

GEPHE SUMMARY

	Gephebase Gene	GepheID
enamelin (ENAM) (https://www.gephebase.org/search-criteria/?and+Gene Gephebase=^enamelin (ENAM)^#gephebase-summary-title)	GP00001939	
Published	Entry Status	Main curator

PHENOTYPIC CHANGE

	Trait Category	
Physiology (https://www.gephebase.org/search-criteria/?and+Trait Category=^Physiology^#gephebase-summary-title)	Trait	
Tooth absence (no enamel production) (https://www.gephebase.org/search-criteria/?and+Trait=^Tooth+absence+(no+enamel+production)^#gephebase-summary-title)	Trait State in Taxon A	
presence of teeth	Trait State in Taxon B	
absence of teeth	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		
Cetacea (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Cetacea^#gephebase-summary-title)	Latin Name	Latin Name
whales	Common Name	Common Name
whales; cetaceans; whale; whales, dolphins, and porpoises	Synonyms	Synonyms
order	Rank	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla	Lineage	Lineage
Cetartiodactyla (whales, hippos, ruminants, pigs, camels etc.) - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=91561)	Parent	Parent
9721 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9721)	NCBI Taxonomy ID	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	is Taxon B an Infraspecies?
Taxon B #1		
Eubalaena glacialis (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Eubalaena+glacialis^#gephebase-summary-title)	Latin Name	Latin Name
North Atlantic right whale	Common Name	Common Name
Balaena glacialis; North Atlantic right whale; northern right whale; Eubalaena glacialis (Mueller, 1776)	Synonyms	Synonyms
species	Rank	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Cetacea; Mysticeti; Balaenidae; Eubalaena	Lineage	Lineage
Eubalaena () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=27605)	Parent	Parent
27606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=27606)	NCBI Taxonomy ID	NCBI Taxonomy ID
No	is Taxon B an Infraspecies?	is Taxon B an Infraspecies?
Taxon B #2		
Megaptera novaeangliae (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Megaptera+novaeangliae^#gephebase-summary-title)	Latin Name	Latin Name
humpback whale	Common Name	Common Name
humpback whale; Megaptera novaeangliae	Synonyms	Synonyms
species	Rank	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Cetacea; Mysticeti; Balaenopteridae; Megaptera	Lineage	Lineage
Megaptera () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9772)	Parent	Parent

9773

<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9773>

is Taxon B an Infraspecies?

No

Taxon B #3

Latin Name

Eschrichtius robustus<https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=%5Ceschrictius+robustus%5C#gephebase-summary-title>

Common Name

grey whale

Synonyms

Balaenoptera robusta; *Eschrichtius gibbosus*; grey whale; California gray whale; gray whale

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; Laurasiatheria; Cetartiodactyla; Cetacea; Mysticeti; Eschrichtiidae; *Eschrichtius*

Parent

Eschrichtius () - (Rank: genus)<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9763>

NCBI Taxonomy ID

9764

<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9764>

is Taxon B an Infraspecies?

No

Taxon B #4

Latin Name

Caperea marginata<https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=%5CCaperea+marginata%5C#gephebase-summary-title>

Common Name

pygmy right whale

Synonyms

pygmy right whale

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; Laurasiatheria; Cetartiodactyla; Cetacea; Mysticeti; Neobalaenidae; *Caperea*

Parent

Caperea () - (Rank: genus)<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 27603>

NCBI Taxonomy ID

27604

<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 27604>

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

UniProtKB Homo sapiens

ENAM

<http://www.uniprot.org/uniprot/Q9NRM1>

Synonyms

GenebankID or UniProtKB

ADA1; Al1C; AlH2

0

String

9606.ENSP00000379383

http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000379383

Sequence Similarities

GO - Molecular Function

GO:0030345 : structural constituent of tooth enamel
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0030345>)

GO - Biological Process

GO:0044267 : cellular protein metabolic process
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0044267>)

