GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00001134

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

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

Trait State in Taxon A

Homo sapiens - straight hair

Trait State in Taxon B

Homo sapiens - curly/wavy hair

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

TCHH

Synonyms

THH; THL; TRHY; UHS3

String

9606.ENSP00000357794 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000357794)

Sequence Similarities

Belongs to the S100-fused protein family.

GO - Molecular Function

GO:0005509 : calcium ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005509)
GO:0046914 : transition metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046914)

GO - Biological Process

UniProtKB Homo sapiens

Q07283 (http://www.uniprot.org/uniprot/Q07283)

GenebankID or UniProtKB

L09190 (https://www.ncbi.nlm.nih.gov/nuccore/L09190)

GO:0045109 : intermediate filament organization (https://www.ebi.ac.uk/QuickGO/term/GO:0045109)
GO:0070268 : cornification (https://www.ebi.ac.uk/QuickGO/term/GO:0070268)

GO - Cellular Component

GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0005856 : cytoskeleton (https://www.ebi.ac.uk/QuickGO/term/GO:0005856)
GO:0001533 : cornified envelope (https://www.ebi.ac.uk/QuickGO/term/GO:0001533)

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

Not identified

Experimental Evidence

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

Main Reference

Common variants in the trichohyalin gene are associated with straight hair in Europeans. (2009) (https://pubmed.ncbi.nlm.nih.gov/19896111)

Authors

Medland SE; Nyholt DR; Painter JN; McEvoy BP; McRae AF; Zhu G; Gordon SD; Ferreira MA; Wright MJ; Henders AK; Campbell MJ; Duffy DL; Hansell NK; Macgregor S; Slutske WS; Heath AC; Montgomery GW; Martin NG

Abstract

Hair morphology is highly differentiated between populations and among people of European ancestry. Whereas hair morphology in East Asian populations has been studied extensively, relatively little is known about the genetics of this trait in Europeans. We performed a genome-wide association scan for hair morphology (straight, wavy, curly) in three Australian samples of European descent. All three samples showed evidence of association implicating the Trichohyalin gene (TCHH), which is expressed in the developing inner root sheath of the hair follicle, and explaining approximately 6% of variance (p=1.5x10(-31)). These variants are at their highest frequency in Northern Europeans, paralleling the distribution of the straight-hair EDAR variant in Asian populations.

Additional References

RELATED GEPHE

Related Genes

1 (P2RY5) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Hair type/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS