GEPHE SUMMARY

Gephebase Gene

Uncoupling protein 1 (UCP1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Uncoupling protein 1 (UCP1)^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00001162

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

Thermoregulation (brown adipose tissue) (https://www.gephebase.org/search-criteria?/and+Trait=^Thermoregulation (brown adipose tissue)^#gephebase-summary-title)

Trait State in Taxon A

Other mammals

Trait State in Taxon B

Sus scrofa; wild boars; Bornean bearded pig (S.barbatus); wart hog (Phacochoerus africanus); and red river hog(Potamochoerus porcus).

Ancestral State

Taxon A

Taxonomic Status

Intergeneric or Higher (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intergeneric or Higher^#gephebase-summary-title)

Taxon A

Latin Name

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

Common Name

mammals

Synonyms

mammals

Rank

class

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota

Parent

Amniota (amniotes) - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32524)

NCBI Taxonomy ID

40674 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 40674)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

-

Synonyms

-

Rank

genus

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

Parent

Suidae (pigs) - (Rank: family) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9821)

NCBI Taxonomy ID

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

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

Ucp1

Synonyms

Ucp; Slc25a7; AI385626

String

10090.ENSMUSP00000034146 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000034146)

Sequence Similarities

Belongs to the mitochondrial carrier (TC 2.A.29) family.

GO - Molecular Function

GO:0005525 : GTP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005525)
GO:0022857 : transmembrane transporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0022857)
GO:0019003 : GDP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019003)
GO:1901612 : cardiolipin binding (https://www.ebi.ac.uk/QuickGO/term/GO:1901612)
GO:0036041 : long-chain fatty acid binding

UniProtKB Mus musculus

P12242 (http://www.uniprot.org/uniprot/P12242)

GenebankID or UniProtKB

AAD26529 (https://www.ncbi.nlm.nih.gov/nuccore/AAD26529)

(https://www.ebi.ac.uk/QuickGO/term/GO:0036041)
GO:0017077 : oxidative phosphorylation uncoupler activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0017077)
GO:0032555 : purine ribonucleotide binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0032555)

GO - Biological Process

GO:0006357 : regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0006357)
GO:0032870 : cellular response to hormone stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:0032870)
GO:0009409 : response to cold (https://www.ebi.ac.uk/QuickGO/term/GO:0009409)
GO:0120162 : positive regulation of cold-induced thermogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0120162)
GO:0009266 : response to temperature stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:0009266)
GO:0050873 : brown fat cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0050873)
GO:1902600 : proton transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:1902600)
GO:0031667 : response to nutrient levels
(https://www.ebi.ac.uk/QuickGO/term/GO:0031667)
GO:1990845 : adaptive thermogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:1990845)
GO:0071398 : cellular response to fatty acid
(https://www.ebi.ac.uk/QuickGO/term/GO:0071398)
GO:0034614 : cellular response to reactive oxygen species
(https://www.ebi.ac.uk/QuickGO/term/GO:0034614)
GO:0002024 : diet induced thermogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0002024)
GO:1990542 : mitochondrial transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:1990542)
GO:0006839 : mitochondrial transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0006839)
GO:1903426 : regulation of reactive oxygen species biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:1903426)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005739 : mitochondrion (https://www.ebi.ac.uk/QuickGO/term/GO:0005739)
GO:0005743 : mitochondrial inner membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005743)
GO:0005740 : mitochondrial envelope
(https://www.ebi.ac.uk/QuickGO/term/GO:0005740)

Presumptive Null

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)

Molecular Type

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

Aberration Type

Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title)

Deletion Size

1-10 kb

Molecular Details of the Mutation

deletion of exons 3 to 5

Experimental Evidence

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

Main Reference

The uncoupling protein 1 gene (UCP1) is disrupted in the pig lineage: a genetic explanation for poor thermoregulation in piglets. (2006) (https://pubmed.ncbi.nlm.nih.gov/16933999)

Authors

Berg F; Gustafson U; Andersson L

Abstract

Piglets appear to lack brown adipose tissue, a specific type of fat that is essential for nonshivering thermogenesis in mammals, and they rely on shivering as the main mechanism for thermoregulation. Here we provide a genetic explanation for the poor thermoregulation in pigs as we demonstrate that the gene for uncoupling protein 1 (UCP1) was disrupted in the pig lineage. UCP1 is exclusively expressed in brown adipose tissue and plays a crucial role for thermogenesis by uncoupling oxidative phosphorylation. We used long-range PCR and genome walking to determine the complete genome sequence of pig UCP1. An alignment with human UCP1 revealed that exons 3 to 5 were eliminated by a deletion in the pig sequence. The presence of this deletion was confirmed in all tested domestic pigs, as well as in European wild boars, Bornean bearded pigs, wart hogs, and red river hogs. Three additional disrupting mutations were detected in the remaining exons. Furthermore, the rate of nonsynonymous substitutions was clearly elevated in the pig sequence compared with the corresponding sequences in humans, cattle, and mice, and we used this increased rate to estimate that UCP1 was disrupted about 20 million years ago.

Additional References

RELATED GEPHE

Related Genes

1 (Prolactin receptor) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^40674^/and+Trait=Thermoregulation/or+Taxon ID=^9822^/and+Trait=Thermoregulation/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Note that the derived phenotype may have appeared during evolution due to another mutation; and that the mutation described here might have occurred subsequently; as a neutral mutation.