Null
Unknown

Molecular Type
Unknown

Aberration Type
Unknown

Molecular Details of the Mutation
unknown

Experimental Evidence
Association Mapping

Main Reference
Sequences associated with human iris pigmentation. (2003)

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)

RELATED GEPHE

Related Genes
14 (Agouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), HERC2, IRF4, Kit ligand, MC1R, Oca2, OPRM1, SLC24A5 (NCKX5), SLC45A2=MATP, TPCN2, tyrosinase (TYR))
Related Haplotypes
1

EXTERNAL LINKS

COMMENTS