

CREBRF (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~CREBRF^#gephebase-summary-title)	Gephebase Gene	GP00001444	GepheID
Published	Entry Status	Prigent	Main curator

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title)		Trait Category	
Body size (obesity) (https://www.gephebase.org/search-criteria?/and+Trait=~Body size (obesity)~#gephebase-summary-title)		Trait	
human Samoan not associated with higher BMI		Trait State in Taxon A	
human Samoan associated with higher BMI		Trait State in Taxon B	
Taxon A		Ancestral State	
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=~Intraspecific~#gephebase-summary-title)		Taxonomic Status	
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 sapien; Homo sapiens; 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 sapien; Homo sapiens; 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)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

CREBRF	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	Q8IUR6 (http://www.uniprot.org/uniprot/Q8IUR6)
LRF; C5orf41	String	GenebankID or UniProtKB
9606.ENSPP00000296953 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000296953)	Sequence Similarities	NM_153607 (https://www.ncbi.nlm.nih.gov/nucore/NM_153607)
Belongs to the bZIP family. CREBRF subfamily.	GO - Molecular Function	
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001228)		
GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000977)		

GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000981>)

GO - Biological Process

GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)

GO:0000122 : negative regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)

GO:0042711 : maternal behavior (<https://www.ebi.ac.uk/QuickGO/term/GO:0042711>)

GO:0030968 : endoplasmic reticulum unfolded protein response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030968>)

GO:0034976 : response to endoplasmic reticulum stress
(<https://www.ebi.ac.uk/QuickGO/term/GO:0034976>)

GO:0045732 : positive regulation of protein catabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045732>)

GO:1900102 : negative regulation of endoplasmic reticulum unfolded protein response
(<https://www.ebi.ac.uk/QuickGO/term/GO:1900102>)

GO:1900170 : negative regulation of glucocorticoid mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:1900170>)

GO:0032388 : positive regulation of intracellular transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032388>)

GO:1902213 : positive regulation of prolactin signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:1902213>)

GO:0051222 : positive regulation of protein transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051222>)

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>)

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>)

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title>)

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title>)

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title>)

Nonsynonymous

c.1370G>A p.Arg457Gln

Association Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Association Mapping~#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

A thrifty variant in CREBRF strongly influences body mass index in Samoans. (2016) (<https://pubmed.ncbi.nlm.nih.gov/27455349>)

Minster RL; Hawley NL; Su CT; Sun G; Kershaw EE; Cheng H; Buhule OD; Lin J; Reupena MS; Viali S; Tuitele J; Naseri T; Urban Z; Deka R; Weeks DE; McGarvey ST

Samoans are a unique founder population with a high prevalence of obesity, making them well suited for identifying new genetic contributors to obesity. We conducted a genome-wide association study (GWAS) in 3,072 Samoans, discovered a variant, rs12513649, strongly associated with body mass index (BMI) (P = 5.3 Å— 10(-14)), and replicated the association in 2,102 additional Samoans (P = 1.2 Å— 10(-9)). Targeted sequencing identified a strongly associated missense variant, rs373863828 (p.Arg457Gln), in CREBRF (meta P = 1.4 Å— 10(-20)). Although this variant is extremely rare in other populations, it is common in Samoans (frequency of 0.259), with an effect size much larger than that of any other known common BMI risk variant (1.36-1.45 kg/m(2) per copy of the risk-associated allele). In comparison to wild-type CREBRF, the Arg457Gln variant when overexpressed selectively decreased energy use and increased fat storage in an adipocyte cell model. These data, in combination with evidence of positive selection of the allele encoding p.Arg457Gln, support a ‘thrifty’ variant hypothesis as a factor in human obesity.

RELATED GEPHE

23 (ADAMTS10, aggrecan, DC-STAMP domain containing 2 (DCST2), DYM, EIF2AK3, FTO, GDF5, GHSR, GPR133, Growth Hormone Receptor (GHR), HMG2, Insulin-like growth factor receptor 1 (IGF1R), JAZF1, KCNQ1, LCORL, LIN28B, natriuretic peptide precursor type C (NPPC), natriuretic peptide receptor 3 (NPR3), Patched1 (Ptc1), PPAR-delta, TRIP11 (=GMAP-210), SMAD family member 2 (SMAD2), stanniocalcin 2 (STC2)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~9606~/and+Trait=Body size/and+groupHaplotypes=true#gephebase-summary-title>)

No matches found.

EXTERNAL LINKS

COMMENTS