

GEPHE SUMMARY

EDAR (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^EDAR^#gephebase-summary-title)	Gephebase Gene	GP00000256	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Trait #1	Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Morphology^#gephebase-summary-title)	Trait
Hair thickness (https://www.gephebase.org/search-criteria?/and+Trait=^Hair thickness^#gephebase-summary-title)	Trait State in Taxon A
Homo sapiens	Trait State in Taxon B
Homo sapiens (Asia)	

Trait #2	Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Morphology^#gephebase-summary-title)	Trait
Ecchine gland density (https://www.gephebase.org/search-criteria?/and+Trait=^Ecchine gland density^#gephebase-summary-title)	Trait State in Taxon A
-	Trait State in Taxon B
-	

Trait #3	Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Morphology^#gephebase-summary-title)	Trait
Ear traits = lobe size / lobe attachment / helix rolling / ear protrusion (https://www.gephebase.org/search-criteria?/and+Trait=^Ear traits = lobe size / lobe attachment / helix rolling / ear protrusion^#gephebase-summary-title)	Trait State in Taxon A
-	Trait State in Taxon B
-	

Trait #4	Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Morphology^#gephebase-summary-title)	Trait
Tooth morphology (shovel-shaped incisors) (https://www.gephebase.org/search-criteria?/and+Trait=^Tooth morphology (shovel-shaped incisors)^#gephebase-summary-title)	Trait State in Taxon A
-	Trait State in Taxon B
-	

	Ancestral State
Taxon A	Taxonomic Status
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=^Intraspecific^#gephebase-summary-title)	

Taxon A	Taxon B
Latin Name Homo sapiens (#gephebase-summary-title) Common Name human 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 Rank species 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 Parent Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605) NCBI Taxonomy ID 9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606) is Taxon A an Intraspecies? No	Latin Name Homo sapiens (#gephebase-summary-title) Common Name human 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 Rank species 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 Parent Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605) NCBI Taxonomy ID 9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606) is Taxon B an Intraspecies? No

GENOTYPIC CHANGE

Generic Gene Name EDAR Synonyms DL; ED3; ED5; ED1R; EDA3; HRM1; EDA1R; ECTD10A; ECTD10B; EDA-A1R String 9606.ENSPO0000258443 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000258443) Sequence Similarities - GO - Molecular Function GO:0004888 : transmembrane signaling receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004888) GO:0038023 : signaling receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0038023) GO - Biological Process GO:0030154 : cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030154) GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473) GO:0010628 : positive regulation of gene expression (https://www.ebi.ac.uk/QuickGO/term/GO:0010628) GO:0001942 : hair follicle development (https://www.ebi.ac.uk/QuickGO/term/GO:0001942) GO:0042475 : odontogenesis of dentin-containing tooth (https://www.ebi.ac.uk/QuickGO/term/GO:0042475) GO:0043123 : positive regulation of I-kappaB kinase/NF-kappaB signaling (https://www.ebi.ac.uk/QuickGO/term/GO:0043123) GO:1901224 : positive regulation of NIK/NF-kappaB signaling (https://www.ebi.ac.uk/QuickGO/term/GO:1901224) GO:0060662 : salivary gland cavitation (https://www.ebi.ac.uk/QuickGO/term/GO:0060662) GO:0006915 : apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006915) GO:0008544 : epidermis development (https://www.ebi.ac.uk/QuickGO/term/GO:0008544) GO:0046330 : positive regulation of JNK cascade (https://www.ebi.ac.uk/QuickGO/term/GO:0046330) GO:0033209 : tumor necrosis factor-mediated signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0033209) 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:0045177 : apical part of cell (https://www.ebi.ac.uk/QuickGO/term/GO:0045177) Presumptive Null No (#gephebase-summary-title)	UniProtKB Homo sapiens Q9UNE0 (http://www.uniprot.org/uniprot/Q9UNE0) GenebankID or UniProtKB AF130988 (https://www.ncbi.nlm.nih.gov/nuccore/AF130988)
---	--

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

p.Val370Ala; functionally validated in mouse

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	Val	Ala	370

Main Reference

A scan for genetic determinants of human hair morphology: EDAR is associated with Asian hair thickness. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18065779>)

Authors

Fujimoto A; Kimura R; Ohashi J; Omi K; Yuliwulandari R; Batubara L; Mustofa MS; Samakkarn U; Settheetham-Ishida W; Ishida T; Morishita Y; Furusawa T; Nakazawa M; Ohtsuka R; Tokunaga K

Abstract

Hair morphology is one of the most differentiated traits among human populations. However, genetic backgrounds of hair morphological differences among populations have not been clarified yet. In addition, little is known about the evolutionary forces that have acted on hair morphology. To identify hair morphology-determining genes, the levels of local genetic differentiation in 170 genes that are related to hair morphogenesis were evaluated by using data from the International HapMap project. Among highly differentiated genes, ectodysplasin A receptor (EDAR) harboring an Asian-specific non-synonymous single nucleotide polymorphism (1540T/C, 370Val/Ala) was identified as a strong candidate. Association studies between genotypes and hair morphology revealed that the Asian-specific 1540C allele is associated with increase in hair thickness. Reporter gene assays suggested that 1540T/C affects the activity of the downstream transcription factor NF-kappaB. It was inferred from geographic distribution of 1540T/C and the long-range haplotype test that 1540C arose after the divergence of Asians from Europeans and its frequency has rapidly increased in East Asian populations. These findings lead us to conclude that EDAR is a major genetic determinant of Asian hair thickness and the 1540C allele spread through Asian populations due to recent positive selection.

Additional References

Modeling recent human evolution in mice by expression of a selected EDAR variant. (2013) (<https://pubmed.ncbi.nlm.nih.gov/23415220>)

A genome-wide association study identifies multiple loci for variation in human ear morphology. (2015) (<https://pubmed.ncbi.nlm.nih.gov/26105758>)

Effects of an Asian-specific nonsynonymous EDAR variant on multiple dental traits. (2012) (<https://pubmed.ncbi.nlm.nih.gov/22648185>)

A common variation in EDAR is a genetic determinant of shovel-shaped incisors. (2009) (<https://pubmed.ncbi.nlm.nih.gov/19804850>)

RELATED GEPHE

Related Genes

1 (FGFR2) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9606^/and+Trait=Hair+thickness/or+Taxon+ID=~9606^/and+Trait=Ecocrine+gland+density/or+Taxon+ID=~9606^/and+Trait=Ear+traits+=+lobe+size+/+lobe+attachment+/+helix+rolling+/+ear+protrusion/or+Taxon+ID=~9606^/and+Trait=Tooth+morphology/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Pleiotropy