

GEPHE SUMMARY

<p>GSDMB (<a +gsdmb+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+GSDMB+"#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001556</p> <p>Prigent</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

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

<p>GSDMB</p> <p>GSDML; PP4052; PRO2521</p> <p>9606.ENSPO0000353465 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPO0000353465)</p> <p>Belongs to the gasdermin family.</p> <p>GO:0005546 : phosphatidylinositol-4,5-bisphosphate binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005546)</p> <p>GO:0070273 : phosphatidylinositol-4-phosphate binding (https://www.ebi.ac.uk/QuickGO/term/GO:0070273)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>Q8TAX9 (http://www.uniprot.org/uniprot/Q8TAX9)</p> <p>0</p>	<p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
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GO:0001786 : phosphatidylserine binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001786>)

GO - Biological Process

GO:0070269 : pyroptosis (<https://www.ebi.ac.uk/QuickGO/term/GO:0070269>)

GO - Cellular Component

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

GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)

Presumptive Null

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null+No+gpebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null+No+gpebase-summary-title))

Molecular Type

Unknown ([https://www.gephebase.org/search-criteria?/and+Molecular Type+Unknown+gpebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type+Unknown+gpebase-summary-title))

Aberration Type

Unknown ([https://www.gephebase.org/search-criteria?/and+Aberration Type+Unknown+gpebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type+Unknown+gpebase-summary-title))

Molecular Details of the Mutation

T>C in associated SNP

Experimental Evidence

Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence+Association Mapping+gpebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence+Association+Mapping+gpebase-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, FTO, GRAMD3, LY86, LYPLAL1, RREB1, TRIB2, UBE2E2) ([https://www.gephebase.org/search-criteria?/or+Taxon ID+9606+and+Trait=Body fat distribution+and+groupHaplotypes=true#gpebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID+9606+and+Trait+Body+fat+distribution+and+groupHaplotypes=true#gpebase-summary-title))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS