GEPHE SUMMARY

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

GepheID
GP00001933

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

Common Name
Cuvier's dwarf caiman

Synonyms
Cuvier's dwarf caiman; MNHN 7530; MNHN:7530

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Crocodylia; Alligatoridae; Caimaninae; Paleosuchus

Parent
Paleosuchus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 38657)

NCBI Taxonomy ID
84099 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 84099)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
chicken

Synonyms
Gallus gallus domesticus; chicken; bantam; chickens

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Galliformes; Phasianidae; Phasianinae; Gallus

Parent
Gallus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9030)

NCBI Taxonomy ID
9031 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9031)

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)

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

GenebankID or UniProtKB
()

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

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

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

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

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

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Additional References

Mutation #3

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

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

Indel Size

-

Molecular Details of the Mutation

also numerous indels 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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Authors

Sire JY; Delgado SC; Girondot M

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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/and+groupHaplotypes=true#gephebase-summary-title)

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).