

GEPHE SUMMARY

Gephebase Gene	Gephebase ID
UBE2E2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^UBE2E2#gephebase-summary-title)	GP00001553
Entry Status	Main curator
Published	Prigent

PHENOTYPIC CHANGE

Trait Category	
Trait	
Body fat distribution (visceral/subcutaneous ratio) (https://www.gephebase.org/search-criteria/?and+Trait)	
Category= [^] Physiology ^#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
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 sapien; 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 sapien; 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?

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
UBE2E2	Synonyms	GenebankID or UniProtKB
UBCH8	0	
9606.ENSP00000379931 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000379931)	String	
Belongs to the ubiquitin-conjugating enzyme family.	Sequence Similarities	
	GO - Molecular Function	
GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)		
GO:0004842 : ubiquitin-protein transferase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004842)		

GO:0061631 : ubiquitin conjugating enzyme activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0061631>)

GO:0042296 : ISG15 transferase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042296>)

GO - Biological Process

GO:0006974 : cellular response to DNA damage stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006974>)

GO:0070534 : protein K63-linked ubiquitination
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070534>)

GO:0032020 : ISG15-protein conjugation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032020>)

GO:1900087 : positive regulation of G1/S transition of mitotic cell cycle
(<https://www.ebi.ac.uk/QuickGO/term/GO:1900087>)

GO:0070979 : protein K11-linked ubiquitination
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070979>)

GO:0070936 : protein K48-linked ubiquitination
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070936>)

GO - Cellular Component

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

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

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

T>C in associated SNP Molecular Details of the Mutation

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

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; Kisseebah AH; Kullberg J; Laqua R; Launer LJ; Nauck M; Olivier M; Peyser PA; Terry JG; Wojczynski 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 JI; 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; $P < 5 \times 10^{-10}$; 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, LY86, LYPLAL1, RREB1, TRIB2) (<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

Ube2e2 knockdown impaired the formation of lipid-containing adipocytes during ex vivo adipogenesis of subcutaneous adipose tissue progenitors and impaired adipogenesis in progenitor cells isolated from visceral adipose tissue. UBE2E2 was also associated to type 2 diabetes.

