GEPHE SUMMARY

Gephebase Gene
natriuretic peptide receptor 3 (NPR3) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^natriuretic peptide receptor 3 (NPR3)^#gephebase-summary-title)

GepheID
GP00000720

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Body size (height) (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 (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name
human

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

Rank
species

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

Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name
human

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

Rank
species

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

Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
NPR3

Synonyms
ATNPR3; K17O22.11; K17O22_11; NPR1-like protein 3; At5g45110

String
3702.AT5G45110.1 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 3702.AT5G45110.1)

Sequence Similarities
-

GO - Molecular Function
GO:1901149 : salicylic acid binding (https://www.ebi.ac.uk/QuickGO/term/GO:1901149)

GO - Biological Process
GO:0009816 : defense response to bacterium, incompatible interaction

UniProtKB Arabidopsis thaliana
Q8L746 (http://www.uniprot.org/uniprot/Q8L746)

GenebankID or UniProtKB
NP_000899 (https://www.ncbi.nlm.nih.gov/nuccore/NP_000899)

(https://www.ebi.ac.uk/QuickGO/term/GO:0009816)
GO:0016567 : protein ubiquitination (https://www.ebi.ac.uk/QuickGO/term/GO:0016567)
GO:0042742 : defense response to bacterium
(https://www.ebi.ac.uk/QuickGO/term/GO:0042742)
GO:0050832 : defense response to fungus
(https://www.ebi.ac.uk/QuickGO/term/GO:0050832)
GO:0009817 : defense response to fungus, incompatible interaction
(https://www.ebi.ac.uk/QuickGO/term/GO:0009817)
GO:0080185 : effector dependent induction by symbiont of host immune response
(https://www.ebi.ac.uk/QuickGO/term/GO:0080185)
GO:2000022 : regulation of jasmonic acid mediated signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:2000022)
GO:0009627 : systemic acquired resistance
(https://www.ebi.ac.uk/QuickGO/term/GO:0009627)

GO - Cellular Component

GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)

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

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)

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(combined) = 3.4 x 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(combined) = 2.1 x 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

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, agrecan, 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 precursor type C (NPPC), 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