GEPHE SUMMARY

Gephebase Gene
SBNO1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SBNO1^#gephebase-summary-title)

GepheID
GP00001023

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

Trait State in Taxon A
Homo sapiens - European ancestry

Trait State in Taxon B
Homo sapiens - European ancestry

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
SBNO1

Synonyms
Sno; MOP3

String
9606.ENSP00000387361 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000387361)

Sequence Similarities
Belongs to the SBNO family.

GO - Molecular Function
-

GO - Biological Process
GO:0006355 : regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0006355)

UniProtKB Homo sapiens
A3KN83 (http://www.uniprot.org/uniprot/A3KN83)

GenebankID or UniProtKB
BC133704 (https://www.ncbi.nlm.nih.gov/nuccore/BC133704)

GO - Cellular Component

GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)

Aberration Type

SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Ser729Asn

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

Common variants at 12q15 and 12q24 are associated with infant head circumference. (2012) (https://pubmed.ncbi.nlm.nih.gov/22504419)

Authors

Taal HR; Pourcain BS; Thiering E; Das S; Mook-Kanamori DO; Warrington NM; Kaakinen M; Kreiner-MÃ¸ller E; Bradfield JP; Freathy RM; Geller F; Guxens M; Cousminer DL; Kerkhof M; Timpson NJ; Ikram MA; Beilin LJ; BÃ¸nnelykke K; Buxton JL; Charoen P; Chawes BLK; Eriksson J; Evans DM; Hofman A; Kemp JP; Kim CE; Klopp N; Lahti J; Lye SJ; McMahon G; Mentch FD; MÃ¼ller M; O'Reilly PF; Prokopenko I; Rivadeneira F; Steegers EAP; Sunyer J; Tiesler C; Yaghootkar H; ; Breteler MMB; Debette S; Fornage M; Gudnason V; Launer LJ; van der Lugt A; Mosley TH; Seshadri S; Smith AV; Vernooij MW; ; Blakemore AI; Chiavacci RM; Feenstra B; Fernandez-Benet J; Grant SFA; Hartikainen AL; van der Heijden AJ; IÃ±iguez C; Lathrop M; McArdle WL; MÃ¸lgaard A; Newnham JP; Palmer LJ; Palotie A; Pouta A; Ring SM; Sovio U; Standl M; Uitterlinden AG; Wichmann HE; Vissing NH; DeCarli C; van Duijn CM; McCarthy MI; Koppelman GH; Estivill X; Hattersley AT; Melbye M; Bisgaard H; Pennell CE; Widen E; Hakonarson H; Smith GD; Heinrich J; Jarvelin MR; ; Jaddoe VWV

Abstract

To identify genetic variants associated with head circumference in infancy, we performed a meta-analysis of seven genome-wide association studies (GWAS) (N = 10,768 individuals of European ancestry enrolled in pregnancy and/or birth cohorts) and followed up three lead signals in six replication studies (combined N = 19,089). rs7980687 on chromosome 12q24 (P = 8.1 Ã— 10(-9)) and rs1042725 on chromosome 12q15 (P = 2.8 Ã— 10(-10)) were robustly associated with head circumference in infancy. Although these loci have previously been associated with adult height, their effects on infant head circumference were largely independent of height (P = 3.8 Ã— 10(-7) for rs7980687 and P = 1.3 Ã— 10(-7) for rs1042725 after adjustment for infant height). A third signal, rs11655470 on chromosome 17q21, showed suggestive evidence of association with head circumference (P = 3.9 Ã— 10(-6)). SNPs correlated to the 17q21 signal have shown genome-wide association with adult intracranial volume, Parkinson's disease and other neurodegenerative diseases, indicating that a common genetic variant in this region might link early brain growth with neurological disease in later life.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS