GEPHE SUMMARY

Gephebase Gene

HMGA2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^HMGA2^#gephebase-summary-title)

GepheID

GP00000486

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

Trait State in Taxon A

Homo sapiens

Trait State in Taxon B

Homo sapiens

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

HMGA2

Synonyms

BABL; LIPO; HMGIC; HMGI-C; STQTL9

String

9606.ENSP00000384026 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000384026)

Sequence Similarities

Belongs to the HMGA family.

GO - Molecular Function

GO:0008134 : transcription factor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008134)
GO:0044212 : transcription regulatory region DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0044212)

UniProtKB Homo sapiens

P52926 (http://www.uniprot.org/uniprot/P52926)

GenebankID or UniProtKB

U28753 (https://www.ncbi.nlm.nih.gov/nuccore/U28753)

GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001077)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000978)
GO:0003680 : AT DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003680)
GO:0001078 : proximal promoter DNA-binding transcription repressor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001078)
GO:0003712 : transcription coregulator activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003712)
GO:0051575 : 5′-deoxyribose-5-phosphate lyase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0051575)
GO:0070742 : C2H2 zinc finger domain binding (https://www.ebi.ac.uk/QuickGO/term/GO:0070742)
GO:0035497 : cAMP response element binding (https://www.ebi.ac.uk/QuickGO/term/GO:0035497)
GO:0008301 : DNA binding, bending (https://www.ebi.ac.uk/QuickGO/term/GO:0008301)
GO:0003906 : DNA-(apurinic or apyrimidinic site) endonuclease activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003906)
GO:0004677 : DNA-dependent protein kinase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004677)
GO:0035501 : MH1 domain binding (https://www.ebi.ac.uk/QuickGO/term/GO:0035501)
GO:0035500 : MH2 domain binding (https://www.ebi.ac.uk/QuickGO/term/GO:0035500)
GO:0031492 : nucleosomal DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031492)
GO:0046332 : SMAD binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046332)

GO - Biological Process

GO:0007275 : multicellular organism development (https://www.ebi.ac.uk/QuickGO/term/GO:0007275)
GO:0009615 : response to virus (https://www.ebi.ac.uk/QuickGO/term/GO:0009615)
GO:0043066 : negative regulation of apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0045944 : positive regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0006355 : regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0006355)
GO:0000122 : negative regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0000122)
GO:0045892 : negative regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0045892)
GO:0045893 : positive regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0051301 : cell division (https://www.ebi.ac.uk/QuickGO/term/GO:0051301)
GO:0010628 : positive regulation of gene expression (https://www.ebi.ac.uk/QuickGO/term/GO:0010628)
GO:2000774 : positive regulation of cellular senescence (https://www.ebi.ac.uk/QuickGO/term/GO:2000774)
GO:0035986 : senescence-associated heterochromatin focus assembly (https://www.ebi.ac.uk/QuickGO/term/GO:0035986)
GO:0006325 : chromatin organization (https://www.ebi.ac.uk/QuickGO/term/GO:0006325)
GO:0048863 : stem cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0048863)
GO:0001837 : epithelial to mesenchymal transition (https://www.ebi.ac.uk/QuickGO/term/GO:0001837)
GO:0048762 : mesenchymal cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0048762)
GO:0048333 : mesodermal cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0048333)
GO:2000648 : positive regulation of stem cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:2000648)
GO:0043065 : positive regulation of apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0043065)
GO:0002062 : chondrocyte differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0002062)
GO:0045444 : fat cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0045444)
GO:0040008 : regulation of growth (https://www.ebi.ac.uk/QuickGO/term/GO:0040008)
GO:0006284 : base-excision repair (https://www.ebi.ac.uk/QuickGO/term/GO:0006284)
GO:0035988 : chondrocyte proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0035988)
GO:0031052 : chromosome breakage (https://www.ebi.ac.uk/QuickGO/term/GO:0031052)
GO:0030261 : chromosome condensation (https://www.ebi.ac.uk/QuickGO/term/GO:0030261)
GO:0042769 : DNA damage response, detection of DNA damage

