

GEPHE SUMMARY

	Gephebase Gene		GepheID
natriuretic peptide precursor type C (NPPC) (<a +natriuretic+peptide+precursor+type+c+(nppc)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+natriuretic+peptide+precursor+type+C+(NPPC)+"#gephebase-summary-title)		GP00000719	
	Entry Status	Martin	Main curator
Published			

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 - Northern European		Trait State in Taxon B	
Homo sapiens - Northern European			
	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 Mus musculus
Nppc		Q61839 (http://www.uniprot.org/uniprot/Q61839)	
	Synonyms		GenebankID or UniProtKB
CNP; lbab; Cnp		ADZ36320 (https://www.ncbi.nlm.nih.gov/nuccore/ADZ36320)	
	String		
10090.ENSMUSP00000027449 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000027449)			
	Sequence Similarities		
Belongs to the natriuretic peptide family.			
	GO - Molecular Function		
GO:0005102 : signaling receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005102)			
GO:0005179 : hormone activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005179)			

GO:0051427 : hormone receptor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051427>)
GO:0005184 : neuropeptide hormone activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005184>)

GO - Biological Process

GO:0006457 : protein folding (<https://www.ebi.ac.uk/QuickGO/term/GO:0006457>)
GO:0019934 : cGMP-mediated signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019934>)
GO:0008285 : negative regulation of cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008285>)
GO:0001503 : ossification (<https://www.ebi.ac.uk/QuickGO/term/GO:0001503>)
GO:0045669 : positive regulation of osteoblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045669>)
GO:0009791 : post-embryonic development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009791>)
GO:0010753 : positive regulation of cGMP-mediated signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010753>)
GO:0040014 : regulation of multicellular organism growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0040014>)
GO:0001666 : response to hypoxia (<https://www.ebi.ac.uk/QuickGO/term/GO:0001666>)
GO:0045471 : response to ethanol (<https://www.ebi.ac.uk/QuickGO/term/GO:0045471>)
GO:0048513 : animal organ development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048513>)
GO:0006182 : cGMP biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006182>)
GO:0003418 : growth plate cartilage chondrocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003418>)
GO:0003419 : growth plate cartilage chondrocyte proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003419>)
GO:0032966 : negative regulation of collagen biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032966>)
GO:2000279 : negative regulation of DNA biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000279>)
GO:0051447 : negative regulation of meiotic cell cycle
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051447>)
GO:1900194 : negative regulation of oocyte maturation
(<https://www.ebi.ac.uk/QuickGO/term/GO:1900194>)
GO:0097755 : positive regulation of blood vessel diameter
(<https://www.ebi.ac.uk/QuickGO/term/GO:0097755>)
GO:0007168 : receptor guanylyl cyclase signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007168>)
GO:0048660 : regulation of smooth muscle cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048660>)
GO:0022414 : reproductive process (<https://www.ebi.ac.uk/QuickGO/term/GO:0022414>)

GO - Cellular Component

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
GO:0032991 : protein-containing complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032991>)
GO:0005615 : extracellular space (<https://www.ebi.ac.uk/QuickGO/term/GO:0005615>)
GO:0030141 : secretory granule (<https://www.ebi.ac.uk/QuickGO/term/GO:0030141>)

	Presumptive Null
Unknown (<a +unknown+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Presumptive Null=">https://www.gephebase.org/search-criteria?/and+Presumptive Null="+Unknown+"#gephebase-summary-title)	
Unknown (<a +unknown+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Molecular Type=">https://www.gephebase.org/search-criteria?/and+Molecular Type="+Unknown+"#gephebase-summary-title)	Molecular Type
Unknown (<a +unknown+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Aberration Type=">https://www.gephebase.org/search-criteria?/and+Aberration Type="+Unknown+"#gephebase-summary-title)	Aberration Type
Unknown (<a +unknown+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=">https://www.gephebase.org/search-criteria?/and+Experimental Evidence="+Unknown+"#gephebase-summary-title)	Molecular Details of the Mutation
unknown	
Association Mapping (<a +association="" href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=" mapping+"#gephebase-summary-title"="">https://www.gephebase.org/search-criteria?/and+Experimental Evidence="+Association Mapping+"#gephebase-summary-title)	Experimental Evidence
A genome-wide association study of northwestern Europeans involves the C-type natriuretic peptide signaling pathway in the etiology of human height variation. (2009) (https://pubmed.ncbi.nlm.nih.gov/19570815)	Main Reference
	Authors

Estrada K; Krawczak M; Schreiber S; van Duijn K; Stolk L; van Meurs JB; Liu F; Penninx BW; Smit JH; Vogelzangs N; Hottenga JJ; Willemsen G; de Geus EJ; Lorentzon M; von Eller-Eberstein H; Lips P; Schoor N; Pop V; de Keijzer J; Hofman A; Aulchenko YS; Oostra BA; Ohlsson C; Boomsma DI; Uitterlinden AG; van Duijn CM; Rivadeneira F; Kayser M

Abstract
Northwestern Europeans are among the tallest of human populations. The increase in body height in these people appears to have reached a plateau, suggesting the ubiquitous presence of an optimal environment in which genetic factors may have exerted a particularly strong influence on human growth. Therefore, we performed a genome-wide association study (GWAS) of body height using 2.2 million markers in 10 074 individuals from three Dutch and one German population-based cohorts. Upon genotyping, the 12 most significantly height-associated single nucleotide polymorphisms (SNPs) from this GWAS in 6912 additional individuals of Dutch and Swedish origin, a genetic variant (rs6717918) on chromosome 2q37.1 was found to be associated with height at a genome-wide significance level ($P(\text{combined}) = 3.4 \times 10^{-9}$). Notably, a second SNP (rs6718438) located approximately 450 bp away and in strong LD ($r^2 = 0.77$) with rs6717918 was previously found to be suggestive of a height association in 29 820 individuals of mainly northwestern European ancestry, and the over-expression of a nearby natriuretic peptide precursor type C (NPPC) gene, has been associated with overgrowth and skeletal anomalies. We also found a SNP (rs10472828) located on 5p14 near the natriuretic peptide receptor 3 (NPR3) gene, encoding a receptor of the NPPC ligand, to be associated with body height ($P(\text{combined}) = 2.1 \times 10^{-7}$). Taken together, these results suggest that variation in the C-type natriuretic peptide signaling pathway, involving the NPPC and NPR3 genes, plays an important role in determining human body height.

Additional References

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), JAZF1, KCNQ1, LCORL, LIN28B, 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