

## GEPHE SUMMARY

<p>DC-STAMP domain containing 2 (DCST2) (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^DC-STAMP+domain+containing+2+(DCST2)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^DC-STAMP domain containing 2 (DCST2)^#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001509</p> <p>Prigent</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

<p>Morphology (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title</a>)</p> <p>Body size (<a href="https://www.gephebase.org/search-criteria?/and+Trait=^Body+size^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Body size^#gephebase-summary-title</a>)</p> <p>Humans measured for body height (Birth; infant; adult)</p> <p>Human with increased body size at birth</p> <p>Unknown</p> <p>Intraspecific (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title</a>)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Homo sapiens (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo sapiens^#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>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</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p>Homo () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a>)</p> <p>NCBI Taxonomy ID</p> <p>9606 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a>)</p> <p>is Taxon A an Infrasppecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Homo sapiens (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo sapiens^#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>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</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p>Homo () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a>)</p> <p>NCBI Taxonomy ID</p> <p>9606 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a>)</p> <p>is Taxon B an Infrasppecies?</p> <p>No</p>
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## GENOTYPIC CHANGE

<p>DCST2</p> <p>-</p> <p>-</p> <p>-</p> <p>-</p> <p>-</p> <p>GO:0016021 : integral component of membrane</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> <p>GO - Biological Process</p> <p>GO - Cellular Component</p>	<p>UniProtKB Homo sapiens</p> <p>Q5T1A1 (<a href="http://www.uniprot.org/uniprot/Q5T1A1">http://www.uniprot.org/uniprot/Q5T1A1</a>)</p> <p>0</p> <p>GenebankID or UniProtKB</p>
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(<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-Määttä E; Kooijman MN; Guxens M; Stergiakouli E; Sääf A; Bradfield JP; Geller F; Hayes MG; Cousminer DL; Kärrer A; Thiering E; Curtin JA; Myhre R; Huikari V; Joro R; Kerkhof M; Warrington NM; Pitkäräinen 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; Bergström 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-Kiukkaanniemi S; Lewin A; Da Silva Couto Alves A; Blakemore AL; Buxton JL; Kaakinen M; Rodriguez A; Sebert S; Vaarasmaki M; Lakka T; Lindi V; Gehring U; Postma DS; Ang W; Newnham JP; Lyttikäinen 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; Melan E; Grant SF; Feenstra B; Lowe WL; Widom E; Sergeev 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; Bäcknelykke 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) ( $P < 1 \times 10^{-6}$ ) 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;  $\hat{I}^2 = 0.046$ , SE = 0.008,  $P = 2.46 \times 10^{-8}$ , explained variance = 0.05%). Rs905938 was also associated with infant length (N = 28 228;  $P = 5.54 \times 10^{-4}$ ) and adult height (N = 127 513;  $P = 1.45 \times 10^{-5}$ ). 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, ZNF1, HHIP, ACAN, HLA locus and HMGA2). This study highlights that common variation in DCST2 influences variation in early growth and adult height.

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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)