GEPHE SUMMARY

Gephebase Gene
ENSA (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^ENSA^#gephebase-summary-title)

GepheID
GP00001558

Entry Status
Published

Main curator
Prigent

PHENOTYPIC CHANGE

Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait
Body fat distribution (pericardial) (https://www.gephebase.org/search-criteria?/and+Trait=^Body fat distribution (pericardial)^#gephebase-summary-title)

Trait State in Taxon A
Human of European & African & Chinese & Hispanic cohorts

Trait State in Taxon B
human of European & African & Chinese & Hispanic 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 sapian; 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 sapian; 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?
No

GENOTYPIC CHANGE

Generic Gene Name
ENSA

Synonyms
ARPP-19e

String
-

Sequence Similarities
Belongs to the endosulfine family.

GO - Molecular Function
GO:0005102 : signaling receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005102)
GO:0019888 : protein phosphatase regulator activity (https://www.ebi.ac.uk/QuickGO/term/GO:0019888)
GO:0008200 : ion channel inhibitor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008200)

UniProtKB Homo sapiens
O43768 (http://www.uniprot.org/uniprot/O43768)

GenebankID or UniProtKB
()

GO:0019212 : phosphatase inhibitor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0019212)
GO:0019870 : potassium channel inhibitor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0019870)
GO:0051721 : protein phosphatase 2A binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0051721)
GO:0004864 : protein phosphatase inhibitor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004864)

GO - Biological Process

GO:0051301 : cell division (https://www.ebi.ac.uk/QuickGO/term/GO:0051301)
GO:0000278 : mitotic cell cycle (https://www.ebi.ac.uk/QuickGO/term/GO:0000278)
GO:0050796 : regulation of insulin secretion
(https://www.ebi.ac.uk/QuickGO/term/GO:0050796)
GO:0009749 : response to glucose (https://www.ebi.ac.uk/QuickGO/term/GO:0009749)
GO:0007584 : response to nutrient (https://www.ebi.ac.uk/QuickGO/term/GO:0007584)
GO:0000086 : G2/M transition of mitotic cell cycle
(https://www.ebi.ac.uk/QuickGO/term/GO:0000086)
GO:0035308 : negative regulation of protein dephosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0035308)

GO - Cellular Component

GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0005654 : nucleoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005654)

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¶lzke 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 Ã— 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, 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 Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS