GEPHE SUMMARY

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

GepheID
GP00001942

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

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

Trait
Tooth composition (no enamel production) (https://www.gephebase.org/search-criteria?/and+Trait=^Tooth composition (no enamel production)^#gephebase-summary-title)

Trait State in Taxon A
presence of enamel

Trait State in Taxon B
absence of enamel in 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

Latin Name
Physeter catodon (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Physeter catodon^#gephebase-summary-title)

Common Name
sperm whale

Synonyms
Physeter macrocephalus; sperm whale; Physeter catodon Linnaeus, 1758

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; Odontoceti; Physeteridae; Physeter

Parent
Physeter () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9753)

NCBI Taxonomy ID
9755 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9755)

is Taxon A an Infraspecies?
No

Taxon B #1

Latin Name
Kogia sima (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Kogia sima^#gephebase-summary-title)

Common Name
dwarf sperm whale

Synonyms
Kogia simus; dwarf sperm whale; Kogia sima (Owen, 1866)

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; Odontoceti; Physeteridae; Kogia

Parent
Kogia () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9751)

NCBI Taxonomy ID
9752 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9752)

is Taxon B an Infraspecies?
No

Taxon B #2

Latin Name
Kogia breviceps (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Kogia breviceps^#gephebase-summary-title)

Common Name
pygmy sperm whale

Synonyms
pygmy sperm 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; Odontoceti; Physeteridae; Kogia

Parent
Kogia () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9751)

NCBI Taxonomy ID

27615
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 27615)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

ENAM

Synonyms

ADAI; AI1C; AIH2

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)

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

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

two frameshift mutations - 1-bp deletion at position 2343 and 2-bp deletion at position 4034-4035 of the alignment

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)

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

UniProtKB Homo sapiens

Q9NRM1 (http://www.uniprot.org/uniprot/Q9NRM1)

GenebankID or UniProtKB

()

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

two frameshift mutations - 1-bp deletion at position 2343 and 2-bp deletion at position 4034-4035 of the alignment

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

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@ParrallelEvolution in baleen whales. There is an additional 2-bp deletion in the coding region in Kogia simus