

GEPHE SUMMARY

<p>P2RY5 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=[^]P2RY5[^]#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001736</p> <p>Courtier</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=[^]Morphology[^]#gephebase-summary-title)</p>		<p>Trait Category</p>		
<p>Hair type (woolly) (https://www.gephebase.org/search-criteria?/and+Trait=[^]Hair type (woolly)[^]#gephebase-summary-title)</p>		<p>Trait</p>		
<p>Homo sapiens</p>		<p>Trait State in Taxon A</p>		
<p>Homo sapiens - woolly hair</p>		<p>Trait State in Taxon B</p>		
<p>Taxon A</p>		<p>Ancestral State</p>		
<p>Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=[^]Intraspecific[^]#gephebase-summary-title)</p>		<p>Taxonomic Status</p>		
<p>Taxon A</p>	<p>Latin Name</p>	<p>Taxon B</p>	<p>Latin Name</p>	
<p>Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Homo sapiens[^]#gephebase-summary-title)</p>	<p>Homo sapiens</p>	<p>Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Homo sapiens[^]#gephebase-summary-title)</p>	<p>Homo sapiens</p>	
<p>human</p>	<p>Common Name</p>	<p>human</p>	<p>Common Name</p>	
<p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens</p>	<p>Synonyms</p>	<p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens</p>	<p>Synonyms</p>	
<p>species</p>	<p>Rank</p>	<p>species</p>	<p>Rank</p>	
<p>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</p>	<p>Lineage</p>	<p>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</p>	<p>Lineage</p>	
<p>Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p>	<p>Parent</p>	<p>Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p>	<p>Parent</p>	
<p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p>	<p>NCBI Taxonomy ID</p>	<p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p>	<p>NCBI Taxonomy ID</p>	
<p>No</p>	<p>is Taxon A an Intraspecies?</p>	<p>Yes</p>	<p>is Taxon B an Intraspecies?</p>	
			<p>Taxon B Description</p>	
			<p>families ARWH5 and ARWH24</p>	

GENOTYPIC CHANGE

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

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

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

SNP ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP)) Aberration Type

Nonsynonymous SNP Coding Change

562A>T which causes I188F Molecular Details of the Mutation

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

	Taxon A	Taxon B	Position
Codon	-	-	562
Amino-acid	Ile	Phe	188

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>) 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>) Related Haplotypes

EXTERNAL LINKS

COMMENTS

