GEPHE SUMMARY

Gephebase Gene

RYR1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^RYR1^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00002338

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

WT

Trait State in Taxon B

Lean meat ; Hyperthermia in homozygotes

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 (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 B an Infraspecies?

Yes

Taxon B Description

-

GENOTYPIC CHANGE

Generic Gene Name

Ryr1

Synonyms

R; s; Ryr; skrr; RYR-1; AI528790

String

10090.ENSMUSP00000137123 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000137123)

Sequence Similarities

Belongs to the ryanodine receptor (TC 1.A.3.1) family. RYR1 subfamily.

GO - Molecular Function

GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)
GO:0019899 : enzyme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019899)
GO:0005516 : calmodulin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005516)
GO:0005509 : calcium ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005509)
GO:0002020 : protease binding (https://www.ebi.ac.uk/QuickGO/term/GO:0002020)

UniProtKB Mus musculus

E9PZQ0 (http://www.uniprot.org/uniprot/E9PZQ0)

GenebankID or UniProtKB

()

GO:0005262 : calcium channel activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0005262)
GO:0005245 : voltage-gated calcium channel activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0005245)
GO:0048763 : calcium-induced calcium release activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0048763)
GO:0005219 : ryanodine-sensitive calcium-release channel activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0005219)

GO - Biological Process

GO:0051480 : regulation of cytosolic calcium ion concentration
(https://www.ebi.ac.uk/QuickGO/term/GO:0051480)
GO:0043588 : skin development (https://www.ebi.ac.uk/QuickGO/term/GO:0043588)
GO:0001666 : response to hypoxia (https://www.ebi.ac.uk/QuickGO/term/GO:0001666)
GO:0006936 : muscle contraction (https://www.ebi.ac.uk/QuickGO/term/GO:0006936)
GO:0006937 : regulation of muscle contraction
(https://www.ebi.ac.uk/QuickGO/term/GO:0006937)
GO:0006816 : calcium ion transport (https://www.ebi.ac.uk/QuickGO/term/GO:0006816)
GO:0003151 : outflow tract morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0003151)
GO:0051209 : release of sequestered calcium ion into cytosol
(https://www.ebi.ac.uk/QuickGO/term/GO:0051209)
GO:0071318 : cellular response to ATP
(https://www.ebi.ac.uk/QuickGO/term/GO:0071318)
GO:0071313 : cellular response to caffeine
(https://www.ebi.ac.uk/QuickGO/term/GO:0071313)
GO:0071277 : cellular response to calcium ion
(https://www.ebi.ac.uk/QuickGO/term/GO:0071277)
GO:0043931 : ossification involved in bone maturation
(https://www.ebi.ac.uk/QuickGO/term/GO:0043931)
GO:0051289 : protein homotetramerization
(https://www.ebi.ac.uk/QuickGO/term/GO:0051289)
GO:0014808 : release of sequestered calcium ion into cytosol by sarcoplasmic reticulum
(https://www.ebi.ac.uk/QuickGO/term/GO:0014808)
GO:0031000 : response to caffeine (https://www.ebi.ac.uk/QuickGO/term/GO:0031000)
GO:0070296 : sarcoplasmic reticulum calcium ion transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0070296)
GO:0048741 : skeletal muscle fiber development
(https://www.ebi.ac.uk/QuickGO/term/GO:0048741)

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:0030659 : cytoplasmic vesicle membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0030659)
GO:0032991 : protein-containing complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0032991)
GO:0030315 : T-tubule (https://www.ebi.ac.uk/QuickGO/term/GO:0030315)
GO:0030018 : Z disc (https://www.ebi.ac.uk/QuickGO/term/GO:0030018)
GO:0005938 : cell cortex (https://www.ebi.ac.uk/QuickGO/term/GO:0005938)
GO:0042383 : sarcolemma (https://www.ebi.ac.uk/QuickGO/term/GO:0042383)
GO:0034704 : calcium channel complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0034704)
GO:0031234 : extrinsic component of cytoplasmic side of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0031234)
GO:0031674 : I band (https://www.ebi.ac.uk/QuickGO/term/GO:0031674)
GO:0031301 : integral component of organelle membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0031301)
GO:0030314 : junctional membrane complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0030314)
GO:1990425 : ryanodine receptor complex
(https://www.ebi.ac.uk/QuickGO/term/GO:1990425)
GO:0016529 : sarcoplasmic reticulum (https://www.ebi.ac.uk/QuickGO/term/GO:0016529)
GO:0033017 : sarcoplasmic reticulum membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0033017)
GO:0005790 : smooth endoplasmic reticulum
(https://www.ebi.ac.uk/QuickGO/term/GO:0005790)
GO:0014802 : terminal cisterna (https://www.ebi.ac.uk/QuickGO/term/GO:0014802)

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

p.R615C

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	CGC	TGC	1843
Amino-acid	Arg	Cys	615

Main Reference

Identification of a mutation in porcine ryanodine receptor associated with malignant hyperthermia. (1991) (https://pubmed.ncbi.nlm.nih.gov/1862346)

Authors

Fujii J; Otsu K; Zorzato F; de Leon S; Khanna VK; Weiler JE; O'Brien PJ; MacLennan DH

Abstract

Malignant hyperthermia (MH) causes neurological, liver, and kidney damage and death in humans and major economic losses in the swine industry. A single point mutation in the porcine gene for the skeletal muscle ryanodine receptor (ryr1) was found to be correlated with MH in five major breeds of lean, heavily muscled swine. Haplotyping suggests that the mutation in all five breeds has a common origin. Assuming that this is the causal mutation for MH, the development of a noninvasive diagnostic test will provide the basis for elimination of the MH gene or its controlled inclusion in swine breeding programs.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage @AllelicSeries