

GEPHE SUMMARY

	Gephebase Gene		GepheID
Muscle-specific adenosine monophosphate-activated protein kinase (PRKAG3) (<a +muscle-specific+adenosine+monophosphate-activated+protein+kinase+(prkag3)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Muscle-specific+adenosine+monophosphate-activated+protein+kinase+(PRKAG3)+"#gephebase-summary-title)		GP00000682	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Physiology (<a +physiology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Physiology+"#gephebase-summary-title)		Trait	
Glycogen content (muscles) (<a +glycogen+content+(muscles)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Glycogen+content+(muscles)+"#gephebase-summary-title)		Trait State in Taxon A	
Sus scrofa		Trait State in Taxon B	
Sus scrofa - Hampshire - muscle with higher glycogen and distinct cooking and curing traits		Ancestral State	
Taxon A		Taxonomic Status	
Domesticated (<a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated+"#gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Sus scrofa (<a +sus+scrofa+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Sus+scrofa+"#gephebase-summary-title)		Sus scrofa domesticus (<a +sus+scrofa+domesticus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Sus+scrofa+domesticus+"#gephebase-summary-title)	
	Common Name		Common Name
pig		domestic pig	
	Synonyms		Synonyms
pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofa		Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig	
	Rank		Rank
species		subspecies	
	Lineage		Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Suina; Suidae; Sus		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Suina; Suidae; Sus; Sus scrofa	
	Parent		Parent
Sus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9822)		Sus scrofa (pig) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9823 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823)		9825 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9825)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		Yes	
			Taxon B Description
		Sus scrofa - Hampshire	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Prkag3		Q8BGM7 (http://www.uniprot.org/uniprot/Q8BGM7)	
	Synonyms		GenebankID or UniProtKB
Amkg3; Ampkg3; AMPKg3L; AMPKg3S		AAP12533 (https://www.ncbi.nlm.nih.gov/nuccore/AAP12533)	
	String		
10090.ENSMUSP00000080342 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000080342)			
	Sequence Similarities		
Belongs to the 5'-AMP-activated protein kinase gamma subunit family.			
	GO - Molecular Function		
GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)			
GO:0019901 : protein kinase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019901)			
GO:0004679 : AMP-activated protein kinase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004679)			

GO:0005978 : glycogen biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005978>)
GO:0006633 : fatty acid biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006633>)
GO:0006096 : glycolytic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0006096>)
GO:0071900 : regulation of protein serine/threonine kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071900>)
GO:0014873 : response to muscle activity involved in regulation of muscle adaptation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014873>)

GO - Cellular Component

GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)
GO:0031588 : nucleotide-activated protein kinase complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031588>)

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

R200Q in CBS1; which is the most conserved region among AMPK g chain isoforms; and R200 is conserved in mammalian and Drosophila AMPK g isoforms

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	Gln	200

Main Reference

A mutation in PRKAG3 associated with excess glycogen content in pig skeletal muscle. (2000) (<https://pubmed.ncbi.nlm.nih.gov/10818001>)

Authors

Milan D; Jeon JT; Looft C; Amarger V; Robic A; Thelander M; Rogel-Gaillard C; Paul S; Iannuccelli N; Rask L; Ronne H; Lundstr m K; Reinsch N; Gellin J; Kalm E; Roy PL; Chardon P; Andersson L

Abstract

A high proportion of purebred Hampshire pigs carries the dominant RN- mutation, which causes high glycogen content in skeletal muscle. The mutation has beneficial effects on meat content but detrimental effects on processing yield. Here, it is shown that the mutation is a nonconservative substitution (R200Q) in the PRKAG3 gene, which encodes a muscle-specific isoform of the regulatory gamma subunit of adenosine monophosphate-activated protein kinase (AMPK). Loss-of-function mutations in the homologous gene in yeast (SNF4) cause defects in glucose metabolism, including glycogen storage. Further analysis of the PRKAG3 signaling pathway may provide insights into muscle physiology as well as the pathogenesis of noninsulin-dependent diabetes mellitus in humans, a metabolic disorder associated with impaired glycogen synthesis.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

1 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Muscle-specific adenosine monophosphate-activated protein kinase \(PRKAG3\)~/and+Taxon ID=~9823~/or+Gene Gephebase=~Muscle-specific adenosine monophosphate-activated protein kinase \(PRKAG3\)~/and+Taxon ID=~9825~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Muscle-specific adenosine monophosphate-activated protein kinase (PRKAG3)~/and+Taxon ID=~9823~/or+Gene Gephebase=~Muscle-specific adenosine monophosphate-activated protein kinase (PRKAG3)~/and+Taxon ID=~9825~#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

@AllelicSeries <https://omia.org/OMIA001085/9823/>

