



LJMU Research Online

Towle, IE, Irish, JD and De Groote, I

Amelogenesis imperfecta in the dentition of a wild chimpanzee.

<http://researchonline.ljmu.ac.uk/id/eprint/7572/>

Article

Citation (please note it is advisable to refer to the publisher's version if you intend to cite from this work)

Towle, IE, Irish, JD and De Groote, I (2017) Amelogenesis imperfecta in the dentition of a wild chimpanzee. Journal of Medical Primatology. ISSN 0047-2565

LJMU has developed **LJMU Research Online** for users to access the research output of the University more effectively. Copyright © and Moral Rights for the papers on this site are retained by the individual authors and/or other copyright owners. Users may download and/or print one copy of any article(s) in LJMU Research Online to facilitate their private study or for non-commercial research. You may not engage in further distribution of the material or use it for any profit-making activities or any commercial gain.

The version presented here may differ from the published version or from the version of the record. Please see the repository URL above for details on accessing the published version and note that access may require a subscription.

For more information please contact researchonline@ljmu.ac.uk

<http://researchonline.ljmu.ac.uk/>

Amelogenesis imperfecta in the dentition of a wild chimpanzee

Ian Towle¹, Joel D. Irish¹, Isabelle De Groot¹

¹ Research Centre in Evolutionary Anthropology and Palaeoecology, School of Natural Sciences and Psychology, John Moores University, Liverpool, United Kingdom, L3 3AF

Number of text pages: 7

Number of figures: 1

Abbreviated title: Amelogenesis imperfecta in primates

Key words: dental defects, hereditary disease, enamel hypoplasia

Contact details:

Ian Towle

Room 352, James Parsons Building, Byrom Street, Liverpool, L3 3AF

I.Towle@2014.ljmu.ac.uk

Abstract (50 words)

This report describes a case of Amelogenesis imperfecta in the dentition of a wild female chimpanzee. Amelogenesis imperfecta is a group of rare genetic conditions that create severe enamel defects, which, although well researched in humans, has not yet been investigated in wild non-human primates.

Introduction

Amelogenesis imperfecta (AI) is a group of genetic conditions that create enamel defects, affecting one in every 700 to 14,000 humans [1, 2]. A variety of different genetic mutations can be responsible for these heritable diseases [3-6]. Pitting and plane form enamel hypoplasia is common, but abnormal enamel density, thickness and coloration is also associated with different forms [7-16]. To differentiate between different types of AI, clinical, histological and radiographic methods have been used [2, 5, 10].

AI has yet to be implicitly stated to be present in a non-human primate dentition, however research has implicated genetic factors in the formation of severe defects and there are also descriptions of specimens that may fit with what is now classified as AI [17-19]. Given the large amount of mutations that can cause these heritable conditions in humans it is perhaps surprising that more examples of similar defects in primates have not been described. This likely reflects the much greater sample sizes as well as proportionally more research that is carried out on humans. In this study the dentitions of gorillas, chimpanzees and baboons are examined for signs of AI.

Materials and Methods

A total of 119 chimpanzee, 93 gorilla, and 43 baboon dentitions were analyzed, with both permanent (n= 5740) and deciduous (n= 1353) teeth being studied. All material is curated at the Powell-Cotton Museum in Kent, UK. The primates were wild shot and collected in the first half of the 20th century [20, 21]. As with most of the chimpanzee sample, the individual discussed in the sections below originates from the Batouri district, Cameroon.

Teeth were examined macroscopically with a 10x hand lens being used to confirm defects. The presence and position of pitting, plane, and linear forms of hypoplasia were recorded for each tooth. Other enamel defects such as coloration change, porosity and reduction in size are recorded, as well as congenitally missing and supernumerary teeth were recorded. Dental pathologies and unusual wear were also recorded for each dentition, including caries, calculus, periodontal disease and abscesses.

Results

Although many dentitions display enamel defects, with multiple types of hypoplasia commonly occurring, only one individual displays defects that are consistent with a diagnosis of AI. This specimen, M 299, is an adult female chimpanzee. Pitting enamel hypoplasia is visible on all anterior teeth as well as the maxillary first premolars and both sets of lower premolars (Fig. 1). There are no visible defects on the molars. Apart from the defects themselves the teeth appear normal in terms of size and morphology.

