

GEPHE SUMMARY

	Gephebase Gene		GepheID
amelogenin (AMEL) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase= [^] amelogenin (AMEL) [^] #gephebase-summary-title)		GP00001937	
	Entry Status	Courtier	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category	
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category= [^] Physiology [^] #gephebase-summary-title)		
	Trait	
Tooth absence (no enamel production) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=<sup>^</sup>Tooth absence (no enamel production)<sup>^</sup>#gephebase-summary-title">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	Taxon B	Latin Name
Cetacea (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Cetacea [^] #gephebase-summary-title)		Mysticeti (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Mysticeti [^] #gephebase-summary-title)	
	Common Name		Common Name
whales		baleen whales	
	Synonyms		Synonyms
whales; cetaceans; whale; whales, dolphins, and porpoises		baleen whales	
	Rank		Rank
order		suborder	
	Lineage		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		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		Parent
Cetartiodactyla (whales, hippos, ruminants, pigs, camels etc.) - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=91561)		Cetacea (whales) - (Rank: order) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9721)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9721 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9721)		9761 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9761)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Amelx		P63277 (http://www.uniprot.org/uniprot/P63277)	
	Synonyms		GenebankID or UniProtKB
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)			

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 ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes))

Molecular Type

Coding ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding))

Aberration Type

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

Molecular Details of the Mutation

J. Gatesy 2010 unpublished data cited in the main reference

Experimental Evidence

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

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.