GO:0043687 : post-translational protein modification
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0043687>)

GO:0031214 : biomineral tissue development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0031214>)

GO:0036305 : ameloblast differentiation
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0036305>)

GO:0097186 : amelogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0097186>)

GO:0070175 : positive regulation of enamel mineralization
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0070175>)

GO:0022604 : regulation of cell morphogenesis
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0022604>)

GO - Cellular Component

GO:0031012 : extracellular matrix (<https://www.ebi.ac.uk/QuickGO/term/GO:0031012>)

GO:0005788 : endoplasmic reticulum lumen
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0005788>)

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

1-9 bp

Molecular Details of the Mutation

1-bp deletion. Various frameshift mutations were found in the distinct species.

Experimental Evidence

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

Main Reference

Molecular decay of the tooth gene Enamelin (ENAM) mirrors the loss of enamel in the fossil record of placental mammals. (2009) (<https://pubmed.ncbi.nlm.nih.gov/19730686>)

Authors

Meredith RW; Gatesy J; Murphy WJ; Ryder OA; Springer MS

Abstract

Vestigial structures occur at both the anatomical and molecular levels, but studies documenting the co-occurrence of morphological degeneration in the fossil record and molecular decay in the genome are rare. Here, we use morphology, the fossil record, and phylogenetics to predict the occurrence of "molecular fossils" of the enamelin (ENAM) gene in four different orders of placental mammals (Tubulidentata, Pholidota, Cetacea, Xenarthra) with toothless and/or enamelless taxa. Our results support the "molecular fossil" hypothesis and demonstrate the occurrence of frameshift mutations and/or stop codons in all toothless and enamelless taxa. We then use a novel method based on selection intensity estimates for codons (omega) to calculate the timing of iterated enamel loss in the fossil record of aardvarks and pangolins, and further show that the molecular evolutionary history of ENAM predicts the occurrence of enamel in basal representatives of Xenarthra (sloths, anteaters, armadillos) even though frameshift mutations are ubiquitous in ENAM sequences of living xenarthrans. The molecular decay of ENAM parallels the morphological degeneration of enamel in the fossil record of placental mammals and provides manifest evidence for the predictive power of Darwin's theory.

Additional References

Pseudogenization of the tooth gene enamelysin (MMP20) in the common ancestor of extant baleen whales. (2011) (<https://pubmed.ncbi.nlm.nih.gov/20861053>)

RELATED GEPHE

Related Genes

3 (ameloblastin (AMBN), amelogenin (AMEL), enamelysin (MMP20)) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=%9721%/and+Trait=Tooth+absence/or+Taxon+ID=%27606%/and+Trait=Tooth+absence/or+Taxon+ID=%9773%/and+Trait=Tooth+absence/or+Taxon+ID=%9764%/and+Trait=Tooth+absence/or+Taxon+ID=%27604%/and+Trait=Tooth+absence/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

1 ([https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=%enamelin+\(ENAM\)%/and+Taxon+ID=%9721%/or+Gene+Gephebase=%enamelin+\(ENAM\)%/and+Taxon+ID=%27606%/or+Gene+Gephebase=%enamelin+\(ENAM\)%/and+Taxon+ID=%9773%/or+Gene+Gephebase=%enamelin+\(ENAM\)%/and+Taxon+ID=%9764%/or+Gene+Gephebase=%enamelin+\(ENAM\)%/and+Taxon+ID=%27604%#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=%enamelin+(ENAM)%/and+Taxon+ID=%9721%/or+Gene+Gephebase=%enamelin+(ENAM)%/and+Taxon+ID=%27606%/or+Gene+Gephebase=%enamelin+(ENAM)%/and+Taxon+ID=%9773%/or+Gene+Gephebase=%enamelin+(ENAM)%/and+Taxon+ID=%9764%/or+Gene+Gephebase=%enamelin+(ENAM)%/and+Taxon+ID=%27604%#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