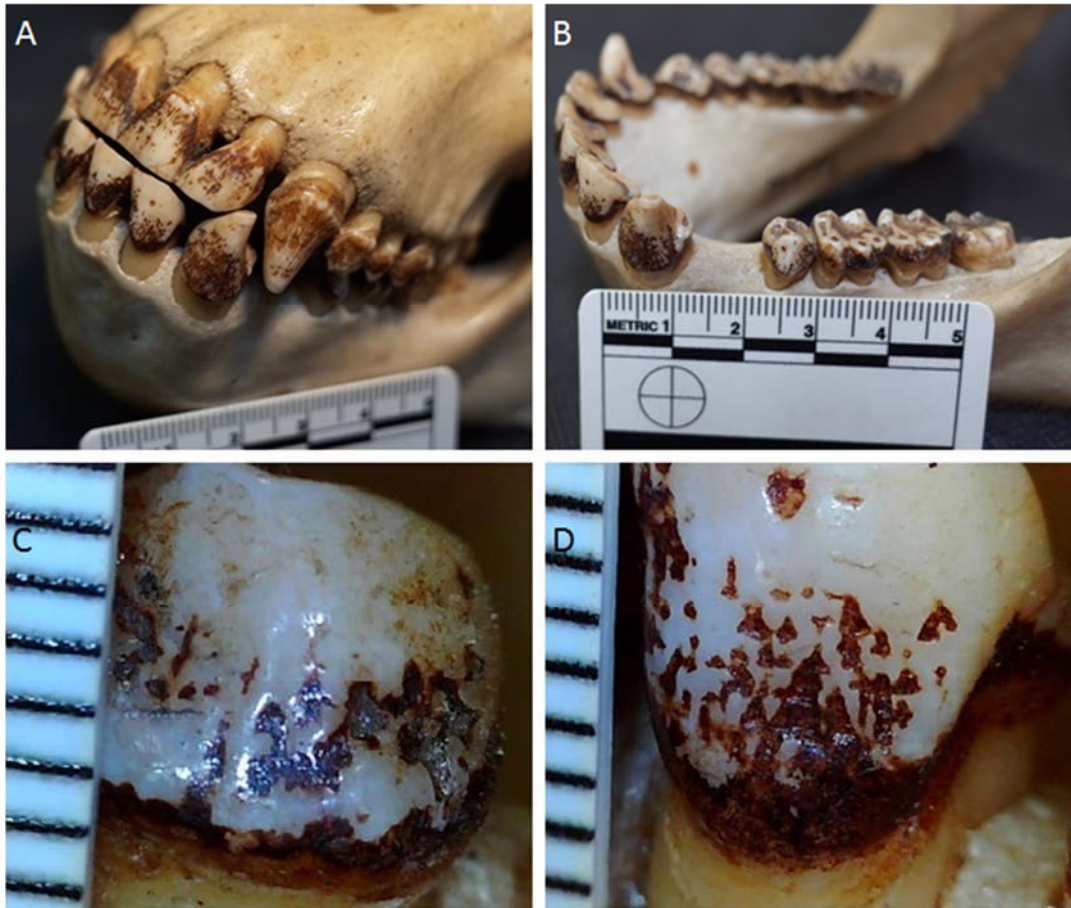


Figure 1. Female chimpanzee showing enamel defects and a congenitally missing tooth. A) buccal/Labial view of enamel defects on anterior teeth; B) congenitally missing left mandibular first premolar; C) buccal view of the lower right first premolar, with clear pitting/plane form defects; D) buccal view of the lower left lateral incisor, with clear pitting/plane form defects.

The pitting is irregular in both shape and size, but small circular depressions are most common (Fig. 1a,d), with some larger irregular defects that resemble plane form defects (Fig. 1c). Hypoplasia is most defined on the labial/buccal surface and the cervical half of the crown. The anterior teeth all appear to be equally effected, with roughly the same position and severity of defects (Fig. 1a,b). There is also a ‘mottled’ or ‘wavey’ appearance to the enamel on these teeth. The color of the enamel appears normal; however, dark post-

mortem coloration may mask defects. The mandibular left first premolar is congenitally absent (Fig. 1b). There is no evidence of caries, ante-mortem enamel fractures, periodontal disease, or severe occlusal attrition. Small amounts of calculus are visible on the posterior mandibular teeth.

