GEPHE SUMMARY

Gephebase Gene

P2RY5 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^P2RY5^#gephebase-summary-title)

GepheID

GP00001734

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

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

Trait State in Taxon A

Homo sapiens

Trait State in Taxon B

Homo sapiens - woolly hair

Ancestral State

Taxon A

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?

Yes

Taxon B Description

family ARWH18

GENOTYPIC CHANGE

Generic Gene Name

LPAR6

Synonyms

LAH3; P2Y5; ARWH1; HYPT8; P2RY5

String

9606.ENSP00000344353 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000344353)

Sequence Similarities

Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function

GO:0004930 : G protein-coupled receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004930)

UniProtKB Homo sapiens

P43657 (http://www.uniprot.org/uniprot/P43657)

GenebankID or UniProtKB

HQ995530 (https://www.ncbi.nlm.nih.gov/nuccore/HQ995530)

GO - Biological Process

GO:0007186 : G protein-coupled receptor signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0007186)
GO:0001835 : blastocyst hatching (https://www.ebi.ac.uk/QuickGO/term/GO:0001835)
GO:0051482 : positive regulation of cytosolic calcium ion concentration involved in phospholipase C-activating G protein-coupled signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0051482)
GO:0035025 : positive regulation of Rho protein signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0035025)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005887 : integral component of plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0043231 : intracellular membrane-bounded organelle (https://www.ebi.ac.uk/QuickGO/term/GO:0043231)

Presumptive Null

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)

Aberration Type

Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title)

Deletion Size

1-9 bp

Molecular Details of the Mutation

2 deletions - one at position 172-175 delAACT and one at position 177 delG - cause frameshift at codon 58 (PTC +31)

Experimental Evidence

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

Main Reference

Disruption of P2RY5, an orphan G protein-coupled receptor, underlies autosomal recessive woolly hair. (2008) (https://pubmed.ncbi.nlm.nih.gov/18297072)

Authors

Shimomura Y; Wajid M; Ishii Y; Shapiro L; Petukhova L; Gordon D; Christiano AM

Abstract

The genetic determinants of hair texture in humans are largely unknown. Several human syndromes exist in which woolly hair comprises a part of the phenotype; however, simple autosomal recessive inheritance of isolated woolly hair has only rarely been reported. To identify a gene involved in controlling hair texture, we performed genetic linkage analysis in six families of Pakistani origin with autosomal recessive woolly hair (ARWH; OMIM 278150). All six families showed linkage to chromosome 13q14.2-14.3 (Z = 17.97). In all cases, we discovered pathogenic mutations in P2RY5, which encodes a G protein-coupled receptor and is a nested gene residing within intron 17 of the retinoblastoma 1 (RB1) gene. P2RY5 is expressed in both Henle's and Huxley's layers of the inner root sheath of the hair follicle. Our findings indicate that disruption of P2RY5 underlies ARWH and, more broadly, uncover a new gene involved in determining hair texture in humans.

Additional References

RELATED GEPHE

Related Genes

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

Related Haplotypes

4 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^P2RY5^/and+Taxon ID=^9606^/or+Gene Gephebase=^P2RY5^/and+Taxon ID=^9606^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS