

GEPHE SUMMARY

Gephebase Gene
ATP4B

Entry Status
Published

GepheID
GP00001909

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Digestion (absence of stomach)

Trait State in Taxon A
presence of stomach and gastric acid production

Trait State in Taxon B
loss of stomach and no gastric acid production

Ancestral State
Taxon A

Taxonomic Status
Intergeneric or Higher

Taxon A #1

Latin Name

Monodelphis domestica

Common Name

gray short-tailed opossum

Synonyms

gray short-tailed opossum; Monodelphis domesticus

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Metatheria; Didelphimorphia; Didelphidae; Didelphinae; Monodelphis

Parent

Monodelphis (short-tailed opossums) - (Rank: genus)

NCBI Taxonomy ID

13616

is Taxon A an Intraspecies?

No

Taxon B

Latin Name

Ornithorhynchus anatinus

Common Name

platypus

Synonyms

platypus; duck-billed platypus; duckbill platypus; Ornithorhynchus anatinus

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Prototheria; Monotremata; Ornithorhynchidae; Ornithorhynchus

Parent

Ornithorhynchus () - (Rank: genus)

NCBI Taxonomy ID

9258

is Taxon B an Intraspecies?

No

Taxon A #2

Latin Name

Homo sapiens

Common Name

human

Synonyms

human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens

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; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo

Parent

Homo () - (Rank: genus)

NCBI Taxonomy ID

9606

is Taxon A an Intraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

ATP4B

UniProtKB Homo sapiens

P51164

Synonyms

ATP6B

GenebankID or UniProtKB

String

9606.ENSP00000334216

Sequence Similarities

Belongs to the X(+)/potassium ATPases subunit beta family.

GO - Molecular Function

GO:0001671 : ATPase activator activity

GO:0008900 : potassium:proton exchanging ATPase activity

GO - Biological Process

GO:0007155 : cell adhesion

GO:0034220 : ion transmembrane transport

GO:0032496 : response to lipopolysaccharide

GO:0030007 : cellular potassium ion homeostasis

GO:0006883 : cellular sodium ion homeostasis

GO:1990573 : potassium ion import across plasma membrane

GO:0036376 : sodium ion export across plasma membrane

GO:0010243 : response to organonitrogen compound

GO:0010248 : establishment or maintenance of transmembrane electrochemical gradient

GO - Cellular Component

GO:0005886 : plasma membrane

GO:0005890 : sodium:potassium-exchanging ATPase complex

Mutation #1

Presumptive Null

Yes

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsense

Molecular Details of the Mutation

two nonsense mutations and one deletion causing a frameshift in exon 7

Experimental Evidence

Candidate Gene

	Taxon A	Taxon B	Position
Codon	-	TAA	-
Amino-acid	Tyr	STP	98

Main Reference

Loss of genes implicated in gastric function during platypus evolution. (2008)

Authors

Ordoñez GR; Hillier LW; Warren WC; Grützner F; López-Otin C; Puente XS

Abstract

The duck-billed platypus (*Ornithorhynchus anatinus*) belongs to the mammalian subclass Prototheria, which diverged from the Theria line early in mammalian evolution. The platypus genome sequence provides a unique opportunity to illuminate some aspects of the biology and evolution of these animals.

We show that several genes implicated in food digestion in the stomach have been deleted or inactivated in platypus. Comparison with other vertebrate genomes revealed that the main genes implicated in the formation and activity of gastric juice have been lost in platypus. These include the aspartyl proteases pepsinogen A and pepsinogens B/C, the hydrochloric acid secretion stimulatory hormone gastrin, and the alpha subunit of the gastric H⁺/K⁺-ATPase. Other genes implicated in gastric functions, such as the beta subunit of the H⁺/K⁺-ATPase and the aspartyl protease cathepsin E, have been inactivated because of the acquisition of loss-of-function mutations. All of these genes are highly conserved in vertebrates, reflecting a unique pattern of evolution in the platypus genome not previously seen in other mammalian genomes.

The observed loss of genes involved in gastric functions might be responsible for the anatomical and physiological differences in gastrointestinal tract between monotremes and other vertebrates, including small size, lack of glands, and high pH of the monotreme stomach. This study contributes to a better understanding of the mechanisms that underlie the evolution of the platypus genome, might extend the less-is-more evolutionary model to monotremes, and provides novel insights into the importance of gene loss events during mammalian evolution.

Additional References

Mutation #2

Presumptive Null

Yes

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsense

Molecular Details of the Mutation

two nonsense mutations and one deletion causing a frameshift in exon 7

Experimental Evidence

Candidate Gene

	Taxon A	Taxon B	Position
Codon	AAG	TAG	-
Amino-acid	Lys	STP	153

Main Reference

Loss of genes implicated in gastric function during platypus evolution. (2008)

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Additional References

Mutation #3

Presumptive Null

Yes

Molecular Type

Coding

Aberration Type

Deletion

Deletion Size

-

Molecular Details of the Mutation

two nonsense mutations and one deletion causing a frameshift in exon 7

Experimental Evidence

Candidate Gene

Main Reference

Loss of genes implicated in gastric function during platypus evolution. (2008)

Authors

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Abstract

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Additional References

RELATED GEPHE

Related Genes

7 (ATP4A, gastrin, neurogenin 3, pepsinogen A, pepsinogen B, pepsinogen C, cathepsin E)

Related Haplotypes

No matches found.

COMMENTS

lack of acid secretion in the platypus stomach - this is a characteristic feature of monotremes whose gastric juice is above pH 6