

GEPHE SUMMARY

JAZF1 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~JAZF1^#gephebase-summary-title)	Gephebase Gene	GP00000507	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 - Northern European		Trait State in Taxon A	
Homo sapiens - Northern European		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

JAZF1	Generic Gene Name	Q86VZ6 (http://www.uniprot.org/uniprot/Q86VZ6)	UniProtKB Homo sapiens
TIP27; ZNF802	Synonyms	BC042441 (https://www.ncbi.nlm.nih.gov/nucore/BC042441)	GenebankID or UniProtKB
9606.ENSPO0000283928 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000283928)	String		
-	Sequence Similarities		
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)	GO - Molecular Function		
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)			
GO:0003714 : transcription corepressor activity			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0003714>)
GO:0003676 : nucleic acid binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003676>)
GO - Biological Process
GO:0000122 : negative regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)
GO:0006629 : lipid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006629>)
GO - Cellular Component
GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0017053 : transcriptional repressor complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0017053>)
GO:0001650 : fibrillar center (<https://www.ebi.ac.uk/QuickGO/term/GO:0001650>)

	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
Common variants in the JAZF1 gene associated with height identified by linkage and genome-wide association analysis. (2009) (https://pubmed.ncbi.nlm.nih.gov/18952825)	
	Authors
Johansson A; Marroni F; Hayward C; Franklin CS; Kirichenko AV; Jonasson I; Hicks AA; Vitart V; Isaacs A; Axenovich T; Campbell S; Dunlop MG; Floyd J; Hastie N; Hofman A; Knott S; Kolcic I; Pichler I; Polasek O; Rivadeneira F; Tenesa A; Uitterlinden AG; Wild SH; Zorkoltseva IV; Meitinger T; Wilson JF; Rudan I; Campbell H; Pattaro C; Pramstaller P; Oostra BA; Wright AF; van Duijn CM; Aulchenko YS; Gyllensten U;	
	Abstract
Genes for height have gained interest for decades, but only recently have candidate genes started to be identified. We have performed linkage analysis and genome-wide association for height in approximately 4000 individuals from five European populations. A total of five chromosomal regions showed suggestive linkage and in one of these regions, two SNPs (rs849140 and rs1635852) were associated with height (nominal P = 7.0 x 10(-8) and P = 9.6 x 10(-7), respectively). In total, five SNPs across the genome showed an association with height that reached the threshold of genome-wide significance (nominal P < 1.6 x 10(-7)). The association with height was replicated for two SNPs (rs1635852 and rs849140) using three independent studies (n = 31 077, n=1268 and n = 5746) with overall meta P-values of 9.4 x 10(-10) and 5.3 x 10(-8). These SNPs are located in the JAZF1 gene, which has recently been associated with type II diabetes, prostate and endometrial cancer. JAZF1 is a transcriptional repressor of NR2C2, which results in low IGF1 serum concentrations, perinatal and early postnatal hypoglycemia and growth retardation when knocked out in mice. Both the linkage and association analyses independently identified the JAZF1 region affecting human height. We have demonstrated, through replication in additional independent populations, the consistency of the effect of the JAZF1 SNPs on height. Since this gene also has a key function in the metabolism of growth, JAZF1 represents one of the strongest candidates influencing human height identified so far.	
	Additional References
Meta-analysis of genome-wide scans for human adult stature identifies novel Loci and associations with measures of skeletal frame size. (2009) (https://pubmed.ncbi.nlm.nih.gov/19343178)	

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), 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