

GEPHE SUMMARY

GRAMD3 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^GRAMD3^#gephebase-summary-title)	Gephebase Gene	GP00001557	GephelD
Published	Entry Status	Prigent	Main curator

PHENOTYPIC CHANGE

	Trait Category		
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)	Trait		
Body fat distribution (visceral) (https://www.gephebase.org/search-criteria?/and+Trait=^Body+fat+distribution+(visceral)^#gephebase-summary-title)	Trait State in Taxon A		
Women of European & African cohorts	Trait State in Taxon B		
women of European & African cohorts	Ancestral State		
Unknown	Taxonomic Status		
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title)	Common Name	Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title)	Common Name
human	Synonyms	human	Synonyms
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Homo sapiens		human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Homo sapiens	
species	Rank	species	Rank
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	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	Lineage
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	Parent	Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	Parent
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	NCBI Taxonomy ID	9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?		is Taxon B an Infraspecies?
	No		

GENOTYPIC CHANGE

GRAMD2B	Generic Gene Name	UniProtKB Homo sapiens
GRAMD3; NS3TP2	Synonyms	GenebankID or UniProtKB
-	String	
-	Sequence Similarities	
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)	GO - Molecular Function	
-	GO - Biological Process	
GO:0005881 : cytoplasmic microtubule	GO - Cellular Component	

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

A>C in associated SNP

Experimental Evidence

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

Main Reference

Multiethnic genome-wide meta-analysis of ectopic fat depots identifies loci associated with adipocyte development and differentiation. (2017) (<https://pubmed.ncbi.nlm.nih.gov/27918534>)

Authors

Chu AY; Deng X; Fisher VA; Drong A; Zhang Y; Feitosa MF; Liu CT; Weeks O; Choh AC; Duan Q; Dyer TD; Eicher JD; Guo X; Heard-Costa NL; Kacprowski T; Kent JW; Lange LA; Liu X; Lohman K; Lu L; Mahajan A; O'Connell JR; Parihar A; Peralta JM; Smith AV; Zhang Y; Homuth G; Kisselbach AH; Kullberg J; Laqua R; Launer LJ; Nauck M; Olivier M; Peyser PA; Terry JG; Wojcynski MK; Yao J; Bielak LF; Blangero J; Borecki IB; Bowden DW; Carr JJ; Czerwinski SA; Ding J; Friedrich N; Gudnason V; Harris TB; Ingelsson E; Johnson AD; Kardia SL; Langefeld CD; Lind L; Liu Y; Mitchell BD; Morris AP; Mosley TH; Rotter JL; Shuldiner AR; Towne B; Väistö H; Wallaschofski H; Wilson JG; Allison M; Lindgren CM; Goessling W; Cupples LA; Steinhauer ML; Fox CS

Abstract

Variation in body fat distribution contributes to the metabolic sequelae of obesity. The genetic determinants of body fat distribution are poorly understood. The goal of this study was to gain new insights into the underlying genetics of body fat distribution by conducting sample-size-weighted fixed-effects genome-wide association meta-analyses in up to 9,594 women and 8,738 men of European, African, Hispanic and Chinese ancestry, with and without sex stratification, for six traits associated with ectopic fat (hereinafter referred to as ectopic-fat traits). In total, we identified seven new loci associated with ectopic-fat traits (ATXN1, UBE2E2, EBF1, RREB1, GSDMB, GRAMD3 and ENSA; $P < 5 \times 10^{-8}$; false discovery rate < 1%). Functional analysis of these genes showed that loss of function of either Atxn1 or Ube2e2 in primary mouse adipose progenitor cells impaired adipocyte differentiation, suggesting physiological roles for ATXN1 and UBE2E2 in adipogenesis. Future studies are necessary to further explore the mechanisms by which these genes affect adipocyte biology and how their perturbations contribute to systemic metabolic disease.

Additional References

RELATED GEPHE

Related Genes

10 (ATXN1, EBF1, ENSA, FTO, GSDMB, LY86, LYPLAL1, RREB1, TRIB2, UBE2E2) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=^9606^/and+Trait=Body+fat+distribution/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS