GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00000537

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

Trait State in Taxon A

Homo sapiens - European

Trait State in Taxon B

Homo sapiens - 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

LCORL

Synonyms

MLR1

String

9606.ENSP00000371661 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000371661)

Sequence Similarities

-

GO - Molecular Function

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 - Biological Process

UniProtKB Homo sapiens

Q8N3X6 (http://www.uniprot.org/uniprot/Q8N3X6)

GenebankID or UniProtKB

BC037322 (https://www.ncbi.nlm.nih.gov/nuccore/BC037322)

GO:0006366 : transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0006366)

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

Not identified

Experimental Evidence

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

Main Reference

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)

Authors

Horikoshi M; Yaghootkar H; Mook-Kanamori DO; Sovio U; Taal HR; Hennig BJ; Bradfield JP; St Pourcain B; Evans DM; Charoen P; Kaakinen M; Cousminer DL; LehtimÃ¤ki T; Kreiner-MÃ¸ller E; Warrington NM; Bustamante M; Feenstra B; Berry DJ; Thiering E; Pfab T; Barton SJ; Shields BM; Kerkhof M; van Leeuwen EM; Fulford AJ; Kutalik Z; Zhao JH; den Hoed M; Mahajan A; Lindi V; Goh LK; Hottenga JJ; Wu Y; Raitakari OT; Harder MN; Meirhaeghe A; Ntalla I; Salem RM; Jameson KA; Zhou K; Monies DM; Lagou V; Kirin M; Heikkinen J; Adair LS; Alkuraya FS; Al-Odaib A; Amouyel P; Andersson EA; Bennett AJ; Blakemore AI; Buxton JL; Dallongeville J; Das S; de Geus EJ; Estivill X; Flexeder C; Froguel P; Geller F; Godfrey KM; Gottrand F; Groves CJ; Hansen T; Hirschhorn JN; Hofman A; Hollegaard MV; Hougaard DM; HyppÃ¶nen E; Inskip HM; Isaacs A; JÃ¸rgensen T; Kanaka-Gantenbein C; Kemp JP; Kiess W; KilpelÃ¤inen TO; Klopp N; Knight BA; Kuzawa CW; McMahon G; Newnham JP; Niinikoski H; Oostra BA; Pedersen L; Postma DS; Ring SM; Rivadeneira F; Robertson NR; Sebert S; Simell O; Slowinski T; Tiesler CM; TÃ¶njes A; Vaag A; Viikari JS; Vink JM; Vissing NH; Wareham NJ; Willemsen G; Witte DR; Zhang H; Zhao J; ; Wilson JF; Stumvoll M; Prentice AM; Meyer BF; Pearson ER; Boreham CA; Cooper C; Gillman MW; Dedoussis GV; Moreno LA; Pedersen O; Saarinen M; Mohlke KL; Boomsma DI; Saw SM; Lakka TA; KÃ¶rner A; Loos RJ; Ong KK; Vollenweider P; van Duijn CM; Koppelman GH; Hattersley AT; Holloway JW; Hocher B; Heinrich J; Power C; Melbye M; Guxens M; Pennell CE; BÃ¸nnelykke K; Bisgaard H; Eriksson JG; WidÃ©n E; Hakonarson H; Uitterlinden AG; Pouta A; Lawlor DA; Smith GD; Frayling TM; McCarthy MI; Grant SF; Jaddoe VW; Jarvelin MR; Timpson NJ; Prokopenko I; Freathy RM;

Abstract

Birth weight within the normal range is associated with a variety of adult-onset diseases, but the mechanisms behind these associations are poorly understood. Previous genome-wide association studies of birth weight identified a variant in the ADCY5 gene associated both with birth weight and type 2 diabetes and a second variant, near CCNL1, with no obvious link to adult traits. In an expanded genome-wide association meta-analysis and follow-up study of birth weight (of up to 69,308 individuals of European descent from 43 studies), we have now extended the number of loci associated at genome-wide significance to 7, accounting for a similar proportion of variance as maternal smoking. Five of the loci are known to be associated with other phenotypes: ADCY5 and CDKAL1 with type 2 diabetes, ADRB1 with adult blood pressure and HMGA2 and LCORL with adult height. Our findings highlight genetic links between fetal growth and postnatal growth and metabolism.

Additional References

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, 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