GEPHE SUMMARY

Gephebase Gene
enamelin (ENAM)

GephelD
GP00001942

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Tooth composition (no enamel production)

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

Taxon A

Latin Name
Physeter catodon

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)

NCBI Taxonomy ID
9755

is Taxon A an Infraspecies?
No

Taxon B #1

Latin Name
Kogia sima

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)

NCBI Taxonomy ID
9752

is Taxon B an Infraspecies?
No

Taxon B #2

Latin Name
Kogia breviceps

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)

NCBI Taxonomy ID
27615

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
ENAM

Synonyms
ADAI; AI1C; AIH2

String
9606.ENSP00000379383

Sequence Similarities
-

GO - Molecular Function
GO:0030345 : structural constituent of tooth enamel

GO - Biological Process
GO:0044267 : cellular protein metabolic process
GO:0043687 : post-translational protein modification
GO:0031214 : biomineral tissue development
GO:0036305 : ameloblast differentiation
GO:0097186 : amelogenesis
GO:0070175 : positive regulation of enamel mineralization
GO:0022604 : regulation of cell morphogenesis

GO - Cellular Component
GO:0031012 : extracellular matrix
GO:0005788 : endoplasmic reticulum lumen

UniProtKB Homo sapiens
Q9NRM1

GenebankID or UniProtKB

Mutation #1
Presumptive Null
Yes

Molecular Type
Coding

Aberration Type
Deletion

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

Main Reference
Molecular decay of the tooth gene Enamelin (ENAM) mirrors the loss of enamel in the fossil record of placental mammals. (2009)

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)

Mutation #2
Presumptive Null
Yes

Molecular Type
Coding

Aberration Type
Deletion

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

Main Reference

Molecular decay of the tooth gene Enamelin (ENAM) mirrors the loss of enamel in the fossil record of placental mammals. (2009)

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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