

GEPHE SUMMARY

	Gephebase Gene		GepheID
Uncoupling protein 1 (UCP1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="Uncoupling protein 1 (UCP1)"#gephebase-summary-title)	GP00001162		
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category="Physiology"#gephebase-summary-title)		Trait	
		Thermoregulation (brown adipose tissue) (<a (brown="" adipose="" href="https://www.gephebase.org/search-criteria?/and+Trait=" thermoregulation="" tissue)"#gephebase-summary-title"="">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 (<i>S.barbatus</i>); wart hog (<i>Phacochoerus africanus</i>); and red river hog(<i>Potamochoerus porcus</i>).	
	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		Taxon B	
	Latin Name		Latin Name
Mammalia (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Mammalia"#gephebase-summary-title)	Sus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Sus"#gephebase-summary-title)		
	Common Name		Common Name
mammals	-		
	Synonyms		Synonyms
mammals	-		
	Rank		Rank
class	genus		
	Lineage		Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota	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		Parent
Amniota (amniotes) - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32524)	Suidae (pigs) - (Rank: family) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9821)		
	NCBI Taxonomy ID		NCBI Taxonomy ID
40674 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 40674)	9822 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9822)		
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No	No		

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Ucp1		P12242 (http://www.uniprot.org/uniprot/P12242)	
	Synonyms		GenebankID or UniProtKB
Ucp; Slc25a7; A1385626		AAD26529 (https://www.ncbi.nlm.nih.gov/nuccore/AAD26529)	
	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			

(<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)		
No matches found.		Related Haplotypes

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.