GEPHE SUMMARY

Gephebase Gene
PPAR-delta (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^PPAR-delta^#gephebase-summary-title)

GepheID
GP00000918

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Body size (height) (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 (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^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 Infraspecies?
No

Taxon B

Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^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 Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
PPARD

Synonyms
FAAR; NUC1; NUCI; NR1C2; NUCII; PPARB

String
9606.ENSP00000310928 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000310928)

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)

UniProtKB Homo sapiens
Q03181 (http://www.uniprot.org/uniprot/Q03181)

GenebankID or UniProtKB
AF246300 (https://www.ncbi.nlm.nih.gov/nuccore/AF246300)

GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000977)
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;

Haiman CA

Abstract

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 approximately10% 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, Pâ€Š=â€Š3.4Ã—10(-12) and 2p14-rs4315565, Pâ€Š=â€Š1.2Ã—10(-8)). As a group, height associations discovered in European-ancestry samples replicate in individuals of African ancestry (Pâ€Š=â€Š1.7Ã—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, agrecan, 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)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS