

GEPHE SUMMARY

<p>Patched1 (Ptc1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="Patched1 (Ptc1)"#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00000855</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>Body size (height) (https://www.gephebase.org/search-criteria?/and+Trait=^Body size (height)"#gephebase-summary-title)</p>		<p>Trait</p>		
<p>Homo sapiens</p>		<p>Trait State in Taxon A</p>		
<p>Homo sapiens</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>Taxon B</p>	
<p>Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Homo sapiens"#gephebase-summary-title)</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>human</p>		<p>Common Name</p>	<p>human</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>species</p>		<p>Rank</p>	<p>species</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>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>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>No</p>		<p>is Taxon A an Intraspecies?</p>	<p>No</p>	
<p>No</p>		<p>is Taxon B an Intraspecies?</p>	<p>No</p>	

GENOTYPIC CHANGE

<p>ptch1</p>	<p>Generic Gene Name</p>	<p>Q98864 (http://www.uniprot.org/uniprot/Q98864)</p>	<p>UniProtKB Danio rerio</p>
<p>ptc1</p>	<p>Synonyms</p>	<p>BAF47711 (https://www.ncbi.nlm.nih.gov/nucleotide/BAF47711)</p>	<p>GenebankID or UniProtKB</p>
<p>7955.ENS DARP00000071771 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=7955.ENS DARP00000071771)</p>		<p>String</p>	
<p>Belongs to the patched family.</p>		<p>Sequence Similarities</p>	
<p>GO:0097108 : hedgehog family protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0097108)</p> <p>GO:0008158 : hedgehog receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008158)</p>		<p>GO - Molecular Function</p>	

GO:0005119 : smoothed binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005119>)
GO - Biological Process

GO:0001649 : osteoblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001649>)
GO:0010002 : cardioblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010002>)
GO:0043010 : camera-type eye development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043010>)
GO:0045879 : negative regulation of smoothed signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045879>)
GO:0048635 : negative regulation of muscle organ development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048635>)
GO:0009954 : proximal/distal pattern formation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009954>)
GO:0031290 : retinal ganglion cell axon guidance
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031290>)
GO:0007224 : smoothed signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007224>)

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>)

Presumptive Null
Unknown ([https://www.gephebase.org/search-criteria?/and+Presumptive+Null="+Unknown^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))
Molecular Type
Unknown ([https://www.gephebase.org/search-criteria?/and+Molecular+Type="+Unknown^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=))
Aberration Type
Unknown ([https://www.gephebase.org/search-criteria?/and+Aberration+Type="+Unknown^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=))
Molecular Details of the Mutation
unknown
Experimental Evidence
Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental+Evidence="+Association+Mapping^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=))
Main Reference
Recent progress in the study of the genetics of height. (2011) (<https://pubmed.ncbi.nlm.nih.gov/21340692>)
Authors
Lettre G
Abstract

Adult height is a classic polygenic trait of high narrow-sense heritability ($h^2 = 0.8$). In the late nineteenth to early twentieth century, variation in adult height was used as a model to set the foundation of the fields of statistics and quantitative genetics. More recently, with our increasing knowledge concerning the extent of genetic variation in the human genome, human geneticists have used genome-wide association studies to identify hundreds of loci robustly associated with adult height, providing new insights into human growth and development, and into the architecture of complex human traits. In this review, I highlight the progress made in the last 2 years in understanding how genetic variation controls height variation in humans, including non-Caucasian populations and children.

Additional References

RELATED GEPHE

23 (ADAMTS10, aggrecan, CREBRF, DC-STAMP domain containing 2 (DCST2), DYM, EIF2AK3, FTO, GDF5, GHSR, GPR133, Growth Hormone Receptor (GHR), HMGA2, Insulin-like growth factor receptor 1 (IGF1R), JAZF1, KCNQ1, LCORL, LIN28B, natriuretic peptide precursor type C (NPPC), natriuretic peptide receptor 3 (NPR3), PPAR-delta, TRIP11 (=GMAP-210), SMAD family member 2 (SMAD2), stanniocalcin 2 (STC2)) ([https://www.gephebase.org/search-criteria?/or+TaxonID="+9606^/and+Trait=Body+size/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+TaxonID=))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS