GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00000099

Entry Status

Published

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Homo sapiens

Trait State in Taxon B

Homo sapiens

Ancestral State

Data not curated

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

APOE

Synonyms

AD2; LPG; APO-E; ApoE4; LDLCQ5

String

9606.ENSP00000252486 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000252486)

Sequence Similarities

Belongs to the apolipoprotein A1/A4/E family.

GO - Molecular Function

GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)
GO:0046983 : protein dimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046983)

UniProtKB Homo sapiens

P02649 (http://www.uniprot.org/uniprot/P02649)

GenebankID or UniProtKB

NP_001289617 (https://www.ncbi.nlm.nih.gov/nuccore/NP_001289617)

GO:0042803 : protein homodimerization activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0001540 : amyloid-beta binding (https://www.ebi.ac.uk/QuickGO/term/GO:0001540)
GO:0016209 : antioxidant activity (https://www.ebi.ac.uk/QuickGO/term/GO:0016209)
GO:0015485 : cholesterol binding (https://www.ebi.ac.uk/QuickGO/term/GO:0015485)
GO:0017127 : cholesterol transporter activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0017127)
GO:0008201 : heparin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008201)
GO:0008289 : lipid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008289)
GO:0005319 : lipid transporter activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0005319)
GO:0071813 : lipoprotein particle binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0071813)
GO:0050750 : low-density lipoprotein particle receptor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0050750)
GO:0046911 : metal chelating activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046911)
GO:0060228 : phosphatidylcholine-sterol O-acyltransferase activator activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0060228)
GO:0005543 : phospholipid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005543)
GO:0044877 : protein-containing complex binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0044877)
GO:0005198 : structural molecule activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0005198)
GO:0048156 : tau protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0048156)
GO:0070326 : very-low-density lipoprotein particle receptor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0070326)

GO - Biological Process

GO:0033344 : cholesterol efflux (https://www.ebi.ac.uk/QuickGO/term/GO:0033344)
GO:0006357 : regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0006357)
GO:0045893 : positive regulation of transcription, DNA-templated
(https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0097113 : AMPA glutamate receptor clustering
(https://www.ebi.ac.uk/QuickGO/term/GO:0097113)
GO:0042982 : amyloid precursor protein metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042982)
GO:0048844 : artery morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0048844)
GO:0006874 : cellular calcium ion homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0006874)
GO:0044267 : cellular protein metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0044267)
GO:0019934 : cGMP-mediated signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0019934)
GO:0006695 : cholesterol biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006695)
GO:0006707 : cholesterol catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006707)
GO:0042632 : cholesterol homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0042632)
GO:0008203 : cholesterol metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0008203)
GO:0034378 : chylomicron assembly (https://www.ebi.ac.uk/QuickGO/term/GO:0034378)
GO:0034382 : chylomicron remnant clearance
(https://www.ebi.ac.uk/QuickGO/term/GO:0034382)
GO:0034371 : chylomicron remodeling
(https://www.ebi.ac.uk/QuickGO/term/GO:0034371)
GO:0007010 : cytoskeleton organization
(https://www.ebi.ac.uk/QuickGO/term/GO:0007010)
GO:0055089 : fatty acid homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0055089)
GO:0007186 : G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007186)
GO:0034380 : high-density lipoprotein particle assembly
(https://www.ebi.ac.uk/QuickGO/term/GO:0034380)
GO:0034384 : high-density lipoprotein particle clearance
(https://www.ebi.ac.uk/QuickGO/term/GO:0034384)
GO:0034375 : high-density lipoprotein particle remodeling
(https://www.ebi.ac.uk/QuickGO/term/GO:0034375)
GO:0046907 : intracellular transport (https://www.ebi.ac.uk/QuickGO/term/GO:0046907)
GO:0010877 : lipid transport involved in lipid storage
(https://www.ebi.ac.uk/QuickGO/term/GO:0010877)
GO:0042158 : lipoprotein biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042158)
GO:0042159 : lipoprotein catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042159)
GO:0042157 : lipoprotein metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042157)
GO:0035641 : locomotory exploration behavior

