## GEPHE SUMMARY

Gephebase Gene

RREB1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^RREB1^#gephebase-summary-title)

GepheID

GP00001555

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

RREB1

Synonyms

HNT; FINB; LZ321; Zep-1; RREB-1

String

-

Sequence Similarities

Belongs to the krueppel C2H2-type zinc-finger protein family.

GO - Molecular Function

GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001228)
GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)
GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000977)

UniProtKB Homo sapiens

Q92766 (http://www.uniprot.org/uniprot/Q92766)

GenebankID or UniProtKB

()

GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000978)

GO - Biological Process

GO:0007275 : multicellular organism development (https://www.ebi.ac.uk/QuickGO/term/GO:0007275)
GO:0045944 : positive regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0006355 : regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0006355)
GO:0000122 : negative regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0000122)
GO:0045893 : positive regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0007265 : Ras protein signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007265)
GO:0006366 : transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0006366)
GO:0010634 : positive regulation of epithelial cell migration (https://www.ebi.ac.uk/QuickGO/term/GO:0010634)
GO:2000394 : positive regulation of lamellipodium morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:2000394)
GO:0033601 : positive regulation of mammary gland epithelial cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0033601)
GO:1900026 : positive regulation of substrate adhesion-dependent cell spreading (https://www.ebi.ac.uk/QuickGO/term/GO:1900026)
GO:1903691 : positive regulation of wound healing, spreading of epidermal cells (https://www.ebi.ac.uk/QuickGO/term/GO:1903691)

GO - Cellular Component

GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0070062 : extracellular exosome (https://www.ebi.ac.uk/QuickGO/term/GO:0070062)
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0016604 : nuclear body (https://www.ebi.ac.uk/QuickGO/term/GO:0016604)
GO:0016607 : nuclear speck (https://www.ebi.ac.uk/QuickGO/term/GO:0016607)
GO:0001650 : fibrillar center (https://www.ebi.ac.uk/QuickGO/term/GO:0001650)

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

C>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, ENSA, FTO, GRAMD3, GSDMB, LY86, LYPLAL1, 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