

GEPHE SUMMARY

LCORL (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~LCORL~#gephebase-summary-title)	Gephebase Gene	GP00000537	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology~#gephebase-summary-title)		Trait Category	
Body size (https://www.gephebase.org/search-criteria?/and+Trait=~Body size~#gephebase-summary-title)		Trait	
Homo sapiens - European		Trait State in Taxon A	
Homo sapiens - European		Trait State in Taxon B	
Data not curated		Ancestral State	
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific~#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Homo sapiens~#gephebase-summary-title)		Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Homo sapiens~#gephebase-summary-title)	
human		human	
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		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	
species		species	
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		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	
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)		Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)		9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	
No		No	

GENOTYPIC CHANGE

LCORL		Generic Gene Name		UniProtKB Homo sapiens
MLR1		Synonyms		GenebankID or UniProtKB
9606.ENSPO0000371661 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000371661)		String		
-		Sequence Similarities		
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)		GO - Molecular Function		
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)		GO - Biological Process		

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>)

Unknown (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Unknown~#gephebase-summary-title>)

Unknown (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Unknown~#gephebase-summary-title>)

Unknown (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~Unknown~#gephebase-summary-title>)

Not identified

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

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>)

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; Lehtimäki T; Kreiner-Miller 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 AL; 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; Hyppönen E; Inskip HM; Isaacs A; Järnsten T; Kanaka-Gantenbein C; Kemp JP; Kiess W; Kilpeläinen 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; Tjønnæs A; Vaag A; Viikari JS; Vink JM; Vissing NH; Wareham NJ; Willemssen 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; Kärner 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; Bønnelykke K; Bisgaard H; Eriksson JG; Widén 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;

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.

RELATED GEPHE

23 (ADAMTS10, aggrecan, 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>)

No matches found.

EXTERNAL LINKS

COMMENTS