

GEPHE SUMMARY

Gephebase Gene		GepheID
PPAR-delta (<a +ppar-delta+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+PPAR-delta+"#gephebase-summary-title)	GP00000918	
Entry Status		Main curator
Published		

PHENOTYPIC CHANGE

Trait Category			
Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)		Trait	
Body size (height) (<a +body+size+(height)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Body+size+(height)+"#gephebase-summary-title)		Trait State in Taxon A	
Homo sapiens		Trait State in Taxon B	
Homo sapiens		Ancestral State	
Data not curated		Taxonomic Status	
Intraspecific (<a +intraspecific+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Intraspecific+"#gephebase-summary-title)			
Taxon A		Taxon B	
Latin Name		Latin Name	
Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens+"#gephebase-summary-title)		Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens+"#gephebase-summary-title)	
Common Name		Common Name	
human		human	
Synonyms		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		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		Rank	
species		species	
Lineage		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		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		Parent	
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)		Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)		9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

Generic Gene Name		UniProtKB Homo sapiens
PPARD		Q03181 (http://www.uniprot.org/uniprot/Q03181)
Synonyms		GenebankID or UniProtKB
FAAR; NUC1; NUCI; NR1C2; NUCII; PPARB		AF246300 (https://www.ncbi.nlm.nih.gov/nuccore/AF246300)
String		
9606.ENSPP00000310928 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000310928)		
Sequence Similarities		
Belongs to the nuclear hormone receptor family. NR1 subfamily.		
GO - Molecular Function		
GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)		
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)		

GO:000977 : RNA polymerase II regulatory region sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:000977>)
GO:0008134 : transcription factor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008134>)
GO:0008270 : zinc ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0008270>)
GO:0003677 : DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003677>)
GO:0008289 : lipid binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0008289>)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000981>)
GO:0004879 : nuclear receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004879>)
GO:0001227 : DNA-binding transcription repressor activity, RNA polymerase II-specific
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001227>)
GO:0001078 : proximal promoter DNA-binding transcription repressor activity, RNA
polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0001078>)
GO:0051059 : NF-kappaB binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0051059>)
GO:0008144 : drug binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0008144>)
GO:0005504 : fatty acid binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005504>)
GO:0038023 : signaling receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0038023>)
GO:0003707 : steroid hormone receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003707>)
GO:0000976 : transcription regulatory region sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000976>)
GO:0030374 : nuclear receptor transcription coactivator activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030374>)
GO:0001103 : RNA polymerase II repressing transcription factor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001103>)
GO:0070539 : linoleic acid binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0070539>)

GO - Biological Process

GO:0007275 : multicellular organism development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007275>)
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:0006357 : regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006357>)
GO:0030154 : cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030154>)
GO:0006091 : generation of precursor metabolites and energy
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006091>)
GO:0009755 : hormone-mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009755>)
GO:0007507 : heart development (<https://www.ebi.ac.uk/QuickGO/term/GO:0007507>)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)
GO:0045892 : negative regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045892>)
GO:0045893 : positive regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045893>)
GO:0008203 : cholesterol metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008203>)
GO:0050728 : negative regulation of inflammatory response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050728>)
GO:0008283 : cell proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0008283>)
GO:0050680 : negative regulation of epithelial cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050680>)
GO:0006367 : transcription initiation from RNA polymerase II promoter
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006367>)
GO:0014823 : response to activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0014823>)
GO:0048662 : negative regulation of smooth muscle cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048662>)
GO:0030308 : negative regulation of cell growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030308>)
GO:0071456 : cellular response to hypoxia
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071456>)
GO:0006915 : apoptotic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0006915>)
GO:0071222 : cellular response to lipopolysaccharide
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071222>)
GO:0009062 : fatty acid catabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009062>)
GO:0006631 : fatty acid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006631>)
GO:0006629 : lipid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006629>)
GO:0042060 : wound healing (<https://www.ebi.ac.uk/QuickGO/term/GO:0042060>)
GO:0060612 : adipose tissue development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060612>)

