GEPHE SUMMARY

Gephebase Gene

Muscle-specific adenosine monophosphate-activated protein kinase (PRKAG3) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Muscle-specific adenosine monophosphate-activated protein kinase (PRKAG3)^#gephebase-summary-title)

GepheID

GP00000682

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

Glycogen content (muscles) (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 (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Sus scrofa (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Sus scrofa^#gephebase-summary-title)

Common Name

pig

Synonyms

pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofus

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; Laurasiatheria; Cetartiodactyla; Suina; Suidae; Sus

Parent

Sus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9822)

NCBI Taxonomy ID

9823 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Sus scrofa domesticus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Sus scrofa domesticus^#gephebase-summary-title)

Common Name

domestic pig

Synonyms

Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig

Rank

subspecies

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; Sus scrofa

Parent

Sus scrofa (pig) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)

NCBI Taxonomy ID

9825 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9825)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Sus scrofa - Hampshire

GENOTYPIC CHANGE

Generic Gene Name

Prkag3

Synonyms

Amkg3; Ampkg3; AMPKg3L; AMPKg3S

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)

UniProtKB Mus musculus

Q8BGM7 (http://www.uniprot.org/uniprot/Q8BGM7)

GenebankID or UniProtKB

AAP12533 (https://www.ncbi.nlm.nih.gov/nuccore/AAP12533)

GO - Biological Process

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; LundstrÃ¶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)

EXTERNAL LINKS

COMMENTS

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