

GEPHE SUMMARY

FTO (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="FTO"#gephebase-summary-title)	Gephebase Gene	GP00001560	GepheID
Published	Entry Status	Prigent	Main curator

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category="Physiology"#gephebase-summary-title)	Trait Category		
Body fat distribution (subcutaneous) (<a (subcutaneous)"#gephebase-summary-title"="" body="" distribution="" fat="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="Body fat distribution (subcutaneous)"#gephebase-summary-title)	Trait		
Human of European & African cohorts	Trait State in Taxon A		
human of European & African cohorts	Trait State in Taxon B		
Unknown	Ancestral State		
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status="Intraspecific"#gephebase-summary-title)	Taxonomic Status		
	Taxon A	Taxon B	
	Latin Name	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)	Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Homo sapiens"#gephebase-summary-title)	
human	Common Name	Common Name	
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	Synonyms	Synonyms	
species	Rank	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	Lineage	
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)	Parent	Parent	
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)	NCBI Taxonomy ID	NCBI Taxonomy ID	
No	is Taxon A an Intraspecies?	is Taxon B an Intraspecies?	

GENOTYPIC CHANGE

FTO	Generic Gene Name	Q9C0B1 (http://www.uniprot.org/uniprot/Q9C0B1)	UniProtKB Homo sapiens
GDFD; ALKBH9; BMIQ14; KIAA1752	Synonyms	0	GenebankID or UniProtKB
9606.ENSPO0000418823 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPO0000418823)	String		
Belongs to the fto family.	Sequence Similarities		
GO:0008198 : ferrous iron binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008198)	GO - Molecular Function		
GO:0043734 : DNA-N1-methyladenine dioxygenase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0043734)			
GO:0035516 : oxidative DNA demethylase activity			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0035516>)
GO:0035515 : oxidative RNA demethylase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035515>)
GO:1990931 : RNA N6-methyladenosine dioxygenase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:1990931>)

GO - Biological Process

GO:0070989 : oxidative demethylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070989>)
GO:0040014 : regulation of multicellular organism growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0040014>)
GO:0060612 : adipose tissue development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060612>)
GO:0006307 : DNA dealkylation involved in DNA repair
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006307>)
GO:0080111 : DNA demethylation (<https://www.ebi.ac.uk/QuickGO/term/GO:0080111>)
GO:0035552 : oxidative single-stranded DNA demethylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035552>)
GO:0035553 : oxidative single-stranded RNA demethylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035553>)
GO:0090335 : regulation of brown fat cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090335>)
GO:0010883 : regulation of lipid storage
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010883>)
GO:0044065 : regulation of respiratory system process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0044065>)
GO:0070350 : regulation of white fat cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070350>)
GO:0042245 : RNA repair (<https://www.ebi.ac.uk/QuickGO/term/GO:0042245>)
GO:0001659 : temperature homeostasis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001659>)

GO - Cellular Component

GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0016607 : nuclear speck (<https://www.ebi.ac.uk/QuickGO/term/GO:0016607>)

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>G 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; Kissebah 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; VÃ¶lzlke H; Wallaschofski H; Wilson JG; Allison M; Lindgren CM; Goessling W; Cupples LA; Steinhauser 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, GRAMD3, 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

1 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^FTO^/and+Taxon ID=^9606^/or+Gene Gephebase=^FTO^/and+Taxon ID=^9606^#gephebase-summary-title>)

EXTERNAL LINKS