Discussion

The pattern of defects and the teeth affected support a diagnosis of AI for M 299. The uniform pattern of defects on anterior teeth, and complete lack of defects on molars, strongly suggests these defects are not related to a physiological stress during development [2, 22, 23]. Additionally, the pattern of defects do not resemble those caused by congenital viruses; vitamin deficiencies, malnutrition, and fluorosis and other mineral contaminants in humans [24-26]. A congenitally missing tooth adds further support to the diagnosis of AI, as these two conditions are commonly found together [10].

Of the four main AI types that have been commonly categorized in humans, the characteristics on this chimpanzee's dentition match those of the Hypoplastic (Type 1) variety [9, 10, 27]. In line with this variety there is little or no coloration change visible, no obvious reduction in size or unusual interproximal spacing, and there is clear enamel pitting and plane form defects covering large areas of the crown in multiple teeth. Specifically the defects look indistinguishable from those recorded in humans as Type 1A [28, 29], which is characterized by enamel with pitting that varies from pinpoint to pinhead in size and is found predominantly on the buccal/labial surfaces of permanent teeth [30]. It is also common in this condition for some teeth to show no visible defects [28], likely explaining why the molars in this individual seem unaffected; however postmortem coloration and ante-mortem wear may mask defects. This type of AI creates the least amount of dental

issues for human patients [29]; therefore potentially explaining the relatively good health, except for the defects themselves, of this chimpanzees teeth.

Although other potential cases of AI in primates need to be confirmed, it is suggestive that the range of defects exhibited in non-human primates may be as diverse as in humans [e.g. 17, 19]. It is also likely that many examples have been overlooked in primate skeletal collections, particularly in light of recent DNA advancement showing just how diverse AI defects can be in humans. DNA analysis of this individual as well as additional observations of AI in the dentitions of other primates would further our understanding of these enamel genetic disorders in primates as a whole.

Acknowledgments

We thank I. Livne from the Powell-Cotton museum for access to the primate collection. This research was supported by a studentship to the first author from Liverpool John Moores University.

References

1. Sundell S, Koch G. Hereditary amelogenesis imperfecta. I. Epidemiology and clinical classification in a Swedish child population. *Swedish Dental Journal*. 1984;9(4):157-169.
2. Crawford PJ, Aldred M, & Bloch-Zupan A. Amelogenesis imperfecta. *Orphanet Journal of Rare Diseases*. 2007;2(1),17.
3. Kelley JL, Swanson WJ. Dietary change and adaptive evolution of enamelin in humans and among primates. *Genetics*. 2008;178(3),1595-1603.
4. Horvath JE, Ramachandran GL, Fedrigo O, Nielsen WJ, Babbitt CC, Clair EMS, Wall CE. Genetic comparisons yield insight into the evolution of enamel thickness during human evolution. *Journal of Human Evolution*. 2014;73,75-87.

