

GEPHE SUMMARY

P2RY5 (<https://www.gephebase.org/search-criteria?/and+Gene>
 Gephebase=[^]P2RY5[^]#gephebase-summary-title) Gephebase Gene GP00000817 GepheID
 Entry Status: Courtier Main curator
 Published

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		Taxon B		
	Latin Name		Latin Name	
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Homo sapiens [^] #gephebase-summary-title)		Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Homo sapiens [^] #gephebase-summary-title)		
	Common Name		Common Name	
human		human		
	Synonyms		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		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		Rank	
species		species		
	Lineage		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		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		Parent	
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)		Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)		
	NCBI Taxonomy ID		NCBI Taxonomy ID	
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)		9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)		
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		Yes		
			Taxon B Description	
			family ARWH2	

GENOTYPIC CHANGE

		Generic Gene Name		
LPAR6			P43657 (http://www.uniprot.org/uniprot/P43657) UniProtKB Homo sapiens	
LAH3; P2Y5; ARWH1; HYPT8; P2RY5		Synonyms	GenebankID or UniProtKB	
9606.ENSPO0000344353 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000344353)			HQ995530 (https://www.ncbi.nlm.nih.gov/nuccore/HQ995530)	
		String		
Belongs to the G-protein coupled receptor 1 family.				
		Sequence Similarities		
		GO - Molecular Function		
GO:0004930 : G protein-coupled receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004930)				

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>)

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes^#gephebase-summary-title)) Presumptive Null
Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding^#gephebase-summary-title)) Molecular Type
Insertion ([https://www.gephebase.org/search-criteria?/and+Aberration Type=~Insertion^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=~Insertion^#gephebase-summary-title)) Aberration Type
1-9 bp Insertion Size
at position 69 insertion of 4bp CATG - causes frameshift at codon 24 (PTC +29) Molecular Details of the Mutation
Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Association Mapping^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Association+Mapping^#gephebase-summary-title)) Experimental Evidence
Disruption of P2RY5, an orphan G protein-coupled receptor, underlies autosomal recessive woolly hair. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18297072>) Main Reference
Shimomura Y; Wajid M; Ishii Y; Shapiro L; Petukhova L; Gordon D; Christiano AM Authors
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. Abstract
Additional References

RELATED GEPHE

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

EXTERNAL LINKS

COMMENTS