

GEPHE SUMMARY

	Gephebase Gene	GephelD
enamelin (ENAM) (https://www.gephebase.org/search-criteria/?and+Gene Gephebase=^enamelin (ENAM)^#gephebase-summary-title)	GP00001943	
Published	Entry Status	Main curator

PHENOTYPIC CHANGE

	Trait Category		
	Trait		
	Trait State in Taxon A		
presence of enamel	Trait State in Taxon B		
absence of enamel	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		Taxon B	
	Latin Name		Latin Name
Elephantulus rufescens (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Elephantulus+rufescens^#gephebase-summary-title)	Orycteropus afer (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Orycteropus+afer^#gephebase-summary-title)		
	Common Name		Common Name
East African long-eared elephant shrew	aardvark		
	Synonyms		Synonyms
East African long-eared elephant shrew; rufous elephant shrew	aardvark; Orycteropus afer (Pallas, 1766)		
	Rank		Rank
species	species		
	Lineage		Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Afrotheria; Macroscelidea; Macroscelididae; Elephantulus	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Afrotheria; Tubulidentata; Orycteropodidae; Orycteropus		
	Parent		Parent
Elephantulus (long-eared elephant shrews) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 28736)	Orycteropus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9817)		
42151 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 42151)	9818 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9818)		
No	is Taxon A an Infraspecies?		is Taxon B an Infraspecies?

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
ENAM	Q9NRM1 (http://www.uniprot.org/uniprot/Q9NRM1)	
	Synonyms	GenebankID or UniProtKB
ADA1; AI1C; AIH2	0	
	String	
9606.ENSP00000379383 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000379383)		
	Sequence Similarities	
-		
	GO - Molecular Function	
GO:0030345 : structural constituent of tooth enamel (https://www.ebi.ac.uk/QuickGO/term/GO:0030345)		
	GO - Biological Process	
GO:0044267 : cellular protein metabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0044267)		
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 - Cellular Component

GO:0031012 : extracellular matrix (<https://www.ebi.ac.uk/QuickGO/term/GO:0031012>)
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
Insertion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Insertion^#gephebase-summary-title)	Aberration Type
1-9 bp	Insertion Size
1-bp insertion at position 2649 and 1-bp insertion at position 3678 and 1-bp deletion at position 4041	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 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.	Abstract
	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
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title)	Aberration Type
1-9 bp	Deletion Size
1-bp insertion at position 2649 and 1-bp insertion at position 3678 and 1-bp deletion at position 4041	Molecular Details of the Mutation
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)	Experimental Evidence
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	Additional References

Mutation #3

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)	Presumptive Null
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Coding (#gephebase-summary-title)	Molecular Type
Deletion (#gephebase-summary-title)	Aberration Type
1-9 bp	Deletion Size
1-bp insertion at position 2649 and 1-bp insertion at position 3678 and 1-bp deletion at position 4041	Molecular Details of the Mutation
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
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	Additional References

RELATED GEPHE

No matches found.	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

@ParallelEvolution in baleen whales. Teeth in the extant aardvark *Orycteropus afer* lack both enamel and a central pulp cavity and are composed of 1500 thin hexagonal tubes of dentin that are bound together by cementum