GEPHE SUMMARY

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

GepheID
GP00001937

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 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

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

Common Name
whales

Synonyms
whales; cetaceans; whale; whales, dolphins, and porpoises

Rank
order

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

Parent
Cetartiodactyla (whales, hippos, ruminants, pigs, camels etc.) - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 91561)

NCBI Taxonomy ID
9721 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9721)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
baleen whales

Synonyms
baleen whales

Rank
suborder

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

Parent
Cetacea (whales) - (Rank: order) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9721)

NCBI Taxonomy ID
9761 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9761)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Amelx

Synonyms
Amg; ALGN; AMGL; AMGX; Amel; LRAP; Rgsc888

String
10090.ENSMUSP00000065966 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000065966)

Sequence Similarities
Belongs to the amelogenin family.

GO - Molecular Function
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)
GO:0005509 : calcium ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005509)
GO:0031402 : sodium ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031402)

UniProtKB Mus musculus
P63277 (http://www.uniprot.org/uniprot/P63277)

GenebankID or UniProtKB
()

GO:0046848 : hydroxyapatite binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0046848)
GO:0030345 : structural constituent of tooth enamel
(https://www.ebi.ac.uk/QuickGO/term/GO:0030345)

GO - Biological Process

GO:0007155 : cell adhesion (https://www.ebi.ac.uk/QuickGO/term/GO:0007155)
GO:0007165 : signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007165)
GO:0042475 : odontogenesis of dentin-containing tooth
(https://www.ebi.ac.uk/QuickGO/term/GO:0042475)
GO:0051260 : protein homooligomerization
(https://www.ebi.ac.uk/QuickGO/term/GO:0051260)
GO:0042127 : regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0042127)
GO:0034505 : tooth mineralization (https://www.ebi.ac.uk/QuickGO/term/GO:0034505)

GO - Cellular Component

GO:0005604 : basement membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005604)
GO:0032991 : protein-containing complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0032991)
GO:0009986 : cell surface (https://www.ebi.ac.uk/QuickGO/term/GO:0009986)
GO:0099080 : supramolecular complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0099080)

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

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

Molecular Details of the Mutation

J. Gatesy 2010 unpublished data cited in the main reference

Experimental Evidence

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

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

3 (ameloblastin (AMBN), enamelin (ENAM), enamelysin (MMP20)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9721^/and+Trait=Tooth absence/or+Taxon ID=^9761^/and+Trait=Tooth absence/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Exact species investigated not mentioned in the paper. Cladistic analyses suggest that functional teeth were lost in the common ancestor of crown-group Mysticeti. The amelogenin (AMEL) gene contains various gene null mutations in various species. it is possible that frameshift mutations and/or stop codons will be discovered in the unsequenced protein-coding regions of one or more of these extracellular matrix protein (EMP) genes. A second possibility is that one or more of these genes were initially silenced by mutations in a regulatory gene region on the ancestral mysticete branch, and that mutations in protein-coding regions accumulated subsequently on descendant branches within crown-group Mysticeti. A third possibility is that a different enamel- or tooth-specific gene was knocked out in the common ancestor of mysticetes and that AMBN ENAM and AMEL acquired molecular cavities on descendant branches within crown-group Mysticeti. Alternatively enamel may have been lost independently in several mysticete lineages rather than once in the common ancestor of crown mysticetes.