GEPHE SUMMARY Gephebase Gene GephelD amelogenin (AMEL) (https://www.gephebase.org/search-criteria?/and+Generals.com/search-criteria. GP00001933 Gephebase=^amelogenin (AMEL)^#gephebase-summary-title) Main curator Entry Status Courtier **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) (https://www.gephebase.org/searchcriteria?/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 Taxon B Latin Name Latin Name Paleosuchus palpebrosus Gallus gallus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Paleosuchus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Gallus palpebrosus^#gephebase-summary-title) gallus^#gephebase-summary-title) Common Name Common Name Cuvier's dwarf caiman chicken Synonyms Synonyms Cuvier's dwarf caiman; MNHN 7530; MNHN:7530 Gallus gallus domesticus; chicken; bantam; chickens Rank Rank species species Lineage Lineage cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Crocodylia; Alligatoridae; Caimaninae; Paleosuchus Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Parent Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus Paleosuchus () - (Rank: genus) Parent $(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=38657\,)$ Gallus () - (Rank: genus) NCBI Taxonomy ID $(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9030\)$ NCBI Taxonomy ID (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 84099) $(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9031\,)$ is Taxon A an Infraspecies? No is Taxon B an Infraspecies? Νo **GENOTYPIC CHANGE**

Generic Gene Name UniProtKB Mus musculus Amelx P63277 (http://www.uniprot.org/uniprot/P63277) GenebankID or UniProtKB Synonyms Amg; ALGN; AMGL; AMGX; Amel; LRAP; Rgsc888 0 String 10090.ENSMUSP00000065966

Belongs to the amelogenin family.

10090.ENSMUSP00000065966)

GO - Molecular Function

Sequence Similarities

GO:0042802: identical protein binding

(https://www.ebi.ac.uk/QuickGO/term/GO:0042802) $\mathsf{GO} \mathpunct{:} \mathsf{0042803} : \mathsf{protein} \ \mathsf{homodimerization} \ \mathsf{activity}$ (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)

Mutation #1

 $Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^\#gephebase-summary-title)$

Molecular Type

Presumptive Null

 $Coding \ (https://www.gephebase.org/search-criteria?/and+Molecular \ Type=^Coding^* \\ gephebase-summary-title)$

Aberration Type

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

Insertion Size

1-9 bp

Molecular Details of the Mutation

synteny of the corresponding region - two 2-bp insertions in exon 2 leading to a reading frameshift which changes the amino acids in the N-terminal region and results in a premature stop codon in exon 6

Experimental Evidence

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

Main Reference

Hen's teeth with enamel cap: from dream to impossibility. (2008) (https://pubmed.ncbi.nlm.nih.gov/18775069)

Authors

Sire JY; Delgado SC; Girondot M

Abstract

The ability to form teeth was lost in an ancestor of all modern birds, approximately 100-80 million years ago. However, experiments in chicken have revealed that the oral epithelium can respond to inductive signals from mouse mesenchyme, leading to reactivation of the odontogenic pathway. Recently, tooth germs similar to crocodile rudimentary teeth were found in a chicken mutant. These "chicken teeth" did not develop further, but the question remains whether functional teeth with enamel cap would have been obtained if the experiments had been carried out over a longer time period or if the chicken mutants had survived. The next odontogenetic step would have been tooth differentiation, involving deposition of dental proteins.

Using bioinformatics, we assessed the fate of the four dental proteins thought to be specific to enamel (amelogenin, AMEL; ameloblastin, AMBN; enamelin, ENAM) and to dentin (dentin sialophosphoprotein, DSPP) in the chicken genome. Conservation of gene synteny in amniotes allowed definition of target DNA regions in which we searched for sequence similarity. We found the full-length chicken AMEL and the only N-terminal region of DSPP, and both are invalidated genes. AMBN and ENAM disappeared after chromosomal rearrangements occurred in the candidate region in a bird ancestor.

These findings not only imply that functional teeth with enamel covering, as present in ancestral Aves, will never be obtained in birds, but they also indicate that these four protein genes were dental specific, at least in the last toothed ancestor of modern birds, a specificity which has been questioned in recent years.

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

 $Insertion \ (https://www.gephebase.org/search-criteria?/and+Aberration \ Type=`lnsertion`\#gephebase-summary-title)$

Aberration Type

Insertion Size

1-9 bp

Molecular Details of the Mutation

synteny of the corresponding region - two 2-bp insertions in exon 2 leading to a reading frameshift which changes the amino acids in the N-terminal region and results in a premature stop codon in exon 6

Experimental Evidence

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

Main Reference

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Additional References

Presumptive Null

Molecular Type

Aberration Type

Indel Size

Mutation #3

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

 $Coding \ (https://www.gephebase.org/search-criteria?/and+Molecular \ Type=^Coding^\#gephebase-summary-title)$

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

also numerous indels in exon 6

Molecular Details of the Mutation

Experimental Evidence

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

Main Reference

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Authors

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Additional References

RELATED GEPHE

Related Genes

3 (ameloblastin (AMBN), dentin sialophosphoprotein (DSPP), enamelin (ENAM)) (https://www.gephebase.org/search-criteria?/or+Taxon ID= $^84099^$ /and+Trait=Tooth absence/or+Taxon ID= $^9031^$ /and+Trait=Tooth absence/or+Taxon ID= $^9031^$ /and+Trait=Tooth absence/and+groupHaplotypes=true#gephebase-summary-title)

 ${\sf Related\ Haplotypes}$

No matches found.

EXTERNAL LINKS

COMMENTS

In zebrafinch AMEL exon 2 there is a deletion of 12 bases and a base substitution leading to a premature stop codon. The AMEL gene mutations in these two bird species indicate that this crucial gene for enamel formation has lost its functional constrainsts long before the split between Passeriformes and Galliformes (Sire et al unpublished data).