

GEPHE SUMMARY

GDF5 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~GDF5^#gephebase-summary-title)	Gephebase Gene	GP00000387	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)		Trait Category	
Body size (height) (https://www.gephebase.org/search-criteria?/and+Trait=~Body size (height)^#gephebase-summary-title)		Trait	
Homo sapiens		Trait State in Taxon A	
Homo sapiens		Trait State in Taxon B	
Data not curated		Ancestral State	
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Homo sapiens^#gephebase-summary-title)		Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Homo sapiens^#gephebase-summary-title)	
human		human	
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	
species		species	
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	
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)	
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)	
No		No	

GENOTYPIC CHANGE

GDF5		Generic Gene Name	
OS5; LAP4; BDA1C; BMP14; CDMP1; LAP-4; SYM1B; SYNS2; BMP-14		Synonyms	
9606.ENSPP00000363489 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000363489)		String	
Belongs to the TGF-beta family.		Sequence Similarities	
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)		GO - Molecular Function	
GO:0005125 : cytokine activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005125)		UniProtKB Homo sapiens	
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)		GenebankID or UniProtKB	

GO:0005160 : transforming growth factor beta receptor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005160>)
GO:0036122 : BMP binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0036122>)
GO - Biological Process

GO:0030326 : embryonic limb morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030326>)
GO:0043524 : negative regulation of neuron apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043524>)
GO:0007267 : cell-cell signaling (<https://www.ebi.ac.uk/QuickGO/term/GO:0007267>)
GO:0050680 : negative regulation of epithelial cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050680>)
GO:0030509 : BMP signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030509>)
GO:0048468 : cell development (<https://www.ebi.ac.uk/QuickGO/term/GO:0048468>)
GO:0010862 : positive regulation of pathway-restricted SMAD protein phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010862>)
GO:0042981 : regulation of apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042981>)
GO:0043408 : regulation of MAPK cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043408>)
GO:0060395 : SMAD protein signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060395>)
GO:0030513 : positive regulation of BMP signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030513>)
GO:0032332 : positive regulation of chondrocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032332>)
GO:0045666 : positive regulation of neuron differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045666>)
GO:0040014 : regulation of multicellular organism growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0040014>)
GO:0060591 : chondroblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060591>)
GO:0002062 : chondrocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002062>)
GO:0035136 : forelimb morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035136>)
GO:0035137 : hindlimb morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035137>)
GO:0032331 : negative regulation of chondrocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032331>)
GO:2001054 : negative regulation of mesenchymal cell apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:2001054>)
GO:0043932 : ossification involved in bone remodeling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043932>)
GO:0060390 : regulation of SMAD protein signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060390>)
GO:0009612 : response to mechanical stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009612>)
GO:0007179 : transforming growth factor beta receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007179>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
GO:0005615 : extracellular space (<https://www.ebi.ac.uk/QuickGO/term/GO:0005615>)

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Unknown~#gephebase-summary-title)	Presumptive Null
	Molecular Type
	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)	Main Reference
Common variants in the GDF5-UQCC region are associated with variation in human height. (2008) (https://pubmed.ncbi.nlm.nih.gov/18193045)	
Authors	
Sanna S; Jackson AU; Nagaraja R; Willer CJ; Chen WM; Bonnycastle LL; Shen H; Timpson N; Lettre G; Usala G; Chines PS; Stringham HM; Scott LJ; Dei M; Lai S; Albai G; Crisponi L; Naitza S; Doheny KF; Pugh EW; Ben-Shlomo Y; Ebrahim S; Lawlor DA; Bergman RN; Watanabe RM; Uda M; Tuomilehto J; Coresh J; Hirschhorn JN; Shuldiner AR; Schlessinger D; Collins FS; Davey Smith G; Boerwinkle E; Cao A; Boehnke M; Abecasis GR; Mohlke KL	
Abstract	
Identifying genetic variants that influence human height will advance our understanding of skeletal growth and development. Several rare genetic variants have been convincingly and reproducibly associated with height in mendelian syndromes, and common variants in the transcription factor gene HMGA2 are associated with variation in height in the general population. Here we report genome-wide association analyses, using genotyped and imputed markers, of 6,669 individuals from Finland and Sardinia, and follow-up analyses in an additional 28,801 individuals. We show that common variants in the osteoarthritis-associated locus GDF5-UQCC contribute to variation in height with an estimated additive effect of 0.44 cm (overall P < 10 ⁻¹⁵). Our results indicate that there may be a link between the genetic basis of height and osteoarthritis, potentially mediated through alterations in bone growth and development.	
Recent progress in the study of the genetics of height. (2011) (https://pubmed.ncbi.nlm.nih.gov/21340692)	
Additional References	

RELATED GEPHE

Related Genes

23 (ADAMTS10, aggrecan, CREBRF, DC-STAMP domain containing 2 (DCST2), DYM, EIF2AK3, FTO, 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), 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

No matches found.

EXTERNAL LINKS

COMMENTS