GEPHE SUMMARY

Gephebase Gene
P2RY5

GephelD
GP00001734

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Hair type (woolly)

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Homo sapiens - woolly hair

Ancestral State
Taxon A

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?
Yes

Taxon B Description
family ARWH18

GENOTYPIC CHANGE

Generic Gene Name
LPAR6

Synonyms
LAH3; P2Y5; ARWH1; HYPT8; P2RY5

String
9606.ENSP00000344353

Sequence Similarities
Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function
GO:0004930 : G protein-coupled receptor activity

GO - Biological Process
GO:0007186 : G protein-coupled receptor signaling pathway
GO:0001835 : blastocyst hatching
GO:0051482 : positive regulation of cytosolic calcium ion concentration involved in

UniProtKB Homo sapiens
P43657

GenebankID or UniProtKB
HQ995530

phospholipase C-activating G protein-coupled signaling pathway
GO:0035025 : positive regulation of Rho protein signal transduction

GO - Cellular Component
GO:0005886 : plasma membrane
GO:0005887 : integral component of plasma membrane
GO:0043231 : intracellular membrane-bounded organelle

Presumptive Null
Yes

Molecular Type
Coding

Aberration Type
Deletion

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

Main Reference
Disruption of P2RY5, an orphan G protein-coupled receptor, underlies autosomal recessive woolly hair. (2008)

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)
Related Haplotypes
4

COMMENTS