

GEPHE SUMMARY

<p>FGFR2 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+FGFR2+Gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00000314</p> <p>Martin</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>Hair thickness (https://www.gephebase.org/search-criteria?/and+Trait+Hair+thickness+Gephebase-summary-title)</p> <p>Homo sapiens</p> <p>Homo sapiens - curly/wavy hair</p> <p>Data not curated</p> <p>Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Intraspecific+Gephebase-summary-title)</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 (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Homo+sapiens+Gephebase-summary-title)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sapiens; Homo sapeins; Homo sapien; 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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p> <p>NCBI Taxonomy ID</p> <p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p> <p>is Taxon A an Intraspecies?</p> <p>No</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>Common Name</p> <p>human</p> <p>Synonyms</p> <p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sapiens; Homo sapeins; Homo sapien; 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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p> <p>NCBI Taxonomy ID</p> <p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p>
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GENOTYPIC CHANGE

<p>FGFR2</p> <p>BEK; JWS; BBDS; CEK3; CFD1; ECT1; KGFR; TK14; TK25; BFR-1; CD332; K-SAM; KSAM</p> <p>9606.ENSPO0000410294 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000410294)</p> <p>Belongs to the protein kinase superfamily. Tyr protein kinase family. Fibroblast growth factor receptor subfamily.</p> <p>GO:0004888 : transmembrane signaling receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004888)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P21802 (http://www.uniprot.org/uniprot/P21802)</p> <p>L49241 (https://www.ncbi.nlm.nih.gov/nuccore/L49241)</p>	<p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
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GO:0005524 : ATP binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005524>)
GO:0042802 : identical protein binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042802>)
GO:0042803 : protein homodimerization activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042803>)
GO:0008201 : heparin binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0008201>)
GO:0046934 : phosphatidylinositol-4,5-bisphosphate 3-kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046934>)
GO:0005088 : Ras guanyl-nucleotide exchange factor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005088>)
GO:0004714 : transmembrane receptor protein tyrosine kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004714>)
GO:0004713 : protein tyrosine kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004713>)
GO:0017134 : fibroblast growth factor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0017134>)
GO:0005007 : fibroblast growth factor-activated receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005007>)
GO:0016303 : 1-phosphatidylinositol-3-kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016303>)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043066>)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
GO:0030154 : cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030154>)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)
GO:0070374 : positive regulation of ERK1 and ERK2 cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070374>)
GO:0000165 : MAPK cascade (<https://www.ebi.ac.uk/QuickGO/term/GO:0000165>)
GO:0008284 : positive regulation of cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008284>)
GO:0051897 : positive regulation of protein kinase B signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051897>)
GO:0007267 : cell-cell signaling (<https://www.ebi.ac.uk/QuickGO/term/GO:0007267>)
GO:0001701 : in utero embryonic development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001701>)
GO:0035264 : multicellular organism growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035264>)
GO:0050680 : negative regulation of epithelial cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050680>)
GO:0043410 : positive regulation of MAPK cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043410>)
GO:0009887 : animal organ morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009887>)
GO:0045165 : cell fate commitment (<https://www.ebi.ac.uk/QuickGO/term/GO:0045165>)
GO:0048565 : digestive tract development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048565>)
GO:0050679 : positive regulation of epithelial cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050679>)
GO:0032496 : response to lipopolysaccharide
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032496>)
GO:0045787 : positive regulation of cell cycle
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045787>)
GO:0030324 : lung development (<https://www.ebi.ac.uk/QuickGO/term/GO:0030324>)
GO:0009791 : post-embryonic development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009791>)
GO:0060348 : bone development (<https://www.ebi.ac.uk/QuickGO/term/GO:0060348>)
GO:0090263 : positive regulation of canonical Wnt signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090263>)
GO:0006915 : apoptotic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0006915>)
GO:0009880 : embryonic pattern specification
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009880>)
GO:0009968 : negative regulation of signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009968>)
GO:0051781 : positive regulation of cell division
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051781>)
GO:0040014 : regulation of multicellular organism growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0040014>)
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GO:0030282 : bone mineralization (<https://www.ebi.ac.uk/QuickGO/term/GO:0030282>)
GO:0046777 : protein autophosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046777>)
GO:0018108 : peptidyl-tyrosine phosphorylation
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GO:0008543 : fibroblast growth factor receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008543>)

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GO:0060667 : branch elongation involved in salivary gland morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060667>)
GO:0060670 : branching involved in labyrinthine layer morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060670>)
GO:0060442 : branching involved in prostate gland morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060442>)
GO:0060445 : branching involved in salivary gland morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060445>)
GO:0048755 : branching morphogenesis of a nerve
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GO:0060449 : bud elongation involved in lung branching
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(<https://www.ebi.ac.uk/QuickGO/term/GO:0071300>)
GO:0071560 : cellular response to transforming growth factor beta stimulus
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(<https://www.ebi.ac.uk/QuickGO/term/GO:0060484>)
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GO:0060915 : mesenchymal cell differentiation involved in lung development
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GO:0060916 : mesenchymal cell proliferation involved in lung development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060916>)
GO:0048333 : mesodermal cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048333>)
GO:0030901 : midbrain development

