

GEPHE SUMMARY

<p>TBX15 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+^TBX15^#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001113</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>Trait Category</p>		
<p>Ear shape (https://www.gephebase.org/search-criteria?/and+Trait+^Ear+shape+^#gephebase-summary-title)</p>		<p>Trait</p>		
<p>Homo sapiens</p>		<p>Trait State in Taxon A</p>		
<p>Homo sapiens - Ear traits = anti-tragus size and antihelix folding</p>		<p>Trait State in Taxon B</p>		
<p>Data not curated</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 (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+^Homo+sapiens+^#gephebase-summary-title)</p>	<p>Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+^Homo+sapiens+^#gephebase-summary-title)</p>	<p>Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+^Homo+sapiens+^#gephebase-summary-title)</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>No</p>	<p>is Taxon B an Intraspecies?</p>	

GENOTYPIC CHANGE

<p>TBX15</p>	<p>Generic Gene Name</p>	<p>Q96SF7 (http://www.uniprot.org/uniprot/Q96SF7)</p>	<p>UniProtKB Homo sapiens</p>
<p>TBX14</p>	<p>Synonyms</p>	<p>NP_689593 (https://www.ncbi.nlm.nih.gov/nuccore/NP_689593)</p>	<p>GenebankID or UniProtKB</p>
<p>-</p>	<p>String</p>		
<p>-</p>	<p>Sequence Similarities</p>		
<p>GO - Molecular Function</p>			
<p>GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)</p>			
<p>GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)</p>			
<p>GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)</p>			

GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding

(<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)

GO:0001078 : proximal promoter DNA-binding transcription repressor activity, RNA polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0001078>)

GO - Biological Process

GO:0048701 : embryonic cranial skeleton morphogenesis

(<https://www.ebi.ac.uk/QuickGO/term/GO:0048701>)

GO - Cellular Component

GO:0070722 : Tle3-Aes complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0070722>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

Tbx15 enhancer variation including rs17023457 T or C alleles that affect CART1 binding on a conserved site

Experimental Evidence

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

Main Reference

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

Authors

Adhikari K; Reales G; Smith AJ; Konka E; Palmén J; Quinto-Sánchez M; Acuña-Alonso V; Jaramillo C; Arias W; Fuentes M; Pizarro M; Barquera Lozano R; MacAn PÁrez G; GÁmez-Valdás J; Villamil-Ramírez H; Hunemeier T; Ramallo V; Silva de Cerqueira CC; Hurtado M; Villegas V; Granja V; Gallo C; Poletti G; Schuler-Faccini L; Salzano FM; Bortolini MC; Canizales-Quinteros S; Rothhammer F; Bedoya G; Calderón R; Rosique J; Cheeseman M; Bhutta MF; Humphries SE; Gonzalez-José R; Headon D; Balding D; Ruiz-Linares A

Abstract

Here we report a genome-wide association study for non-pathological pinna morphology in over 5,000 Latin Americans. We find genome-wide significant association at seven genomic regions affecting: lobe size and attachment, folding of antihelix, helix rolling, ear protrusion and antitragus size (linear regression P values 2×10^{-8} to 3×10^{-14}). Four traits are associated with a functional variant in the Ectodysplasin A receptor (EDAR) gene, a key regulator of embryonic skin appendage development. We confirm expression of Edar in the developing mouse ear and that Edar-deficient mice have an abnormally shaped pinna. Two traits are associated with SNPs in a region overlapping the T-Box Protein 15 (TBX15) gene, a major determinant of mouse skeletal development. Strongest association in this region is observed for SNP rs17023457 located in an evolutionarily conserved binding site for the transcription factor Cartilage paired-class homeoprotein 1 (CART1), and we confirm that rs17023457 alters in vitro binding of CART1.

Additional References

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

Functional Validation: luciferase expression with CART1 binding shift assay