

## GEPHE SUMMARY

<p>tyrosinase-related protein 1 (TYRP1) (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^tyrosinase-related+protein+1+(TYRP1)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^tyrosinase-related+protein+1+(TYRP1)^#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>GP00001155</p> <p>Martin</p> <p>Entry Status</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

<p>Morphology (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title</a>)</p> <p>Coloration (coat) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat)^#gephebase-summary-title</a>)</p> <p>Homo sapiens - Melanesia - dark hair</p> <p>Homo sapiens - Melanesia -blond hair</p> <p>Taxon A</p> <p>Intraspecific (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title</a>)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Homo sapiens (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>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</p> <p>Rank</p> <p>species</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>Parent</p> <p>Homo () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a>)</p> <p>NCBI Taxonomy ID</p> <p>9606 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a>)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Homo sapiens (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>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</p> <p>Rank</p> <p>species</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>Parent</p> <p>Homo () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a>)</p> <p>NCBI Taxonomy ID</p> <p>9606 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a>)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p>
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## GENOTYPIC CHANGE

<p>Tyrp1</p> <p>b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1</p> <p>10090.ENSMUSP00000006151 (<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151</a>)</p> <p>Belongs to the tyrosinase family.</p> <p>GO:0042803 : protein homodimerization activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042803">https://www.ebi.ac.uk/QuickGO/term/GO:0042803</a>)</p> <p>GO:0046982 : protein heterodimerization activity</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>UniProtKB Mus musculus</p> <p>P07147 (<a href="http://www.uniprot.org/uniprot/P07147">http://www.uniprot.org/uniprot/P07147</a>)</p> <p>GenebankID or UniProtKB</p> <p>AAC15468 (<a href="https://www.ncbi.nlm.nih.gov/nuccore/AAC15468">https://www.ncbi.nlm.nih.gov/nuccore/AAC15468</a>)</p>
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(<https://www.ebi.ac.uk/QuickGO/term/GO:0046982>)  
 GO:0046872 : metal ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0046872>)  
 GO:0004497 : monoxygenase activity  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0004497>)

GO - Biological Process

GO:0032438 : melanosome organization  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)  
 GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)  
 GO:0048023 : positive regulation of melanin biosynthetic process  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)  
 GO:0006583 : melanin biosynthetic process from tyrosine  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0006583>)  
 GO:0030318 : melanocyte differentiation  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)  
 GO:0043438 : acetoacetic acid metabolic process  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0043438>)  
 GO:0006582 : melanin metabolic process  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0006582>)

GO - Cellular Component

GO:0016021 : integral component of membrane  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)  
 GO:0030669 : clathrin-coated endocytic vesicle membrane  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0030669>)  
 GO:0010008 : endosome membrane  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0010008>)  
 GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)  
 GO:0033162 : melanosome membrane  
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0033162>)

Presumptive Null

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title>)

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

R93C

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

Melanesian blond hair is caused by an amino acid change in TYRP1. (2012) (<https://pubmed.ncbi.nlm.nih.gov/22556244>)

Authors

Kenny EE; Timpson NJ; Sikora M; Yee MC; Moreno-Estrada A; Eng C; Huntsman S; Burchard EG; Stoneking M; Bustamante CD; Myles S

Abstract

Naturally blond hair is rare in humans and found almost exclusively in Europe and Oceania. Here, we identify an arginine-to-cysteine change at a highly conserved residue in tyrosinase-related protein 1 (TYRP1) as a major determinant of blond hair in Solomon Islanders. This missense mutation is predicted to affect catalytic activity of TYRP1 and causes blond hair through a recessive mode of inheritance. The mutation is at a frequency of 26% in the Solomon Islands, is absent outside of Oceania, represents a strong common genetic effect on a complex human phenotype, and highlights the importance of examining genetic associations worldwide.

Additional References

RELATED GEPHE

Related Genes

14 (Agouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, Kit ligand, MC1R, MFSD12, Oca2, OPRM1, SLC24A5 (NCKX5), SLC45A2=MATP, TPCN2, tyrosinase (TYR))  
 (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

1 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^tyrosinase-related protein 1 \(TYRP1\)^/and+Taxon ID=^9606^/or+Gene Gephebase=^tyrosinase-related protein 1 \(TYRP1\)^/and+Taxon ID=^9606^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^/and+Taxon ID=^9606^/or+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^/and+Taxon ID=^9606^#gephebase-summary-title))

EXTERNAL LINKS

