

GEPHE SUMMARY

GPR133 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=GPR133#gephebase-summary-title)	Gephebase Gene	GP00000412	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; male-limited) (https://www.gephebase.org/search-criteria?/and+Trait=Body size (height; male-limited)#gephebase-summary-title)		Trait	
Homo sapiens (5 European cohorts)		Trait State in Taxon A	
Homo sapiens (5 European cohorts)		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

ADGRD1		Generic Gene Name		UniProtKB Homo sapiens
PGR25; GPR133		Synonyms		GenebankID or UniProtKB
9606.ENSPO0000261654 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000261654)		String		
Belongs to the G-protein coupled receptor 2 family. Adhesion G-protein coupled receptor (ADGR) subfamily.		Sequence Similarities		
GO:0004930 : G protein-coupled receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004930)		GO - Molecular Function		
		GO - Biological Process		

GO:0007186 : G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007186>)
GO:0007166 : cell surface receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007166>)
GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007189>)

GO - Cellular Component

GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005887 : integral component of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)

	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
Genetic variation in GPR133 is associated with height: genome wide association study in the self-contained population of Sorbs. (2009) (https://pubmed.ncbi.nlm.nih.gov/19729412)	
	Authors
TÄñjes A; Koriath M; Schleinitz D; Dietrich K; BÄtttcher Y; Rayner NW; Almgren P; Enigk B; Richter O; Rohm S; Fischer-Rosinsky A; Pfeiffer A; Hoffmann K; Krohn K; Aust G; Spranger J; Groop L; BIAÿher M; Kovacs P; Sturmvoll M	
	Abstract
Recently, associations of several common genetic variants with height have been reported in different populations. We attempted to identify further variants associated with adult height in a self-contained population (the Sorbs in Eastern Germany) as discovery set. We performed a genome wide association study (GWAS) (approximately 390,000 genetic polymorphisms, Affymetrix gene arrays) on adult height in 929 Sorbian individuals. Subsequently, the best SNPs (P < 0.001) were taken forward to a meta-analysis together with two independent cohorts [Diabetes Genetics Initiative, British 1958 Birth Cohort, (58BC, publicly available)]. Furthermore, we genotyped our best signal for replication in two additional German cohorts (Leipzig, n = 1044 and Berlin, n = 1728). In the primary Sorbian GWAS, we identified 5 loci with a P-value < 10(-5) and 455 SNPs with P-value < 0.001. In the meta-analysis on those 455 SNPs, only two variants in GPR133 (rs1569019 and rs1976930; in LD with each other) retained a P-value at or below 10(-6) and were associated with height in the three cohorts individually. Upon replication, the SNP rs1569019 showed significant effects on height in the Leipzig cohort (P = 0.004, beta = 1.166) and in 577 men of the Berlin cohort (P = 0.049, beta = 1.127) though not in women. The combined analysis of all five cohorts (n = 6,687) resulted in a P-value of 4.7 x 10(-8) (beta = 0.949). In conclusion, our GWAS suggests novel loci influencing height. In view of the robust replication in five different cohorts, we propose GPR133 to be a novel gene associated with adult height.	
	Additional References

RELATED GEPHE

	Related Genes
23 (ADAMTS10, agrecan, CREBRF, DC-STAMP domain containing 2 (DCST2), DYM, EIF2AK3, FTO, GDF5, GHSR, 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