

**Amelogenesis Imperfecta in an early twentieth century population from central
Portugal**

Wasterlain, S. N.^{1*} and Dias, G.J.²

¹ Department of Anthropology, University of Coimbra, 3000-056 Coimbra, Portugal

² Department of Anatomy and Structural Biology, School of Medical Sciences,
University of Otago, Dunedin, New Zealand

Short title: Amelogenesis imperfecta in Portuguese

*Correspondence to: Sofia N. Wasterlain, Departamento de Antropologia, Universidade
de Coimbra, 3000-056 Coimbra, Portugal

Telephone: 351 239 829051

Fax: 351 239 823491

Email: sofiawas@antrop.uc.pt

Abstract

This paper details differential diagnosis of unusual tooth morphology observed in two adult males from the identified osteological collections held at the Museum of Anthropology of the Coimbra University (Portugal). The differential diagnosis is based on the morphological and radiographic analysis of the teeth of these individuals and gave rise to several possible pathological conditions, namely, regurgitation erosion, mottled enamel due to fluorosis, dentinogenesis imperfecta, amelogenesis imperfecta and osteogenesis imperfecta. Of these, amelogenesis imperfecta remains the most probable cause for one individual and although dentinogenesis imperfecta was considered for the other based on the morphological features, the final diagnosis was changed to the former based on accurate radiological evidence. Since there are only a few reported cases of both these conditions in the osteoarchaeological literature, we find it to be interesting and important to report these two cases. Besides, the scarcity of reports in the paleopathology literature, compared to the prevalence of these tooth structure anomalies in living populations, lead us to consider amelogenesis imperfecta and dentinogenesis imperfecta to be probably misdiagnosed, and hence under reported.

Key words: Amelogenesis imperfecta; dentinogenesis imperfecta; hereditary opalescent dentin; oral pathology; identified osteological collections.

Introduction

Two skulls with unusual tooth morphology were found in the identified osteological collections (19th and early 20th century) held at the Museum of Anthropology of University of Coimbra, Portugal. Tooth anomalies included marked hypoplastic changes in the crowns, grey-white discoloration of the enamel, destruction of tooth tissue at the amelodentinal junctions and chipping of enamel. The excellent state of preservation of these skulls, as well as the availability of detailed information about the individuals, allowed us to make differential diagnosis, which we report in this paper.

Case Report

The individuals reported in this paper are number 36 of the “International Exchange Skull Collection” (*Colecção das Trocas Internacionais*) and number 214 of the “Identified Skeletal Collection” (*Colecção dos Esqueletos Identificados*), curated by the Anthropological Museum from the Faculty of Sciences and Technology at the University of Coimbra. The establishment of Identified Skeletal Collection’s (ISC) was probably initiated in 1915 (Rocha 1995), by E. Tamagnini (1880-1972), who was the director of the Anthropology Museum. The 505 individuals in this collection died between 1904 and 1938 and the skeletons were exhumed from the main Coimbra cemetery (Conchada Cemetery). All except nine individuals were of Portuguese nationality. The foreigners were from Africa (6), Spain (2) and Brazil (1). E. Tamagnini was also responsible for the International Exchange Skull Collection (IESC), which was established with the aim of exchanging the osteological material with other countries. It

consists of 1075 complete skulls collected between 1932 and 1942, all of which originate from Conchada cemetery in Coimbra. The majority of the individuals was born in Portugal (N=1062), and the others were from Africa (1), Spain (6), France (1), Brazil (4) and Italy (1). The 524 males and 561 females died between 1904 and 1938 (Rocha, 1995; Cunha and Wasterlain, in press). Information about each individual, namely birthplace, sex and age at death, year and place of death, illness or cause of death, and occupation, amongst others, is compiled in a Record Book.

Case 1 (Skull 36 of the International Exchange Skull Collection)

Identification

This skull belonged to a 26-year-old Portuguese male, who lived in Coimbra during the first quarter of the twentieth century.

State of preservation

All the upper teeth were present; both lower first molars were lost ante-mortem and the lower left second molar was lost post-mortem. There was post-mortem fracture of the upper central incisors and lower right incisors.

Pathology

Periodontal condition: fair to good, without signs of gross calculus, crestal bone loss or tooth mobility. There was no gross root exposure.

Caries: lower right second molar showed gross crown destruction.

Description of the condition

Tooth crowns had a grey-white colour, reduced lucency, chipping and pitting of the enamel. The pits in the enamel have allowed stains to accumulate, causing discoloration. Mottling of enamel in relation to the crowns of the pre-molar and molar teeth (Figures 1, 2 & 3) was noticed. The occlusal enamel shelf was found to be fracturing off. On the amelocemental junction on labial surfaces of the anterior teeth, a deep groove completely separated the enamel and the cementum (Figures 4 & 5). At these areas, the margins of the enamel seemed to be chipping off (appeared to be due to weak adhesion to the underlying dentine) and this was also seen on the palatal surface of the right upper molar teeth. Although the lesions in the upper central incisors had the appearance of interproximal caries from the palatal view (Figure 2), on the labial view (Figure 4) it soon becomes apparent that these interproximal lesions are continuous with the labial lesions in the cervical region. Furthermore, the lesions abruptly stopped at the amelocemental junction without extending apically to involve the cementum on the root surfaces. Carious lesions at the cervical region would have involved both the crown and root surfaces.

Radiological features

The radiograph (Figure 6) reveals that radiodensity of enamel layer of teeth to be reduced to the extent it to be similar to that of dentin. No abnormalities in the roots or pulp chambers were apparent.

