

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>) Presumptive Null
- Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title>) Molecular Type
- SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP^#gephebase-summary-title>) Aberration Type
- Nonsynonymous SNP Coding Change
- 188A>T which causes D63V Molecular Details of the Mutation
- Association Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Association Mapping^#gephebase-summary-title>) Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Asp	Val	63

- 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

