

GEPHE SUMMARY

ATXN1 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^ATXN1^#gephebase-summary-title) Published	Gephebase Gene	GP00001552	GepheID
	Entry Status	Prigent	Main curator

PHENOTYPIC CHANGE

		Trait Category	
Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Physiology^#gephebase-summary-title)		Trait	
Body fat distribution (attenuation) (https://www.gephebase.org/search-criteria?/and+Trait=^Body fat distribution (attenuation)^#gephebase-summary-title)		Trait State in Taxon A	
Men of European & African cohorts		Trait State in Taxon B	
men of European & African cohorts		Ancestral State	
Unknown		Taxonomic Status	
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=^Intraspecific^#gephebase-summary-title)			
Taxon A		Taxon B	
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		human	
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		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	
species		species	
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		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	
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)		Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)		9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	
No		No	

GENOTYPIC CHANGE

		Generic Gene Name	
ATXN1		P54253 (http://www.uniprot.org/uniprot/P54253)	
ATX1; SCA1; D6S504E		0	
9606.ENSPP00000244769 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000244769)			
Belongs to the ATXN1 family.		Sequence Similarities	
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)		GO - Molecular Function	
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)			
GO:0008022 : protein C-terminus binding			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0008022>)
GO:0034046 : poly(G) binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0034046>)
GO:0008266 : poly(U) RNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008266>)
GO:0043621 : protein self-association (<https://www.ebi.ac.uk/QuickGO/term/GO:0043621>)
GO - Biological Process
GO:0000122 : negative regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)
GO:0045892 : negative regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045892>)
GO:0035176 : social behavior (<https://www.ebi.ac.uk/QuickGO/term/GO:0035176>)
GO:0007612 : learning (<https://www.ebi.ac.uk/QuickGO/term/GO:0007612>)
GO:0007420 : brain development (<https://www.ebi.ac.uk/QuickGO/term/GO:0007420>)
GO:0048286 : lung alveolus development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048286>)
GO:0007613 : memory (<https://www.ebi.ac.uk/QuickGO/term/GO:0007613>)
GO:0051168 : nuclear export (<https://www.ebi.ac.uk/QuickGO/term/GO:0051168>)
GO:0006396 : RNA processing (<https://www.ebi.ac.uk/QuickGO/term/GO:0006396>)
GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
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:0005730 : nucleolus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005730>)
GO:0016363 : nuclear matrix (<https://www.ebi.ac.uk/QuickGO/term/GO:0016363>)
GO:0042405 : nuclear inclusion body
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042405>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title)	Presumptive Null
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
	Molecular Details of the Mutation
A>G in associated SNP	
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; 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 Å— 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

10 (EBF1, ENSA, FTO, 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 Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

Atxn1 knockdown impaired the formation of lipid-containing adipocytes during ex vivo adipogenesis of subcutaneous adipose tissue progenitors

