GEPHE SUMMARY

Gephebase Gene

Insulin-like growth factor receptor 1 (IGF1R) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Insulin-like growth factor receptor 1 (IGF1R)^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00000503

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

IGF1R

Synonyms

IGFR; CD221; IGFIR; JTK13

String

9606.ENSP00000268035 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000268035)

Sequence Similarities

Belongs to the protein kinase superfamily. Tyr protein kinase family. Insulin receptor subfamily.

GO - Molecular Function

GO:0004888 : transmembrane signaling receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004888)

UniProtKB Homo sapiens

P08069 (http://www.uniprot.org/uniprot/P08069)

GenebankID or UniProtKB

AB425196 (https://www.ncbi.nlm.nih.gov/nuccore/AB425196)

GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)
GO:0042802 : identical protein binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0042802)
GO:0004714 : transmembrane receptor protein tyrosine kinase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004714)
GO:0004713 : protein tyrosine kinase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004713)
GO:0043559 : insulin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043559)
GO:0043560 : insulin receptor substrate binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0043560)
GO:0005009 : insulin-activated receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0005009)
GO:0043548 : phosphatidylinositol 3-kinase binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0043548)
GO:0005158 : insulin receptor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0005158)
GO:0005520 : insulin-like growth factor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0005520)
GO:0031994 : insulin-like growth factor I binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0031994)
GO:0005010 : insulin-like growth factor-activated receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0005010)

GO - Biological Process

GO:0006955 : immune response (https://www.ebi.ac.uk/QuickGO/term/GO:0006955)
GO:0007165 : signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007165)
GO:0043066 : negative regulation of apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0030154 : cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030154)
GO:0070374 : positive regulation of ERK1 and ERK2 cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0070374)
GO:0008284 : positive regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0008284)
GO:0009968 : negative regulation of signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0009968)
GO:0008286 : insulin receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0008286)
GO:0048009 : insulin-like growth factor receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0048009)
GO:0046777 : protein autophosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0046777)
GO:0120162 : positive regulation of cold-induced thermogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0120162)
GO:0030335 : positive regulation of cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0030335)
GO:0042593 : glucose homeostasis (https://www.ebi.ac.uk/QuickGO/term/GO:0042593)
GO:1904646 : cellular response to amyloid-beta
(https://www.ebi.ac.uk/QuickGO/term/GO:1904646)
GO:0014065 : phosphatidylinositol 3-kinase signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0014065)
GO:0048015 : phosphatidylinositol-mediated signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0048015)
GO:0097242 : amyloid-beta clearance
(https://www.ebi.ac.uk/QuickGO/term/GO:0097242)
GO:0097062 : dendritic spine maintenance
(https://www.ebi.ac.uk/QuickGO/term/GO:0097062)
GO:0051389 : inactivation of MAPKK activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0051389)
GO:0038083 : peptidyl-tyrosine autophosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0038083)
GO:0043243 : positive regulation of protein complex disassembly
(https://www.ebi.ac.uk/QuickGO/term/GO:0043243)
GO:0051262 : protein tetramerization (https://www.ebi.ac.uk/QuickGO/term/GO:0051262)
GO:0046328 : regulation of JNK cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0046328)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0016020 : membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)
GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0043235 : receptor complex (https://www.ebi.ac.uk/QuickGO/term/GO:0043235)
GO:0030424 : axon (https://www.ebi.ac.uk/QuickGO/term/GO:0030424)
GO:0043231 : intracellular membrane-bounded organelle
(https://www.ebi.ac.uk/QuickGO/term/GO:0043231)
GO:0005899 : insulin receptor complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0005899)
GO:0035867 : alphav-beta3 integrin-IGF-1-IGF1R complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0035867)

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

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

Related Genes

23 (ADAMTS10, agrecan, CREBRF, DC-STAMP domain containing 2 (DCST2), DYM, EIF2AK3, FTO, GDF5, GHSR, GPR133, Growth Hormone Receptor (GHR), HMGA2, JAZF1, KCNQ1, LCORL, LIN28B, natriuretic peptide precursor type C (NPPC), natriuretic peptide receptor 3 (NPR3), Patched1 (Ptc1), PPAR-delta, 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

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Insulin-like growth factor receptor 1 (IGF1R)^/and+Taxon ID=^9606^/or+Gene Gephebase=^Insulin-like growth factor receptor 1 (IGF1R)^/and+Taxon ID=^9606^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS