

GEPHE SUMMARY

| | | | |
|---|---|-----------------------------------|------------------------------------|
| <p>ATP4B (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^ATP4B^#gephebase-summary-title)</p> <p>Published</p> | <p>Gephebase Gene</p> <p>Entry Status</p> | <p>GP00001909</p> <p>Courtier</p> | <p>GepheID</p> <p>Main curator</p> |
|---|---|-----------------------------------|------------------------------------|

PHENOTYPIC CHANGE

| | |
|---|---|
| <p>Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Physiology^#gephebase-summary-title)</p> <p>Digestion (absence of stomach) (https://www.gephebase.org/search-criteria?/and+Trait=^Digestion+absence+of+stomach^#gephebase-summary-title)</p> <p>presence of stomach and gastric acid production</p> <p>loss of stomach and no gastric acid production</p> <p>Taxon A</p> <p>Intergeneric or Higher (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intergeneric+or+Higher^#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> |
|---|---|

| Taxon A #1 | Latin Name |
|---|-----------------------------|
| Monodelphis domestica (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Monodelphis+domestica^#gephebase-summary-title) | |
| gray short-tailed opossum | Common Name |
| gray short-tailed opossum; Monodelphis domesticus | Synonyms |
| species | Rank |
| 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 | Lineage |
| Monodelphis (short-tailed opossums) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=13615) | Parent |
| 13616 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=13616) | NCBI Taxonomy ID |
| No | is Taxon A an Intraspecies? |

| Taxon B | Latin Name |
|---|-----------------------------|
| Ornithorhynchus anatinus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ornithorhynchus+anatinus^#gephebase-summary-title) | |
| platypus | Common Name |
| platypus; duck-billed platypus; duckbill platypus; Ornithorhynchus anatinus | Synonyms |
| species | Rank |
| cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Prototheria; Monotremata; Ornithorhynchidae; Ornithorhynchus | Lineage |
| Ornithorhynchus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9257) | Parent |
| 9258 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9258) | NCBI Taxonomy ID |
| No | is Taxon B an Intraspecies? |

| Taxon A #2 | Latin Name |
|--|-------------|
| Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title) | |
| human | Common Name |
| 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 | Synonyms |
| species | Rank |
| 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 | Lineage |

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 Intraspecies?

No

GENOTYPIC CHANGE

| | | | |
|---|-------------------------|--|-------------------------|
| ATP4B | Generic Gene Name | P51164 (http://www.uniprot.org/uniprot/P51164) | UniProtKB Homo sapiens |
| ATP6B | Synonyms | 0 | GenebankID or UniProtKB |
| 9606.ENSP00000334216 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000334216) | String | | |
| Belongs to the X(+)/potassium ATPases subunit beta family. | Sequence Similarities | | |
| GO:0001671 : ATPase activator activity (https://www.ebi.ac.uk/QuickGO/term/GO:0001671) | GO - Molecular Function | | |
| GO:0008900 : potassium;proton exchanging ATPase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008900) | | | |
| GO:0007155 : cell adhesion (https://www.ebi.ac.uk/QuickGO/term/GO:0007155) | GO - Biological Process | | |
| GO:0034220 : ion transmembrane transport (https://www.ebi.ac.uk/QuickGO/term/GO:0034220) | | | |
| GO:0032496 : response to lipopolysaccharide (https://www.ebi.ac.uk/QuickGO/term/GO:0032496) | | | |
| GO:0030007 : cellular potassium ion homeostasis (https://www.ebi.ac.uk/QuickGO/term/GO:0030007) | | | |
| GO:0006883 : cellular sodium ion homeostasis (https://www.ebi.ac.uk/QuickGO/term/GO:0006883) | | | |
| GO:1990573 : potassium ion import across plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:1990573) | | | |
| GO:0036376 : sodium ion export across plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0036376) | | | |
| GO:0010243 : response to organonitrogen compound (https://www.ebi.ac.uk/QuickGO/term/GO:0010243) | | | |
| GO:0010248 : establishment or maintenance of transmembrane electrochemical gradient (https://www.ebi.ac.uk/QuickGO/term/GO:0010248) | | | |
| GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886) | GO - Cellular Component | | |
| GO:0005890 : sodium;potassium-exchanging ATPase complex (https://www.ebi.ac.uk/QuickGO/term/GO:0005890) | | | |

Mutation #1

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

Presumptive Null

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

Molecular Type

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

Aberration Type

Nonsense

SNP Coding Change

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

Molecular Details of the Mutation

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

Experimental Evidence

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

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

Main Reference

Authors

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

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

Presumptive Null

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

Molecular 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#gephebase-summary-title))

Aberration Type

Nonsense

SNP Coding Change

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

Molecular Details of the Mutation

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

Experimental Evidence

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

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

Main Reference

Ordoñez GR; Hillier LW; Warren WC; Grätzner F; López-Otáñez C; Puente XS

Authors

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.

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Additional References

Mutation #3

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

Presumptive Null

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

Molecular Type

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

Aberration Type

-

Deletion Size

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

Molecular Details of the Mutation

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

Experimental Evidence

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

Main Reference

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

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.

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Additional References

RELATED GEPHE

Related Genes

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

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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