

GEPHE SUMMARY

Gephebase Gene		GepheID	
CHRNA3/5 nicotine receptor cluster (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^CHRNA3/5+nicotine+receptor+cluster^#gephebase-summary-title)		GP00000185	
Published		Main curator	
Entry Status		Martin	

PHENOTYPIC CHANGE

Trait #1		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			

Trait #2		Trait Category	
Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Physiology^#gephebase-summary-title)		Trait	
Smoking behavior (https://www.gephebase.org/search-criteria?/and+Trait=^Smoking+behavior^#gephebase-summary-title)		Trait State in Taxon A	
-		Trait State in Taxon B	
-			

Ancestral State			
Data not curated		Taxonomic Status	
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title)			
Taxon A		Taxon B	
Homo sapiens		Homo sapiens	
(https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title)		(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 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	
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)		Homo () - (Rank: genus)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)	
9606		9606	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)	
is Taxon A an Infrspecies?		is Taxon B an Infrspecies?	
No		No	

GENOTYPIC CHANGE

CHRNA3	Generic Gene Name	UniProtKB Homo sapiens
		P32297 (http://www.uniprot.org/uniprot/P32297)
	Synonyms	GenebankID or UniProtKB
	LNCR2; PAOD2; NACHRA3	AAH06114 (https://www.ncbi.nlm.nih.gov/nuccore/AAH06114)
9606.ENSPO0000315602 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000315602)	String	
	Sequence Similarities	
	Belongs to the ligand-gated ion channel (TC 1.A.9) family. Acetylcholine receptor (TC 1.A.9.1) subfamily. Alpha-3/CHRNA3 sub-subfamily.	
GO:0042166 : acetylcholine binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042166) GO:0015464 : acetylcholine receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015464) GO:0022848 : acetylcholine-gated cation-selective channel activity (https://www.ebi.ac.uk/QuickGO/term/GO:0022848) GO:0015276 : ligand-gated ion channel activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015276)	GO - Molecular Function	
GO:0007165 : signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007165) GO:0007268 : chemical synaptic transmission (https://www.ebi.ac.uk/QuickGO/term/GO:0007268) GO:0007399 : nervous system development (https://www.ebi.ac.uk/QuickGO/term/GO:0007399) GO:0007271 : synaptic transmission, cholinergic (https://www.ebi.ac.uk/QuickGO/term/GO:0007271) GO:0034220 : ion transmembrane transport (https://www.ebi.ac.uk/QuickGO/term/GO:0034220) GO:0050877 : nervous system process (https://www.ebi.ac.uk/QuickGO/term/GO:0050877) GO:0007274 : neuromuscular synaptic transmission (https://www.ebi.ac.uk/QuickGO/term/GO:0007274) GO:0042391 : regulation of membrane potential (https://www.ebi.ac.uk/QuickGO/term/GO:0042391) GO:0035094 : response to nicotine (https://www.ebi.ac.uk/QuickGO/term/GO:0035094) GO:0095500 : acetylcholine receptor signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0095500) GO:0007171 : activation of transmembrane receptor protein tyrosine kinase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0007171) GO:0035095 : behavioral response to nicotine (https://www.ebi.ac.uk/QuickGO/term/GO:0035095) GO:0060079 : excitatory postsynaptic potential (https://www.ebi.ac.uk/QuickGO/term/GO:0060079) GO:0006811 : ion transport (https://www.ebi.ac.uk/QuickGO/term/GO:0006811) GO:0007626 : locomotory behavior (https://www.ebi.ac.uk/QuickGO/term/GO:0007626) GO:0014056 : regulation of acetylcholine secretion, neurotransmission (https://www.ebi.ac.uk/QuickGO/term/GO:0014056) GO:0048814 : regulation of dendrite morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0048814) GO:0006940 : regulation of smooth muscle contraction (https://www.ebi.ac.uk/QuickGO/term/GO:0006940) GO:1905144 : response to acetylcholine (https://www.ebi.ac.uk/QuickGO/term/GO:1905144) GO:0060084 : synaptic transmission involved in micturition (https://www.ebi.ac.uk/QuickGO/term/GO:0060084)	GO - Biological Process	
GO:0016021 : integral component of membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016021) GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886) GO:0005887 : integral component of plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005887) GO:0030054 : cell junction (https://www.ebi.ac.uk/QuickGO/term/GO:0030054) GO:0030425 : dendrite (https://www.ebi.ac.uk/QuickGO/term/GO:0030425) GO:0043025 : neuronal cell body (https://www.ebi.ac.uk/QuickGO/term/GO:0043025) GO:0043005 : neuron projection (https://www.ebi.ac.uk/QuickGO/term/GO:0043005) GO:0045211 : postsynaptic membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0045211) GO:0045202 : synapse (https://www.ebi.ac.uk/QuickGO/term/GO:0045202) GO:0005892 : acetylcholine-gated channel complex (https://www.ebi.ac.uk/QuickGO/term/GO:0005892) GO:0044853 : plasma membrane raft (https://www.ebi.ac.uk/QuickGO/term/GO:0044853) GO:0014069 : postsynaptic density (https://www.ebi.ac.uk/QuickGO/term/GO:0014069)	GO - Cellular Component	
Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)		Presumptive Null

Unknown (https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Unknown~#gephebase-summary-title)	Molecular Type
Unknown (https://www.gephebase.org/search-criteria?/and+Aberration+Type=~Unknown~#gephebase-summary-title)	Aberration Type
unknown; complex variation with SNP association signal	Molecular Details of the Mutation
Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Association+Mapping~#gephebase-summary-title)	Experimental Evidence
Variants near CHRNA3/5 and APOE have age- and sex-related effects on human lifespan. (2016) (https://pubmed.ncbi.nlm.nih.gov/27029810)	Main Reference
Joshi PK; Fischer K; Schraut KE; Campbell H; Esko T; Wilson JF	Authors
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 ε ₄ differentially influencing maternal lifespan (P=4.2 Å— 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 Å— 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.	Abstract
	Additional References

RELATED GEPHE

1 (APOE (apolipoprotein E)) (https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9606~/and+Trait=Aging/or+Taxon+ID=~9606~/and+Trait=Smoking+behavior/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

Locus may affect age-specific susceptibility to death via smoking behavior - not a longevity gene