

GEPHE SUMMARY

<p>RYR1 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+RYR1+Gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002338</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category+Physiology+Gephebase-summary-title)</p> <p>Meat content (https://www.gephebase.org/search-criteria?/and+Trait+Meat+content+Gephebase-summary-title)</p> <p>WT</p> <p>Lean meat ; Hyperthermia in homozygotes</p> <p>Taxon A</p> <p>Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+Gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Taxon B</p>	<p>Taxon A</p> <p>Taxon B</p>	<p>Latin Name</p> <p>Latin Name</p> <p>Common Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Synonyms</p> <p>Rank</p> <p>Rank</p> <p>Lineage</p> <p>Lineage</p> <p>Parent</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Intraspecies?</p> <p>is Taxon B an Intraspecies?</p> <p>Taxon B Description</p>
<p>Sus scrofa (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Sus+scrofa+Gephebase-summary-title)</p> <p>pig</p> <p>pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofaus</p> <p>species</p> <p>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</p> <p>Sus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9822)</p> <p>9823 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823)</p> <p>No</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Intraspecies?</p>	<p>Sus scrofa (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Sus+scrofa+Gephebase-summary-title)</p> <p>pig</p> <p>pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofaus</p> <p>species</p> <p>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</p> <p>Sus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9822)</p> <p>9823 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823)</p> <p>Yes</p> <p>-</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p> <p>Taxon B Description</p>	

GENOTYPIC CHANGE

<p>Ryr1</p> <p>R; s; Ryr; skrr; RYR-1; A1528790</p> <p>10090.ENSMUSP00000137123 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000137123)</p> <p>Belongs to the ryanodine receptor (TC 1.A.3.1) family. RYR1 subfamily.</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>E9PZQ0 (http://www.uniprot.org/uniprot/E9PZQ0)</p> <p>0</p>	<p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
<p>GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)</p> <p>GO:0019899 : enzyme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019899)</p> <p>GO:0005516 : calmodulin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005516)</p> <p>GO:0005509 : calcium ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005509)</p> <p>GO:0002020 : protease binding (https://www.ebi.ac.uk/QuickGO/term/GO:0002020)</p>	<p>GO - Molecular Function</p>	<p>GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)</p> <p>GO:0019899 : enzyme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019899)</p> <p>GO:0005516 : calmodulin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005516)</p> <p>GO:0005509 : calcium ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005509)</p> <p>GO:0002020 : protease binding (https://www.ebi.ac.uk/QuickGO/term/GO:0002020)</p>	<p>GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)</p> <p>GO:0019899 : enzyme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019899)</p> <p>GO:0005516 : calmodulin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005516)</p> <p>GO:0005509 : calcium ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005509)</p> <p>GO:0002020 : protease binding (https://www.ebi.ac.uk/QuickGO/term/GO:0002020)</p>

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](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type="+Coding+"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=))

Aberration Type

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type="+SNP+"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

p.R615C

Experimental Evidence

	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