GEPHE SUMMARY

Gephebase Gene

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

Entry Status

Published

GepheID

GP00001910

Main curator

Courtier

PHENOTYPIC CHANGE

Trait Category

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

Trait

Digestion (absence of stomach) (https://www.gephebase.org/search-criteria?/and+Trait=^Digestion (absence of stomach)^#gephebase-summary-title)

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

Taxon A #1

Latin Name

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

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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 13615)

NCBI Taxonomy ID

13616 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 13616)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

platypus

Synonyms

platypus; duck-billed platypus; duckbill platypus; Ornythorhynchus 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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9257)

NCBI Taxonomy ID

9258 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9258)

is Taxon B an Infraspecies?

No

Taxon A #2

Latin Name

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

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)
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID

9606
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon A an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

CTSE

Synonyms

CATE

String

9606.ENSP00000350911
(http://string-db.org/newstring_cgi/show_network_section.pl?identifier=
9606.ENSP00000350911)

Sequence Similarities

Belongs to the peptidase A1 family.

GO - Molecular Function

GO:0042803 : protein homodimerization activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0004190 : aspartic-type endopeptidase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004190)
GO:0008233 : peptidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008233)

GO - Biological Process

GO:0006508 : proteolysis (https://www.ebi.ac.uk/QuickGO/term/GO:0006508)
GO:0019886 : antigen processing and presentation of exogenous peptide antigen via MHC class II (https://www.ebi.ac.uk/QuickGO/term/GO:0019886)
GO:0016540 : protein autoprocessing
(https://www.ebi.ac.uk/QuickGO/term/GO:0016540)
GO:0030163 : protein catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0030163)

GO - Cellular Component

GO:0005768 : endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005768)

UniProtKB Homo sapiens

P14091 (http://www.uniprot.org/uniprot/P14091)

GenebankID or UniProtKB

()

Mutation #1

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

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

premature stop codon in exon 7 (Lys295Ter) and deletion leading to the loss of six of its nine exons

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Lys	STP	295

Main Reference

Loss of genes implicated in gastric function during platypus evolution. (2008) (https://pubmed.ncbi.nlm.nih.gov/18482448)

Authors

OrdoÃ±ez GR; Hillier LW; Warren WC; GrÃ¼tzner F; LÃ³pez-OtÃn 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 (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

-

Molecular Details of the Mutation

deletion leading to the loss of six of its nine exons - the high abundance of repetitive elements in the CTSE region (more than 3.8 interspersed elements per kilobase as compared with 2 for the genome average) might have contributed to the deletion of six out of the nine exons of CTSE by nonallelic homologous recombination between these repetitive elements - the CTSE gene has been disrupted by the insertion of long interspersed elements (LINEs) and short interspersed elements (SINEs) in exons 7 and 9; disrupting the protein coding region - Exon 9 was disrupted by the insertion of a LINE2 Plat1m element which was further disrupted by the insertion of a SINE Mon1f3 element

Experimental Evidence

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

Main Reference

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Additional References

RELATED GEPHE

Related Genes

7 (ATP4A, gastrin, neurogenin 3, pepsinogen A, pepsinogen B, pepsinogen C, ATP4B) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^13616^/and+Trait=Digestion/or+Taxon ID=^9606^/and+Trait=Digestion/or+Taxon ID=^9258^/and+Trait=Digestion/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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