

GEPHE SUMMARY

Gephebase Gene		GepheID
EIF2AK3 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^EIF2AK3^#gephebase-summary-title)		GP00000264
Entry Status		Main curator
Published		

PHENOTYPIC CHANGE

Trait Category			
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Morphology^#gephebase-summary-title)		Trait	
Body size (height) (https://www.gephebase.org/search-criteria?/and+Trait=^Body size (height)^#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+TaxonomicStatus=^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)	
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)		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)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
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 Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

Generic Gene Name		UniProtKB Homo sapiens
EIF2AK3		Q9NZJ5 (http://www.uniprot.org/uniprot/Q9NZJ5)
Synonyms		GenebankID or UniProtKB
PEK; WRS; PERK		BC126354 (https://www.ncbi.nlm.nih.gov/nucore/BC126354)
String		
9606.ENSPO0000307235		
(http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000307235)		
Sequence Similarities		
Belongs to the protein kinase superfamily. Ser/Thr protein kinase family. GCN2 subfamily.		
GO - Molecular Function		
GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)		
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)		
GO:0042803 : protein homodimerization activity		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042803>)
GO:0019899 : enzyme binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0019899>)
GO:0004674 : protein serine/threonine kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004674>)
GO:0004694 : eukaryotic translation initiation factor 2alpha kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004694>)
GO:0051879 : Hsp90 protein binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0051879>)
GO:0004672 : protein kinase activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0004672>)
GO:0019903 : protein phosphatase binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019903>)

GO - Biological Process

GO:0002063 : chondrocyte development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002063>)
GO:0001501 : skeletal system development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001501>)
GO:0010628 : positive regulation of gene expression
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010628>)
GO:0019722 : calcium-mediated signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019722>)
GO:0006468 : protein phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006468>)
GO:0001503 : ossification (<https://www.ebi.ac.uk/QuickGO/term/GO:0001503>)
GO:0018105 : peptidyl-serine phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0018105>)
GO:0051260 : protein homooligomerization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051260>)
GO:0006919 : activation of cysteine-type endopeptidase activity involved in apoptotic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0006919>)
GO:0001525 : angiogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0001525>)
GO:0030282 : bone mineralization (<https://www.ebi.ac.uk/QuickGO/term/GO:0030282>)
GO:0034198 : cellular response to amino acid starvation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0034198>)
GO:0070417 : cellular response to cold
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070417>)
GO:0042149 : cellular response to glucose starvation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042149>)
GO:0036492 : eIF2alpha phosphorylation in response to endoplasmic reticulum stress
(<https://www.ebi.ac.uk/QuickGO/term/GO:0036492>)
GO:0031018 : endocrine pancreas development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031018>)
GO:0007029 : endoplasmic reticulum organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007029>)
GO:0030968 : endoplasmic reticulum unfolded protein response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030968>)
GO:0006983 : ER overload response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006983>)
GO:0048009 : insulin-like growth factor receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048009>)
GO:0031642 : negative regulation of myelination
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031642>)
GO:0017148 : negative regulation of translation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0017148>)
GO:0032057 : negative regulation of translational initiation in response to stress
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032057>)
GO:0036499 : PERK-mediated unfolded protein response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0036499>)
GO:1900182 : positive regulation of protein localization to nucleus
(<https://www.ebi.ac.uk/QuickGO/term/GO:1900182>)
GO:0045943 : positive regulation of transcription by RNA polymerase I
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045943>)
GO:0010575 : positive regulation of vascular endothelial growth factor production
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010575>)
GO:0046777 : protein autophosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046777>)
GO:0060734 : regulation of endoplasmic reticulum stress-induced eIF2 alpha phosphorylation (<https://www.ebi.ac.uk/QuickGO/term/GO:0060734>)
GO:1902235 : regulation of endoplasmic reticulum stress-induced intrinsic apoptotic signaling pathway (<https://www.ebi.ac.uk/QuickGO/term/GO:1902235>)
GO:0010998 : regulation of translational initiation by eIF2 alpha phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010998>)
GO:0034976 : response to endoplasmic reticulum stress
(<https://www.ebi.ac.uk/QuickGO/term/GO:0034976>)
GO:1990737 : response to manganese-induced endoplasmic reticulum stress
(<https://www.ebi.ac.uk/QuickGO/term/GO:1990737>)

GO - Cellular Component

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:0030176 : integral component of endoplasmic reticulum membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030176>)
GO:0005783 : endoplasmic reticulum
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005783>)
GO:0005789 : endoplasmic reticulum membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005789>)
GO:0022626 : cytosolic ribosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0022626>)
GO:0048471 : perinuclear region of cytoplasm
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048471>)

	Presumptive Null
Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#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
unknown	
	Experimental Evidence
Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)	
	Main Reference
Recent progress in the study of the genetics of height. (2011) (https://pubmed.ncbi.nlm.nih.gov/21340692)	
	Authors
Lettre G	
	Abstract
Adult height is a classic polygenic trait of high narrow-sense heritability ($h^2 = 0.8$). In the late nineteenth to early twentieth century, variation in adult height was used as a model to set the foundation of the fields of statistics and quantitative genetics. More recently, with our increasing knowledge concerning the extent of genetic variation in the human genome, human geneticists have used genome-wide association studies to identify hundreds of loci robustly associated with adult height, providing new insights into human growth and development, and into the architecture of complex human traits. In this review, I highlight the progress made in the last 2 years in understanding how genetic variation controls height variation in humans, including non-Caucasian populations and children.	
	Additional References

RELATED GEPHE

	Related Genes
23 (ADAMTS10, aggrecan, CREBRF, DC-STAMP domain containing 2 (DCST2), DYM, 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)	
	Related Haplotypes
No matches found.	

EXTERNAL LINKS

COMMENTS