(https://www.ebi.ac.uk/QuickGO/term/GO:0042769)
GO:0035987 : endodermal cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0035987)
GO:0031507 : heterochromatin assembly
(https://www.ebi.ac.uk/QuickGO/term/GO:0031507)
GO:0035978 : histone H2A-S139 phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0035978)
GO:0003131 : mesodermal-endodermal cell signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0003131)
GO:0007095 : mitotic G2 DNA damage checkpoint
(https://www.ebi.ac.uk/QuickGO/term/GO:0007095)
GO:0043922 : negative regulation by host of viral transcription
(https://www.ebi.ac.uk/QuickGO/term/GO:0043922)
GO:2000773 : negative regulation of cellular senescence
(https://www.ebi.ac.uk/QuickGO/term/GO:2000773)
GO:0043392 : negative regulation of DNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0043392)
GO:2001033 : negative regulation of double-strand break repair via nonhomologous end
joining (https://www.ebi.ac.uk/QuickGO/term/GO:2001033)
GO:0045869 : negative regulation of single stranded viral RNA replication via double
stranded DNA intermediate (https://www.ebi.ac.uk/QuickGO/term/GO:0045869)
GO:0090402 : oncogene-induced cell senescence
(https://www.ebi.ac.uk/QuickGO/term/GO:0090402)
GO:0045766 : positive regulation of angiogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0045766)
GO:0071158 : positive regulation of cell cycle arrest
(https://www.ebi.ac.uk/QuickGO/term/GO:0071158)
GO:0071864 : positive regulation of cell proliferation in bone marrow
(https://www.ebi.ac.uk/QuickGO/term/GO:0071864)
GO:2000685 : positive regulation of cellular response to X-ray
(https://www.ebi.ac.uk/QuickGO/term/GO:2000685)
GO:2001022 : positive regulation of response to DNA damage stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:2001022)
GO:2000679 : positive regulation of transcription regulatory region DNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:2000679)
GO:0010564 : regulation of cell cycle process
(https://www.ebi.ac.uk/QuickGO/term/GO:0010564)
GO:2001038 : regulation of cellular response to drug
(https://www.ebi.ac.uk/QuickGO/term/GO:2001038)
GO:2000036 : regulation of stem cell population maintenance
(https://www.ebi.ac.uk/QuickGO/term/GO:2000036)

GO - Cellular Component

GO:0005654 : nucleoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005654)
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0035985 : senescence-associated heterochromatin focus
(https://www.ebi.ac.uk/QuickGO/term/GO:0035985)
GO:0000228 : nuclear chromosome (https://www.ebi.ac.uk/QuickGO/term/GO:0000228)
GO:0032993 : protein-DNA complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0032993)
GO:0071141 : SMAD protein complex (https://www.ebi.ac.uk/QuickGO/term/GO:0071141)

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 common variant of HMGA2 is associated with adult and childhood height in the general population. (2007) (https://pubmed.ncbi.nlm.nih.gov/17767157)

Authors

Weedon MN; Lettre G; Freathy RM; Lindgren CM; Voight BF; Perry JR; Elliott KS; Hackett R; Guiducci C; Shields B; Zeggini E; Lango H; Lyssenko V; Timpson NJ; Burtt NP; Rayner NW; Saxena R; Ardlie K; Tobias JH; Ness AR; Ring SM; Palmer CN; Morris AD; Peltonen L; Salomaa V; ; ; Davey Smith G; Groop LC; Hattersley AT; McCarthy MI; Hirschhorn JN; Frayling TM

Abstract

Human height is a classic, highly heritable quantitative trait. To begin to identify genetic variants influencing height, we examined genome-wide association data from 4,921 individuals. Common variants in the HMGA2 oncogene, exemplified by rs1042725, were associated with height (P = 4 x 10(-8)). HMGA2 is also a strong biological candidate for height, as rare, severe mutations in this gene alter body size in mice and humans, so we tested rs1042725 in additional samples. We confirmed the association in 19,064 adults from four further studies (P = 3 x 10(-11), overall P = 4 x 10(-16), including the genome-wide association data). We also observed the association in children (P = 1 x 10(-6), N = 6,827) and a tall/short case-control study (P = 4 x 10(-6), N = 3,207). We estimate that rs1042725 explains approximately 0.3% of population variation in height (approximately 0.4 cm increased adult height per C allele). There are few examples of common genetic variants reproducibly associated with human quantitativetraits; these results represent, to our knowledge, the first consistently replicated association with adult and childhood height.

Additional References

HMGA2 is confirmed to be associated with human adult height. (2010) (https://pubmed.ncbi.nlm.nih.gov/19930247)
Association of a high mobility group gene (HMGA2) variant with bone mineral density. (2009) (https://pubmed.ncbi.nlm.nih.gov/19376282)
New loci associated with birth weight identify genetic links between intrauterine growth and adult height and metabolism. (2013) (https://pubmed.ncbi.nlm.nih.gov/23202124)

RELATED GEPHE

Related Genes

23 (ADAMTS10, agrecan, CREBRF, DC-STAMP domain containing 2 (DCST2), DYM, EIF2AK3, FTO, GDF5, GHSR, GPR133, Growth Hormone Receptor (GHR), 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