GEPHE SUMMARY

Gephebase Gene

DC-STAMP domain containing 2 (DCST2) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^DC-STAMP domain containing 2 (DCST2)^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00001509

Main curator

Prigent

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Humans measured for body height (Birth; infant; adult)

Trait State in Taxon B

Human with increased body size at birth

Ancestral State

Unknown

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

DCST2

Synonyms

-

String

-

Sequence Similarities

-

GO - Molecular Function

-

GO - Biological Process

-

GO - Cellular Component

GO:0016021 : integral component of membrane

UniProtKB Homo sapiens

Q5T1A1 (http://www.uniprot.org/uniprot/Q5T1A1)

GenebankID or UniProtKB

()

(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)

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

SNP rs905938 C allele associated with an increase in birth length but probably not the causative mutation

Experimental Evidence

Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)

Main Reference

A novel common variant in DCST2 is associated with length in early life and height in adulthood. (2015) (https://pubmed.ncbi.nlm.nih.gov/25281659)

Authors

van der Valk RJ; Kreiner-MÃ¸ller E; Kooijman MN; Guxens M; Stergiakouli E; SÃ¤Ã¤f A; Bradfield JP; Geller F; Hayes MG; Cousminer DL; KÃ¶rner A; Thiering E; Curtin JA; Myhre R; Huikari V; Joro R; Kerkhof M; Warrington NM; PitkÃ¤nen N; Ntalla I; Horikoshi M; Veijola R; Freathy RM; Teo YY; Barton SJ; Evans DM; Kemp JP; St Pourcain B; Ring SM; Davey Smith G; BergstrÃ¶m A; Kull I; Hakonarson H; Mentch FD; Bisgaard H; Chawes B; Stokholm J; Waage J; Eriksen P; Sevelsted A; Melbye M; ; van Duijn CM; Medina-Gomez C; Hofman A; de Jongste JC; Taal HR; Uitterlinden AG; ; Armstrong LL; Eriksson J; Palotie A; Bustamante M; Estivill X; Gonzalez JR; Llop S; Kiess W; Mahajan A; Flexeder C; Tiesler CM; Murray CS; Simpson A; Magnus P; Sengpiel V; Hartikainen AL; Keinanen-Kiukaanniemi S; Lewin A; Da Silva Couto Alves A; Blakemore AI; Buxton JL; Kaakinen M; Rodriguez A; Sebert S; Vaarasmaki M; Lakka T; Lindi V; Gehring U; Postma DS; Ang W; Newnham JP; LyytikÃ¤inen LP; Pahkala K; Raitakari OT; Panoutsopoulou K; Zeggini E; Boomsma DI; Groen-Blokhuis M; Ilonen J; Franke L; Hirschhorn JN; Pers TH; Liang L; Huang J; Hocher B; Knip M; Saw SM; Holloway JW; MelÃ©n E; Grant SF; Feenstra B; Lowe WL; WidÃ©n E; Sergeyev E; Grallert H; Custovic A; Jacobsson B; Jarvelin MR; Atalay M; Koppelman GH; Pennell CE; Niinikoski H; Dedoussis GV; Mccarthy MI; Frayling TM; Sunyer J; Timpson NJ; Rivadeneira F; BÃ¸nnelykke K; Jaddoe VW;

Abstract

Common genetic variants have been identified for adult height, but not much is known about the genetics of skeletal growth in early life. To identify common genetic variants that influence fetal skeletal growth, we meta-analyzed 22 genome-wide association studies (Stage 1; N = 28 459). We identified seven independent top single nucleotide polymorphisms (SNPs) (P < 1 Ã— 10(-6)) for birth length, of which three were novel and four were in or near loci known to be associated with adult height (LCORL, PTCH1, GPR126 and HMGA2). The three novel SNPs were followed-up in nine replication studies (Stage 2; N = 11 995), with rs905938 in DC-STAMP domain containing 2 (DCST2) genome-wide significantly associated with birth length in a joint analysis (Stages 1 + 2; Î² = 0.046, SE = 0.008, P = 2.46 Ã— 10(-8), explained variance = 0.05%). Rs905938 was also associated with infant length (N = 28 228; P = 5.54 Ã— 10(-4)) and adult height (N = 127 513; P = 1.45 Ã— 10(-5)). DCST2 is a DC-STAMP-like protein family member and DC-STAMP is an osteoclast cell-fusion regulator. Polygenic scores based on 180 SNPs previously associated with human adult stature explained 0.13% of variance in birth length. The same SNPs explained 2.95% of the variance of infant length. Of the 180 known adult height loci, 11 were genome-wide significantly associated with infant length (SF3B4, LCORL, SPAG17, C6orf173, PTCH1, GDF5, ZNFX1, HHIP, ACAN, HLA locus and HMGA2). This study highlights that common variation in DCST2 influences variation in early growth and adult height.

Additional References

RELATED GEPHE

Related Genes

23 (ADAMTS10, agrecan, CREBRF, 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), 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

DCST2 is a DC-STAMP-like protein family member and DC-STAMP is an important regulator of osteoclast cell-fusion in bone homeostasis. However the 1q22 locus is a complex region harboring multiple interesting genes that could affect birth length. We emphasize that we could not specifically pinpoint the causal gene(s)