

GEPHE SUMMARY

	Gephebase Gene		GepheID
FTO (<a +fto+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+FTO+"#gephebase-summary-title)		GP00000378	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology, Physiology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)			
	Trait		
Body size (weight; variance) (<a +body+size+(weight;+variance)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Body+size+(weight;+variance)+"#gephebase-summary-title)			
	Trait State in Taxon A		
Homo sapiens - Europe			
	Trait State in Taxon B		
Homo sapiens - Europe			
	Ancestral State		
Data not curated			
	Taxonomic Status		
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)			
	Taxon A		Taxon B
	Latin Name		Latin Name
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)		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)	
	Common Name		Common Name
human		human	
	Synonyms		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		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		Rank
species		species	
	Lineage		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		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		Parent
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)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
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	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
FTO		Q9C0B1 (http://www.uniprot.org/uniprot/Q9C0B1)	
	Synonyms		GenebankID or UniProtKB
GDFD; ALKBH9; BMIQ14; KIAA1752		NP_001073901 (https://www.ncbi.nlm.nih.gov/nuccore/NP_001073901)	
	String		
9606.ENSPP00000418823 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000418823)			
	Sequence Similarities		
Belongs to the fto family.			
	GO - Molecular Function		
GO:0008198 : ferrous iron binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008198)			
GO:0043734 : DNA-N1-methyladenine dioxygenase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0043734)			
GO:0035516 : oxidative DNA demethylase activity			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0035516>)
GO:0035515 : oxidative RNA demethylase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035515>)
GO:1990931 : RNA N6-methyladenosine dioxygenase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:1990931>)

GO - Biological Process

GO:0070989 : oxidative demethylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070989>)
GO:0040014 : regulation of multicellular organism growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0040014>)
GO:0060612 : adipose tissue development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060612>)
GO:0006307 : DNA dealkylation involved in DNA repair
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006307>)
GO:0080111 : DNA demethylation (<https://www.ebi.ac.uk/QuickGO/term/GO:0080111>)
GO:0035552 : oxidative single-stranded DNA demethylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035552>)
GO:0035553 : oxidative single-stranded RNA demethylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035553>)
GO:0090335 : regulation of brown fat cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090335>)
GO:0010883 : regulation of lipid storage
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010883>)
GO:0044065 : regulation of respiratory system process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0044065>)
GO:0070350 : regulation of white fat cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070350>)
GO:0042245 : RNA repair (<https://www.ebi.ac.uk/QuickGO/term/GO:0042245>)
GO:0001659 : temperature homeostasis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001659>)

GO - Cellular Component

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:0016607 : nuclear speck (<https://www.ebi.ac.uk/QuickGO/term/GO:0016607>)

	Presumptive Null
No (<a +no+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Presumptive Null=">https://www.gephebase.org/search-criteria?/and+Presumptive Null="+No+"#gephebase-summary-title)	
	Molecular Type
Cis-regulatory (<a +cis-regulatory+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Molecular Type=">https://www.gephebase.org/search-criteria?/and+Molecular Type="+Cis-regulatory+"#gephebase-summary-title)	
	Aberration Type
SNP (<a +snp+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Aberration Type=">https://www.gephebase.org/search-criteria?/and+Aberration Type="+SNP+"#gephebase-summary-title)	
	Molecular Details of the Mutation
rs1421085 T-to-C single-nucleotide variant disrupts a conserved motif for the ARID5B repressor	
	Experimental Evidence
Association Mapping (<a +association="" href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=" mapping+"#gephebase-summary-title"="">https://www.gephebase.org/search-criteria?/and+Experimental Evidence="+Association Mapping+"#gephebase-summary-title)	
	Main Reference
A common variant in the FTO gene is associated with body mass index and predisposes to childhood and adult obesity. (2007) (https://pubmed.ncbi.nlm.nih.gov/17434869)	
	Authors
Frayling TM; Timpson NJ; Weedon MN; Zeggini E; Freathy RM; Lindgren CM; Perry JR; Elliott KS; Lango H; Rayner NW; Shields B; Harries LW; Barrett JC; Ellard S; Groves CJ; Knight B; Patch AM; Ness AR; Ebrahim S; Lawlor DA; Ring SM; Ben-Shlomo Y; Jarvelin MR; Sovio U; Bennett AJ; Melzer D; Ferrucci L; Loos RJ; Barroso I; Wareham NJ; Karpe F; Owen KR; Cardon LR; Walker M; Hitman GA; Palmer CN; Doney AS; Morris AD; Smith GD; Hattersley AT; McCarthy MI	
	Abstract
Obesity is a serious international health problem that increases the risk of several common diseases. The genetic factors predisposing to obesity are poorly understood. A genome-wide search for type 2 diabetes-susceptibility genes identified a common variant in the FTO (fat mass and obesity associated) gene that predisposes to diabetes through an effect on body mass index (BMI). An additive association of the variant with BMI was replicated in 13 cohorts with 38,759 participants. The 16% of adults who are homozygous for the risk allele weighed about 3 kilograms more and had 1.67-fold increased odds of obesity when compared with those not inheriting a risk allele. This association was observed from age 7 years upward and reflects a specific increase in fat mass.	
	Additional References
Variation in FTO contributes to childhood obesity and severe adult obesity. (2007) (https://pubmed.ncbi.nlm.nih.gov/17496892) Genome-wide association scan shows genetic variants in the FTO gene are associated with obesity-related traits. (2007) (https://pubmed.ncbi.nlm.nih.gov/17658951) FTO genotype is associated with phenotypic variability of body mass index. (2012) (https://pubmed.ncbi.nlm.nih.gov/22982992) FTO Obesity Variant Circuitry and Adipocyte Browning in Humans. (2015) (https://pubmed.ncbi.nlm.nih.gov/26287746)	

RELATED GEPHE

	Related Genes
23 (ADAMTS10, agrecan, CREBRF, DC-STAMP domain containing 2 (DCST2), DYM, EIF2AK3, 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)) (<a +9606+"="" and+grouphaplotypes='true#gephebase-summary-title"' and+trait="Body" href="https://www.gephebase.org/search-criteria?/or+Taxon ID=" size="">https://www.gephebase.org/search-criteria?/or+Taxon ID="+9606+"/and+Trait=Body size/and+groupHaplotypes=true#gephebase-summary-title)	
	Related Haplotypes
1 (<a #gephebase-summary-title"="" +fto"="" and+taxon="" gephebase="+FTO" href="https://www.gephebase.org/search-criteria?/or+Gene Gephebase=" id="+9606" or+gene="">https://www.gephebase.org/search-criteria?/or+Gene Gephebase="+FTO"/and+Taxon ID="+9606"/or+Gene Gephebase="+FTO"/and+Taxon ID="+9606"#gephebase-summary-title)	

EXTERNAL LINKS

COMMENTS

Functional validation in 26287746