

GEPHE SUMMARY

	Gephebase Gene	GephelD
LY86 (https://www.gephebase.org/search-criteria/?and+Gene Gephebase=^LY86^#gephebase-summary-title)	GP00001562	Main curator
Published	Entry Status	Prigent

PHENOTYPIC CHANGE

	Trait Category
Physiology (https://www.gephebase.org/search-criteria/?and+Trait Category=^Physiology^#gephebase-summary-title)	Trait
Body fat distribution (visceral/subcutaneous ratio) (https://www.gephebase.org/search-criteria/?and+Trait=^Body+fat+distribution+(visceral/subcutaneous+ratio)^#gephebase-summary-title)	Trait State in Taxon A
Human of European & African cohorts	Trait State in Taxon B
human 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)		Homo sapiens (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title)	
human	Common Name	human	Common Name
human; man; Homo sapiens Linnaeus, 1758; Homo sapiens; Homo sapiens; Homo sapien; Homo sapien; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Homo sapiens	Synonyms	human; man; Homo sapiens Linnaeus, 1758; Homo sapiens; Homo sapiens; Homo sapien; Homo sapien; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Homo sapiens	Synonyms
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?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

LY86	Generic Gene Name	UniProtKB Homo sapiens
MD1; MD-1; MMD-1; dJ80N2.1	Synonyms	GenebankID or UniProtKB
9606.ENSP00000230568 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000230568)	String	0
-	Sequence Similarities	
-	GO - Molecular Function	
GO:0045087 : innate immune response	GO - Biological Process	

(<https://www.ebi.ac.uk/QuickGO/term/GO:0045087>)
GO:0008283 : cell proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0008283>)
GO:0006954 : inflammatory response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006954>)
GO:0031666 : positive regulation of lipopolysaccharide-mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031666>)
GO:0006915 : apoptotic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0006915>)
GO:0006959 : humoral immune response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006959>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005615 : extracellular space (<https://www.ebi.ac.uk/QuickGO/term/GO:0005615>)

Presumptive Null

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>T 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; Parikh A; Peralta JM; Smith AV; Zhang Y; Hornuth G; Kisseeah 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; Steinbauer 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; $P < 5 \times 10^{-10}$; 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, GRAMD3, GSDMB, 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