(https://www.ebi.ac.uk/QuickGO/term/GO:0035641)
GO:0015909 : long-chain fatty acid transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0015909)
GO:0007616 : long-term memory (https://www.ebi.ac.uk/QuickGO/term/GO:0007616)
GO:0034374 : low-density lipoprotein particle remodeling
(https://www.ebi.ac.uk/QuickGO/term/GO:0034374)
GO:0051651 : maintenance of location in cell
(https://www.ebi.ac.uk/QuickGO/term/GO:0051651)
GO:1902430 : negative regulation of amyloid-beta formation
(https://www.ebi.ac.uk/QuickGO/term/GO:1902430)
GO:0030195 : negative regulation of blood coagulation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030195)
GO:0043537 : negative regulation of blood vessel endothelial cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0043537)
GO:0090090 : negative regulation of canonical Wnt signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0090090)
GO:0032269 : negative regulation of cellular protein metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0032269)
GO:0045541 : negative regulation of cholesterol biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0045541)
GO:0090370 : negative regulation of cholesterol efflux
(https://www.ebi.ac.uk/QuickGO/term/GO:0090370)
GO:0061000 : negative regulation of dendritic spine development
(https://www.ebi.ac.uk/QuickGO/term/GO:0061000)
GO:1902951 : negative regulation of dendritic spine maintenance
(https://www.ebi.ac.uk/QuickGO/term/GO:1902951)
GO:0001937 : negative regulation of endothelial cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0001937)
GO:0010629 : negative regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010629)
GO:0050728 : negative regulation of inflammatory response
(https://www.ebi.ac.uk/QuickGO/term/GO:0050728)
GO:0051055 : negative regulation of lipid biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0051055)
GO:1903001 : negative regulation of lipid transport across blood-brain barrier
(https://www.ebi.ac.uk/QuickGO/term/GO:1903001)
GO:1900272 : negative regulation of long-term synaptic potentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:1900272)
GO:0043407 : negative regulation of MAP kinase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0043407)
GO:0043524 : negative regulation of neuron apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043524)
GO:1901215 : negative regulation of neuron death
(https://www.ebi.ac.uk/QuickGO/term/GO:1901215)
GO:0010977 : negative regulation of neuron projection development
(https://www.ebi.ac.uk/QuickGO/term/GO:0010977)
GO:1902999 : negative regulation of phospholipid efflux
(https://www.ebi.ac.uk/QuickGO/term/GO:1902999)
GO:0010544 : negative regulation of platelet activation
(https://www.ebi.ac.uk/QuickGO/term/GO:0010544)
GO:1901627 : negative regulation of postsynaptic membrane organization
(https://www.ebi.ac.uk/QuickGO/term/GO:1901627)
GO:1901630 : negative regulation of presynaptic membrane organization
(https://www.ebi.ac.uk/QuickGO/term/GO:1901630)
GO:0090209 : negative regulation of triglyceride metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0090209)
GO:0007263 : nitric oxide mediated signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0007263)
GO:0097114 : NMDA glutamate receptor clustering
(https://www.ebi.ac.uk/QuickGO/term/GO:0097114)
GO:0033700 : phospholipid efflux (https://www.ebi.ac.uk/QuickGO/term/GO:0033700)
GO:0044794 : positive regulation by host of viral process
(https://www.ebi.ac.uk/QuickGO/term/GO:0044794)
GO:1905908 : positive regulation of amyloid fibril formation
(https://www.ebi.ac.uk/QuickGO/term/GO:1905908)
GO:1902004 : positive regulation of amyloid-beta formation
(https://www.ebi.ac.uk/QuickGO/term/GO:1902004)
GO:0010875 : positive regulation of cholesterol efflux
(https://www.ebi.ac.uk/QuickGO/term/GO:0010875)
GO:0010873 : positive regulation of cholesterol esterification
(https://www.ebi.ac.uk/QuickGO/term/GO:0010873)
GO:0060999 : positive regulation of dendritic spine development
(https://www.ebi.ac.uk/QuickGO/term/GO:0060999)
GO:1902952 : positive regulation of dendritic spine maintenance
(https://www.ebi.ac.uk/QuickGO/term/GO:1902952)
GO:0045807 : positive regulation of endocytosis
(https://www.ebi.ac.uk/QuickGO/term/GO:0045807)
GO:0070374 : positive regulation of ERK1 and ERK2 cascade