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GO:0021769 : orbitofrontal cortex development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0021769>)
GO:0035265 : organ growth (<https://www.ebi.ac.uk/QuickGO/term/GO:0035265>)
GO:0030916 : otic vesicle formation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030916>)
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GO:0060045 : positive regulation of cardiac muscle cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060045>)
GO:0060501 : positive regulation of epithelial cell proliferation involved in lung morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0060501>)
GO:0002053 : positive regulation of mesenchymal cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002053>)
GO:0010518 : positive regulation of phospholipase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010518>)
GO:0048661 : positive regulation of smooth muscle cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048661>)
GO:0030177 : positive regulation of Wnt signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030177>)
GO:0060527 : prostate epithelial cord arborization involved in prostate glandular acinus morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0060527>)
GO:0060523 : prostate epithelial cord elongation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060523>)
GO:0060512 : prostate gland morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060512>)
GO:0021860 : pyramidal neuron development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0021860>)
GO:0060687 : regulation of branching involved in prostate gland morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060687>)
GO:0010453 : regulation of cell fate commitment
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010453>)
GO:0070372 : regulation of ERK1 and ERK2 cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070372>)
GO:0040036 : regulation of fibroblast growth factor receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0040036>)
GO:0060688 : regulation of morphogenesis of a branching structure
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060688>)
GO:0045667 : regulation of osteoblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045667>)
GO:0033688 : regulation of osteoblast proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033688>)
GO:0051150 : regulation of smooth muscle cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051150>)
GO:0008589 : regulation of smoothened signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008589>)
GO:0048608 : reproductive structure development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048608>)
GO:0048705 : skeletal system morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048705>)
GO:0060529 : squamous basal epithelial stem cell differentiation involved in prostate gland acinus development (<https://www.ebi.ac.uk/QuickGO/term/GO:0060529>)
GO:0001657 : ureteric bud development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001657>)
GO:0055010 : ventricular cardiac muscle tissue morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0055010>)
GO:0021847 : ventricular zone neuroblast division
(<https://www.ebi.ac.uk/QuickGO/term/GO:0021847>)
GO:0042060 : wound healing (<https://www.ebi.ac.uk/QuickGO/term/GO:0042060>)

GO - Cellular Component

GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)
GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)
GO:0005887 : integral component of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0043235 : receptor complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0043235>)
GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
GO:0005794 : Golgi apparatus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005794>)
GO:0031410 : cytoplasmic vesicle (<https://www.ebi.ac.uk/QuickGO/term/GO:0031410>)
GO:0043231 : intracellular membrane-bounded organelle
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043231>)
GO:0009986 : cell surface (<https://www.ebi.ac.uk/QuickGO/term/GO:0009986>)

GO:0005938 : cell cortex (<https://www.ebi.ac.uk/QuickGO/term/GO:0005938>)
GO:0060076 : excitatory synapse (<https://www.ebi.ac.uk/QuickGO/term/GO:0060076>)

Unknown (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title>) Presumptive Null
Cis-regulatory (<https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title>) Molecular Type
Unknown (<https://www.gephebase.org/search-criteria?/and+Aberration Type=^Unknown^#gephebase-summary-title>) Aberration Type
Unknown Molecular Details of the Mutation
Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title>) Experimental Evidence
FGFR2 is associated with hair thickness in Asian populations. (2009) (<https://pubmed.ncbi.nlm.nih.gov/19590514>) Main Reference

Fujimoto A; Nishida N; Kimura R; Miyagawa T; Yuliwulandari R; Batubara L; Mustofa MS; Samakkam U; Settheetham-Ishida W; Ishida T; Morishita Y; Tsunoda T; Tokunaga K; Ohashi J
Authors
Abstract

Hair morphology is one of the most differentiated traits among human populations. A previous study has shown that a nonsynonymous single nucleotide polymorphism (SNP) in the EDAR gene, EDAR 1540T/C, is strongly associated with hair thickness in Asian populations. However, the contributions of other genes remain to be elucidated. In this study, 12 SNPs on 10 hair formation-related genes with high differentiation between Asian and other populations were examined to further identify genes associated with hair morphology. A multiple regression analysis adjusted for age, sex, population and the effect of EDAR 1540T/C revealed an SNP in intron 9 of FGFR2, rs4752566, to be significantly associated with hair thickness (cross-sectional area; P-value=0.0052, small diameter; P-value=0.029 and large diameter; P-value=0.0015). In the genomic region containing the FGFR2 gene, rs4752566 was not in strong linkage disequilibrium (LD) with the surrounding SNPs, indicating that the significant association of rs4752566 with the hair thickness is not due to LD with polymorphisms of the other genes. The rs4752566-T allele of FGFR2, associated with thicker hair, was also shown to be associated with higher mRNA level of FGFR2 (P-value=0.0270). These results suggest that the FGFR2 polymorphism affects the variation in hair thickness in Asia through alteration in the expression level of FGFR2.

Additional References

RELATED GEPHE

1 (EDAR) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Hair thickness/and+groupHaplotypes=true#gephebase-summary-title>) Related Genes
No matches found. Related Haplotypes

EXTERNAL LINKS

COMMENTS