

GEPHE SUMMARY

<p>enamelin (ENAM) (https://www.gephebase.org/search-criteria?/and+GeneGephebase=enamelin (ENAM)^#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001942</p> <p>Courtier</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=Physiology^#gephebase-summary-title)</p> <p>Tooth composition (no enamel production) (https://www.gephebase.org/search-criteria?/and+Trait=Tooth composition (no enamel production)^#gephebase-summary-title)</p> <p>presence of enamel</p> <p>absence of enamel in teeth</p> <p>Taxon A</p> <p>Intergeneric or Higher (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=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>
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Taxon A	
<p>Physeter catodon (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=Physeter catodon^#gephebase-summary-title)</p> <p>sperm whale</p> <p>Physeter macrocephalus; sperm whale; Physeter catodon Linnaeus, 1758</p> <p>species</p> <p>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</p> <p>Physeter () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9753)</p> <p>9755 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9755)</p> <p>No</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Intraspecies?</p>

Taxon B #1	
<p>Kogia sima (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=Kogia sima^#gephebase-summary-title)</p> <p>dwarf sperm whale</p> <p>Kogia simus; dwarf sperm whale; Kogia sima (Owen, 1866)</p> <p>species</p> <p>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</p> <p>Kogia () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9751)</p> <p>9752 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9752)</p> <p>No</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p>

Taxon B #2	
<p>Kogia breviceps (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=Kogia breviceps^#gephebase-summary-title)</p> <p>pygmy sperm whale</p> <p>pygmy sperm whale</p> <p>species</p> <p>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</p> <p>Kogia () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9751)</p> <p>NCBI Taxonomy ID</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p>

27615

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

is Taxon B an Intraspecies?

No

GENOTYPIC CHANGE

ENAM	Generic Gene Name	Q9NRM1 (http://www.uniprot.org/uniprot/Q9NRM1)	UniProtKB Homo sapiens
ADAI; AI1C; AIH2	Synonyms	0	GenebankID or UniProtKB
9606.ENSPO0000379383 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000379383)	String		
-	Sequence Similarities		
GO:0030345 : structural constituent of tooth enamel (https://www.ebi.ac.uk/QuickGO/term/GO:0030345)	GO - Molecular Function		
GO:0044267 : cellular protein metabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0044267)	GO - Biological Process		
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:0031012 : extracellular matrix (https://www.ebi.ac.uk/QuickGO/term/GO:0031012)	GO - Cellular Component		
GO:0005788 : endoplasmic reticulum lumen (https://www.ebi.ac.uk/QuickGO/term/GO:0005788)			

Mutation #1

Yes (<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>)

Molecular Type

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

Aberration Type

1-9 bp

Deletion Size

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

Molecular Details of the Mutation

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

Experimental Evidence

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>)

Main Reference

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

Authors

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 enamel 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.

Abstract

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

Additional References

Mutation #2

Yes (<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>)

Molecular Type

Aberration 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 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](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=))

Main Reference

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Authors

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Abstract

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Additional References

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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