(https://www.ebi.ac.uk/QuickGO/term/GO:0070374)
GO:1905855 : positive regulation of heparan sulfate binding
(https://www.ebi.ac.uk/QuickGO/term/GO:1905855)
GO:1905860 : positive regulation of heparan sulfate proteoglycan binding
(https://www.ebi.ac.uk/QuickGO/term/GO:1905860)
GO:0046889 : positive regulation of lipid biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0046889)
GO:1903002 : positive regulation of lipid transport across blood-brain barrier
(https://www.ebi.ac.uk/QuickGO/term/GO:1903002)
GO:0032805 : positive regulation of low-density lipoprotein particle receptor catabolic
process (https://www.ebi.ac.uk/QuickGO/term/GO:0032805)
GO:0051044 : positive regulation of membrane protein ectodomain proteolysis
(https://www.ebi.ac.uk/QuickGO/term/GO:0051044)
GO:1902998 : positive regulation of neurofibrillary tangle assembly
(https://www.ebi.ac.uk/QuickGO/term/GO:1902998)
GO:1901216 : positive regulation of neuron death
(https://www.ebi.ac.uk/QuickGO/term/GO:1901216)
GO:0010976 : positive regulation of neuron projection development
(https://www.ebi.ac.uk/QuickGO/term/GO:0010976)
GO:0051000 : positive regulation of nitric-oxide synthase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0051000)
GO:1902995 : positive regulation of phospholipid efflux
(https://www.ebi.ac.uk/QuickGO/term/GO:1902995)
GO:1901628 : positive regulation of postsynaptic membrane organization
(https://www.ebi.ac.uk/QuickGO/term/GO:1901628)
GO:1901631 : positive regulation of presynaptic membrane organization
(https://www.ebi.ac.uk/QuickGO/term/GO:1901631)
GO:0043687 : post-translational protein modification
(https://www.ebi.ac.uk/QuickGO/term/GO:0043687)
GO:0017038 : protein import (https://www.ebi.ac.uk/QuickGO/term/GO:0017038)
GO:0006898 : receptor-mediated endocytosis
(https://www.ebi.ac.uk/QuickGO/term/GO:0006898)
GO:1905906 : regulation of amyloid fibril formation
(https://www.ebi.ac.uk/QuickGO/term/GO:1905906)
GO:1900221 : regulation of amyloid-beta clearance
(https://www.ebi.ac.uk/QuickGO/term/GO:1900221)
GO:0030516 : regulation of axon extension
(https://www.ebi.ac.uk/QuickGO/term/GO:0030516)
GO:2000822 : regulation of behavioral fear response
(https://www.ebi.ac.uk/QuickGO/term/GO:2000822)
GO:0032489 : regulation of Cdc42 protein signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0032489)
GO:1905890 : regulation of cellular response to very-low-density lipoprotein particle
stimulus (https://www.ebi.ac.uk/QuickGO/term/GO:1905890)
GO:0090181 : regulation of cholesterol metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0090181)
GO:0032374 : regulation of cholesterol transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0032374)
GO:0045088 : regulation of innate immune response
(https://www.ebi.ac.uk/QuickGO/term/GO:0045088)
GO:1901214 : regulation of neuron death
(https://www.ebi.ac.uk/QuickGO/term/GO:1901214)
GO:0048168 : regulation of neuronal synaptic plasticity
(https://www.ebi.ac.uk/QuickGO/term/GO:0048168)
GO:0061136 : regulation of proteasomal protein catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0061136)
GO:0032462 : regulation of protein homooligomerization
(https://www.ebi.ac.uk/QuickGO/term/GO:0032462)
GO:0051246 : regulation of protein metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0051246)
GO:1902947 : regulation of tau-protein kinase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:1902947)
GO:0061771 : response to caloric restriction
(https://www.ebi.ac.uk/QuickGO/term/GO:0061771)
GO:0002021 : response to dietary excess
(https://www.ebi.ac.uk/QuickGO/term/GO:0002021)
GO:0000302 : response to reactive oxygen species
(https://www.ebi.ac.uk/QuickGO/term/GO:0000302)
GO:0001523 : retinoid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0001523)
GO:0043691 : reverse cholesterol transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0043691)
GO:0007271 : synaptic transmission, cholinergic
(https://www.ebi.ac.uk/QuickGO/term/GO:0007271)
GO:0019433 : triglyceride catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0019433)
GO:0070328 : triglyceride homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0070328)