5. Wang X, Zhao Y, Yang Y, Qin M. Novel ENAM and LAMB3 mutations in Chinese families with hypoplastic amelogenesis imperfecta. *PloS one*. 2015;10(3),e0116514.
6. Smith CE, Murillo G, Brookes SJ, Poulter JA, Silva S, Kirkham J, Mighell AJ. Deletion of amelotin exons 3–6 is associated with amelogenesis imperfecta. *Human Molecular Genetics*. 2016;ddw203.
7. Mårdh CK, Bäckman B, Holmgren G, Hu JC, Simmer JP, Forsman-Semb K. A nonsense mutation in the enamelin gene causes local hypoplastic autosomal dominant amelogenesis imperfecta (AIH2). *Human Molecular Genetics*. 2002;11(9),1069-1074.
8. Ozdemir D, Hart PS, Firatli E, Aren G, Ryu OH, Hart TC. Phenotype of ENAM mutations is dosage-dependent. *Journal of Dental Research*, 2005;84(11),1036-1041.
9. Wright JT. The diagnosis and treatment of dentinogenesis imperfecta and amelogenesis imperfecta. *Hellenic Dent J*. 1993;2,17-24.
10. Mehta DN, Shah J, Thakkar B. Amelogenesis imperfecta: Four case reports. *Journal of Natural Science, Biology, and Medicine*. 2013;4(2), 462.
11. Huckert M, Stoetzel C, Morkmued S, Laugel-Haushalter V, Geoffroy V, Muller J, Switala M. Mutations in the latent TGF-beta binding protein 3 (LTBP3) gene cause brachyolmia with amelogenesis imperfecta. *Human Molecular Genetics*. 2015;ddv053.
12. Wright JT. Analysis of a kindred with amelogenesis imperfecta. *Journal of Oral Pathology & Medicine*. 1985;14(5):366-374.
13. Aldred M, Savarirayan R, Crawford P. Amelogenesis imperfecta: A classification and catalogue for the 21st century. *Oral Diseases*. 2003;9(1):19-23.
14. Chamarthi V, Varma BR, Jayanthi M. Amelogenesis imperfecta: A clinician's challenge. *Journal of Indian Society of Pedodontics and Preventive Dentistry*. 2012;30(1), 70.
15. Schuurs A. Pathology of the hard dental tissues. John Wiley & Sons. 2012
16. Poulter JA, El-Sayed W, Shore RC, Kirkham J, Inglehearn CF, Mighell AJ. Whole-exome sequencing, without prior linkage, identifies a mutation in LAMB3 as a cause of dominant

- hypoplastic amelogenesis imperfecta. *European Journal of Human Genetics*. 2014;22(1),132-135.
17. Tomes CS. Partial suppression of teeth in a very hairy monkey (*Colobus caudatus*). *Trans. Odontoi. Soc. Gr. Brit.* 1898;30,30-35.
 18. Jones TS, Cave A JE. Diet, longevity and dental disease in the Sierra Leone chimpanzee. *Journal of Zoology*. 1960;135(1),147-155.
 19. Miles AEW, Grigson C. Colyer's Variations and Diseases of the Teeth of Animals. Cambridge University Press. 2003
 20. Guatelli-Steinberg D, Skinner M. Prevalence and etiology of linear enamel hypoplasia in monkeys and apes from Asia and Africa. *Folia Primatologica*. 2000;71(3),115-132.
 21. Macho GA, Lee-Thorp JA. Niche partitioning in sympatric Gorilla and Pan from Cameroon: Implications for life history strategies and for reconstructing the evolution of hominin life history. *PloS one*. 2014;9(7),e102794.
 22. Guatelli-Steinberg D. Micro-to Macroscopic. A companion to dental anthropology, 450. 2015
 23. Boughner JC, Der J, Kuykendall KL. A multivariate approach to assess variation in tooth mineralization using free-lived and captive-raised chimpanzees (*P. troglodytes*). *American Journal of Physical Anthropology*. 2015;158(3),452-462.
 24. Hillson S. Tooth development in human evolution and bioarchaeology. Cambridge University Press. 2014
 25. Ioannou S, Henneberg M, Henneberg RJ, Anson T. Diagnosis of mercurial teeth in a possible case of congenital syphilis and tuberculosis in a 19th century child skeleton. *Journal of Anthropology*. 2015;2015.
 26. Thylstrup A, Fejerskov O. Clinical appearance of dental fluorosis in permanent teeth in relation to histologic changes. *Community Dentistry and Oral Epidemiology*. 1978;6(6),315-328.

27. Shivhare P, Shankarnarayan L, Gupta A, Sushma P. Amelogenesis Imperfecta: A Review. *Journal of Advanced Oral Research*. 2016;7(1).
28. Witkop CJ. Amelogenesis imperfecta, dentinogenesis imperfecta and dentin dysplasia revisited: problems in classification. *Journal of Oral Pathology & Medicine*. 1988;17(9-10),547-553.
29. Seow WK. Clinical diagnosis and management strategies of amelogenesis imperfecta variants. *Pediatric Dentistry*. 1992;15(6),384-393.
30. Witkop CJ, Sauk JJ. Heritable defects of enamel. *Oral Facial Genetics*. 1976