

GEPHE SUMMARY

	Gephebase Gene	GepheID
Insulin-like growth factor receptor 1 (IGF1R) (<a +insulin-like+growth+factor+receptor+1+(igf1r)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Insulin-like+growth+factor+receptor+1+(IGF1R)^#gephebase-summary-title)	GP00000503	
	Martin	Main curator
Published	Entry Status	

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
IGF1R		P08069 (http://www.uniprot.org/uniprot/P08069)
	Synonyms	GenebankID or UniProtKB
IGFR; CD221; IGFIR; JTK13		AB425196 (https://www.ncbi.nlm.nih.gov/nucleotide/AB425196)
	String	
9606.ENSPO0000268035 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000268035)		
	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)		

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>)

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

EXTERNAL LINKS

COMMENTS