GEPHE SUMMARY

Gephebase Gene
enamelysin (MMP20) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^enamelysin (MMP20)^#gephebase-summary-title)

GepheID
GP00001941

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
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 A an Infraspecies?
No

Taxon B

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
MMP20

Synonyms
AI2A2; MMP-20

String
9606.ENSP00000260228 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000260228)

Sequence Similarities
Belongs to the peptidase M10A family.

GO - Molecular Function
GO:0004222 : metalloendopeptidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004222)
GO:0008270 : zinc ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008270)

GO - Biological Process
GO:0030198 : extracellular matrix organization (https://www.ebi.ac.uk/QuickGO/term/GO:0030198)

UniProtKB Homo sapiens
O60882 (http://www.uniprot.org/uniprot/O60882)

GenebankID or UniProtKB
()

GO:0006508 : proteolysis (https://www.ebi.ac.uk/QuickGO/term/GO:0006508)
GO:0030163 : protein catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0030163)
GO:0030574 : collagen catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0030574)
GO:0022617 : extracellular matrix disassembly
(https://www.ebi.ac.uk/QuickGO/term/GO:0022617)
GO:0097186 : amelogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0097186)
GO:0070173 : regulation of enamel mineralization
(https://www.ebi.ac.uk/QuickGO/term/GO:0070173)

GO - Cellular Component

GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0031012 : extracellular matrix (https://www.ebi.ac.uk/QuickGO/term/GO:0031012)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)

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

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

opal stop codon (TGA) in the propeptide-coding region of MMP20 exon 2 in a single individual of the pygmy sperm whale Kogia breviceps

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	TGA	-
Amino-acid	-	STP	-

Main Reference

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

Authors

Meredith RW; Gatesy J; Cheng J; Springer MS

Abstract

Whales in the suborder Mysticeti are filter feeders that use baleen to sift zooplankton and small fish from ocean waters. Adult mysticetes lack teeth, although tooth buds are present in foetal stages. Cladistic analyses suggest that functional teeth were lost in the common ancestor of crown-group Mysticeti. DNA sequences for the tooth-specific genes, ameloblastin (AMBN), enamelin (ENAM) and amelogenin (AMEL), have frameshift mutations and/or stop codons in this taxon, but none of these molecular cavities are shared by all extant mysticetes. Here, we provide the first evidence for pseudogenization of a tooth gene, enamelysin (MMP20), in the common ancestor of living baleen whales. Specifically, pseudogenization resulted from the insertion of a CHR-2 SINE retroposon in exon 2 of MMP20. Genomic and palaeontological data now provide congruent support for the loss of enamel-capped teeth on the common ancestral branch of crown-group mysticetes. The new data for MMP20 also document a polymorphic stop codon in exon 2 of the pygmy sperm whale (Kogia breviceps), which has enamel-less teeth. These results, in conjunction with the evidence for pseudogenization of MMP20 in Hoffmann's two-toed sloth (Choloepus hoffmanni), another enamel-less species, support the hypothesis that the only unique, non-overlapping function of the MMP20 gene is in enamel formation.

Additional References

RELATED GEPHE

Related Genes

1 (enamelin (ENAM)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9752^/and+Trait=Tooth composition/or+Taxon ID=^27615^/and+Trait=Tooth composition/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@ParallelEvolution in baleen whales.