GO:0006641 : triglyceride metabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006641)
GO:0042311 : vasodilation (https://www.ebi.ac.uk/QuickGO/term/GO:0042311)
GO:0034447 : very-low-density lipoprotein particle clearance (https://www.ebi.ac.uk/QuickGO/term/GO:0034447)
GO:0034372 : very-low-density lipoprotein particle remodeling (https://www.ebi.ac.uk/QuickGO/term/GO:0034372)
GO:0019068 : virion assembly (https://www.ebi.ac.uk/QuickGO/term/GO:0019068)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0016020 : membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)
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:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0005794 : Golgi apparatus (https://www.ebi.ac.uk/QuickGO/term/GO:0005794)
GO:0043083 : synaptic cleft (https://www.ebi.ac.uk/QuickGO/term/GO:0043083)
GO:0062023 : collagen-containing extracellular matrix (https://www.ebi.ac.uk/QuickGO/term/GO:0062023)
GO:0098978 : glutamatergic synapse (https://www.ebi.ac.uk/QuickGO/term/GO:0098978)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)
GO:0072562 : blood microparticle (https://www.ebi.ac.uk/QuickGO/term/GO:0072562)
GO:0042627 : chylomicron (https://www.ebi.ac.uk/QuickGO/term/GO:0042627)
GO:0030669 : clathrin-coated endocytic vesicle membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0030669)
GO:0030425 : dendrite (https://www.ebi.ac.uk/QuickGO/term/GO:0030425)
GO:0034365 : discoidal high-density lipoprotein particle (https://www.ebi.ac.uk/QuickGO/term/GO:0034365)
GO:0005769 : early endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005769)
GO:0071682 : endocytic vesicle lumen (https://www.ebi.ac.uk/QuickGO/term/GO:0071682)
GO:0005783 : endoplasmic reticulum (https://www.ebi.ac.uk/QuickGO/term/GO:0005783)
GO:0005788 : endoplasmic reticulum lumen (https://www.ebi.ac.uk/QuickGO/term/GO:0005788)
GO:1903561 : extracellular vesicle (https://www.ebi.ac.uk/QuickGO/term/GO:1903561)
GO:0034364 : high-density lipoprotein particle (https://www.ebi.ac.uk/QuickGO/term/GO:0034364)
GO:0034363 : intermediate-density lipoprotein particle (https://www.ebi.ac.uk/QuickGO/term/GO:0034363)
GO:1990777 : lipoprotein particle (https://www.ebi.ac.uk/QuickGO/term/GO:1990777)
GO:0034362 : low-density lipoprotein particle (https://www.ebi.ac.uk/QuickGO/term/GO:0034362)
GO:0043025 : neuronal cell body (https://www.ebi.ac.uk/QuickGO/term/GO:0043025)
GO:0034361 : very-low-density lipoprotein particle (https://www.ebi.ac.uk/QuickGO/term/GO:0034361)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Cys112Arg

Experimental Evidence

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

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

Main Reference

Variants near CHRNA3/5 and APOE have age- and sex-related effects on human lifespan. (2016) (https://pubmed.ncbi.nlm.nih.gov/27029810)

Authors

Joshi PK; Fischer K; Schraut KE; Campbell H; Esko T; Wilson JF

Abstract

Lifespan is a trait of enormous personal interest. Research into the biological basis of human lifespan, however, is hampered by the long time to death. Using a novel approach of regressing (272,081) parental lifespans beyond age 40 years on participant genotype in a new large data set (UK Biobank), we here show that common variants near the apolipoprotein E and nicotinic acetylcholine receptor subunit alpha 5 genes are associated with lifespan. The effects are strongly sex and age dependent, with APOE É›4 differentially influencing maternal lifespan (P=4.2 Ã— 10(-15), effect -1.24 years of maternal life per imputed risk allele in parent; sex difference, P=0.011), and a locus near CHRNA3/5 differentially affecting paternal lifespan (P=4.8 Ã— 10(-11), effect -0.86 years per allele; sex difference P=0.075). Rare homozygous carriers of the risk alleles at both loci are predicted to have 3.3-3.7 years shorter lives.

Additional References

RELATED GEPHE

Related Genes

1 (CHRNA3/5 nicotine receptor cluster) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Aging/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

LD with SNP previously associated to other age-related conditions and traits including AIzheimer