GO:0046697 : decidualization (<https://www.ebi.ac.uk/QuickGO/term/GO:0046697>)
GO:0006006 : glucose metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006006>)
GO:0032024 : positive regulation of insulin secretion
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032024>)
GO:0009749 : response to glucose (<https://www.ebi.ac.uk/QuickGO/term/GO:0009749>)
GO:0033993 : response to lipid (<https://www.ebi.ac.uk/QuickGO/term/GO:0033993>)
GO:0014068 : positive regulation of phosphatidylinositol 3-kinase signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014068>)
GO:0045600 : positive regulation of fat cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045600>)
GO:2000288 : positive regulation of myoblast proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000288>)
GO:0045662 : negative regulation of myoblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045662>)
GO:0032966 : negative regulation of collagen biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032966>)
GO:0097755 : positive regulation of blood vessel diameter
(<https://www.ebi.ac.uk/QuickGO/term/GO:0097755>)
GO:0015908 : fatty acid transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015908>)
GO:0010887 : negative regulation of cholesterol storage
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010887>)
GO:1902894 : negative regulation of pri-miRNA transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:1902894>)
GO:0019216 : regulation of lipid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019216>)
GO:0097190 : apoptotic signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0097190>)
GO:0008366 : axon ensheathment (<https://www.ebi.ac.uk/QuickGO/term/GO:0008366>)
GO:0031589 : cell-substrate adhesion (<https://www.ebi.ac.uk/QuickGO/term/GO:0031589>)
GO:0007566 : embryo implantation (<https://www.ebi.ac.uk/QuickGO/term/GO:0007566>)
GO:0006635 : fatty acid beta-oxidation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006635>)
GO:1904659 : glucose transmembrane transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904659>)
GO:0051546 : keratinocyte migration (<https://www.ebi.ac.uk/QuickGO/term/GO:0051546>)
GO:0043616 : keratinocyte proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043616>)
GO:0009299 : mRNA transcription (<https://www.ebi.ac.uk/QuickGO/term/GO:0009299>)
GO:0014912 : negative regulation of smooth muscle cell migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014912>)
GO:0008654 : phospholipid biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008654>)
GO:0045684 : positive regulation of epidermis development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045684>)
GO:0043415 : positive regulation of skeletal muscle tissue regeneration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043415>)
GO:0006029 : proteoglycan metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006029>)
GO:0014842 : regulation of skeletal muscle satellite cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014842>)
GO:0033189 : response to vitamin A (<https://www.ebi.ac.uk/QuickGO/term/GO:0033189>)
GO:0006776 : vitamin A metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006776>)

GO - Cellular Component

GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0090575 : RNA polymerase II transcription factor complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090575>)
GO:0000790 : nuclear chromatin (<https://www.ebi.ac.uk/QuickGO/term/GO:0000790>)

Presumptive Null

Unknown (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title>)

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

unknown

Experimental Evidence

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

Main Reference

Identification, replication, and fine-mapping of Loci associated with adult height in individuals of african ancestry. (2011) (<https://pubmed.ncbi.nlm.nih.gov/21998595/>)

Authors

N'Diaye A; Chen GK; Palmer CD; Ge B; Tayo B; Mathias RA; Ding J; Nalls MA; Adeyemo A; Adoue V; Ambrosone CB; Atwood L; Bandera EV; Becker LC; Berndt SI; Bernstein L; Blot WJ; Boerwinkle E; Britton A; Casey G; Chanock SJ; Demerath E; Deming SL; Diver WR; Fox C; Harris TB; Hernandez DG; Hu JJ; Ingles SA; John EM; Johnson C; Keating B; Kittles RA; Kolonel LN; Kritchevsky SB; Le Marchand L; Lohman K; Liu J; Millikan RC; Murphy A; Musani S; Neslund-Dudas C; North KE; Nyante S; Ogunniyi A; Ostrander EA; Papanicolaou G; Patel S; Pettaway CA; Press MF; Redline S; Rodriguez-Gil JL; Rotimi C; Rybicki BA; Salako B; Schreiner PJ; Signorello LB; Singleton AB; Stanford JL; Stram AH; Stram DO; Strom SS; Suktitipat B; Thun MJ; Witte JS; Yanek LR; Ziegler RG; Zheng W; Zhu X; Zmuda JM; Zonderman AB; Evans MK; Liu Y; Becker DM; Cooper RS; Pastinen T; Henderson BE; Hirschhorn JN; Lettre G;

Adult height is a classic polygenic trait of high heritability (h^2 approximately 0.8). More than 180 single nucleotide polymorphisms (SNPs), identified mostly in populations of European descent, are associated with height. These variants convey modest effects and explain approximately 10% of the variance in height. Discovery efforts in other populations, while limited, have revealed loci for height not previously implicated in individuals of European ancestry. Here, we performed a meta-analysis of genome-wide association (GWA) results for adult height in 20,427 individuals of African ancestry with replication in up to 16,436 African Americans. We found two novel height loci (Xp22-rs12393627, $P=3.4 \times 10^{-12}$) and 2p14-rs4315565, $P=1.2 \times 10^{-8}$). As a group, height associations discovered in European-ancestry samples replicate in individuals of African ancestry ($P=1.7 \times 10^{-4}$ for overall replication). Fine-mapping of the European height loci in African-ancestry individuals showed an enrichment of SNPs that are associated with expression of nearby genes when compared to the index European height SNPs ($P<0.01$). Our results highlight the utility of genetic studies in non-European populations to understand the etiology of complex human diseases and traits.

Additional References

Hundreds of variants clustered in genomic loci and biological pathways affect human height. (2010) (<https://pubmed.ncbi.nlm.nih.gov/20881960>)

RELATED GEPHE

Related Genes

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), Patched1 (Ptc1), TRIP11 (=GMAP-210), SMAD family member 2 (SMAD2), stanniocalcin 2 (STC2)) ([https://www.gephebase.org/search-criteria?/or+Taxon ID="+9606+"/and+Trait=Body size/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS