GEPHE SUMMARY

Gephebase Gene

TPCN2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^TPCN2^#gephebase-summary-title)

GepheID

GP00001131

Entry Status

Published

Main curator

Courtier

PHENOTYPIC CHANGE

Trait Category

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait

Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#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

TPCN2

Synonyms

TPC2; SHEP10

String

9606.ENSP00000294309 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000294309)

Sequence Similarities

Belongs to the calcium channel alpha-1 subunit (TC 1.A.1.11) family. Two pore calcium channel subfamily.

GO - Molecular Function

GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)
GO:0019901 : protein kinase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019901)

UniProtKB Homo sapiens

Q8NHX9 (http://www.uniprot.org/uniprot/Q8NHX9)

GenebankID or UniProtKB

AY029200 (https://www.ncbi.nlm.nih.gov/nuccore/AY029200)

GO:0072345 : NAADP-sensitive calcium-release channel activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0072345)
GO:0005245 : voltage-gated calcium channel activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0005245)

GO - Biological Process

GO:0006874 : cellular calcium ion homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0006874)
GO:0034220 : ion transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0034220)
GO:0019722 : calcium-mediated signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0019722)
GO:0034765 : regulation of ion transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0034765)
GO:0007040 : lysosome organization
(https://www.ebi.ac.uk/QuickGO/term/GO:0007040)
GO:0010506 : regulation of autophagy
(https://www.ebi.ac.uk/QuickGO/term/GO:0010506)
GO:0033280 : response to vitamin D (https://www.ebi.ac.uk/QuickGO/term/GO:0033280)
GO:0006939 : smooth muscle contraction
(https://www.ebi.ac.uk/QuickGO/term/GO:0006939)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0010008 : endosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0010008)
GO:0005764 : lysosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005764)
GO:0005765 : lysosomal membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005765)

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

Met484Leu and/or Gly734Gln

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

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

Authors

Sulem P; Gudbjartsson DF; Stacey SN; Helgason A; Rafnar T; Jakobsdottir M; Steinberg S; Gudjonsson SA; Palsson A; Thorleifsson G; PÃilsson S; Sigurgeirsson B; Thorisdottir K; Ragnarsson R; Benediktsdottir KR; Aben KK; Vermeulen SH; Goldstein AM; Tucker MA; Kiemeney LA; Olafsson JH; Gulcher J; Kong A; Thorsteinsdottir U; Stefansson K

Abstract

We present results from a genome-wide association study for variants associated with human pigmentation characteristics among 5,130 Icelanders, with follow-up analyses in 2,116 Icelanders and 1,214 Dutch individuals. Two coding variants in TPCN2 are associated with hair color, and a variant at the ASIP locus shows strong association with skin sensitivity to sun, freckling and red hair, phenotypic characteristics similar to those affected by well-known mutations in MC1R.

Additional References

RELATED GEPHE

Related Genes

14 (Agouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, Kit ligand, MC1R, MFSD12, Oca2, OPRM1, SLC24A5 (NCKX5), SLC45A2=MATP, 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