Case 2 (Skull 214 from Identified Skeletal Collection)

Identification

This skull belonged to a 38-year-old Portuguese male, who lived in the periphery of Coimbra during the first quarter of the twentieth century.

State of preservation

All the teeth were present except the right lower first premolar, which was lost post-mortem.

Pathology

Periodontal condition: horizontal bone loss was even around the dental arcade, which constituted “plane” form of alveolar bone loss. There was root exposure up to the bifurcations in both upper and lower molars. Deposits of calculus, especially on the buccal surfaces of the posterior teeth, were present. There was interproximal crestal cortical bone loss, exposing the underlying porous cancellous bone. All of these symptoms suggested periodontal disease (Clarke & Hirsch, 1991; Hillson, 1996).

Linear enamel hypoplasia [evidence of a disruption in enamel growth (Hillson & Bond, 1997)] was apparent on upper and lower canines (Figure 7), upper incisors, lower left incisors, and lower second premolars, suggesting that this individual experienced nutritional deficiency, infection or fever during childhood.

Occlusion: there is crowding in the upper and lower anterior teeth (Figures 8 & 9). Malocclusion Class 2, division 2; cross bite in the posterior region, with both upper and lower teeth lingually tilted (Figure 10). The incisor overjet was high (6mm). Despite the very unusual arch-form of the dentition, suggestive of congenital malocclusion and lateral cross-bites, no other skeletal abnormalities were apparent. The

collection files stated the cause of death as general paralysis, which is vague and subjective, no further clinical details about this individual were available.

Description of the condition

There were hypoplastic changes in the crowns of the upper and lower posterior teeth, similar to those described for the earlier skull. The colour of the enamel was distinctly grey and it was envisaged that during life these teeth would have been grey and translucent. There was no marked destruction at the amelocemental junctions in the upper or lower teeth. The buccal cortical alveolar bone in relation to the upper posterior teeth was extremely porous indicating bone remodelling. In the upper incisors, the palatal enamel was chipped off although there were no signs of traumatic occlusion with the lower teeth indicating that there was weak adhesion between the enamel and the underlying dentine. The crowns of both upper canines were almost totally worn down. We are unable to explain this phenomenon. There was pitting with brown discoloration on the palatal surfaces of the molars, and on the upper first and second right molars there were deep grooves at the amelocemental junction (Figure 8).

Radiological features

The initial orthopantomograph (OPG) of the dental arches (Figure 11) revealed that the roots of all the molars were comparatively very short, and this being more pronounced on the lower left molars. Furthermore, on these teeth, the pulp chambers and the root canals were not visible (obliterated). Root apices appeared to be open in several teeth. The crowns of the lower molar teeth were bulbous. But this OPG was found to be distorted and a subsequent accurate OPG (Figure 12) did not reveal any of the above features apart from absence of apical pathologies, and the enamel being less

opaque in relation to normal teeth and the distinction between enamel and dentine being relatively vague.

Differential diagnosis

Several conditions were considered when making the differential diagnosis of these two cases, namely regurgitation erosion, mottled enamel due to fluorosis, dentinogenesis imperfecta, amelogenesis imperfecta and osteogenesis imperfecta.

Mottled enamel due to fluorosis

These two males lived at the same time in the region of Coimbra, located in the central region of Portugal. Fluoridation was never introduced in Portugal, despite some experiments in few places later in the twentieth century, and current natural soil fluorine levels in the region of Coimbra are very low (between 0.03 mg F/l and 0.19 mg F/l) (Pinto *et al.*, 1999; Santos and Coimbra, 2004). Consequently, people living in the centre of Portugal would not have ingested fluorides in any significant amounts, until more recent times, when fluoridated toothpastes were introduced. Also, the examination of thousands of teeth from the three osteological collections held at the Museum of Anthropology did not reveal any fluoride related enamel hypoplasia. Therefore, mottled enamel due to fluorosis was eliminated.

Regurgitation erosion

As described earlier, the lesions on the crowns did not appear to be erosive in nature and also the areas affected did not correspond to the features typical of regurgitation erosion (Robb *et al.*, 1991).

Disturbances of development and growth

From the disturbances of development and growth affecting teeth, the most likely diagnosis is amelogenesis imperfecta, hypomaturation type, for Case 1 and dentinogenesis imperfecta, type II, for Case 2.

Amelogenesis imperfecta (AI)

This encompasses a group of conditions that demonstrate developmental alterations in the structure of the enamel in the absence of systemic disorder. The most widely accepted classification system for the subtypes of this condition is that developed by Witkop (1998). The hereditary defects of the formation of enamel are divided into hypoplastic, hypocalcified, and hypomaturation. The estimated frequency of AI in the population varies between 1:8000 and 1:700 (Neville *et al.*, 1995).

In hypoplastic AI there is inadequate deposition of enamel, pinpoint to pinhead sized pits are scattered across the surface of the teeth, and the buccal surfaces are more severely affected. The enamel between the pits is of normal thickness, hardness, and coloration.

In hypocalcified AI, the enamel matrix is laid down appropriately but no significant mineralization occurs and it is easily lost. It is often stained brown to black

and with years of function, much of the coronal enamel is removed, except for the cervical portion.

In a person with hypomaturation AI, the enamel matrix is laid down appropriately and begins to mineralize, but there is a defect in the maturation of the enamel crystal structure. Affected teeth are normal in shape but exhibit a mottled, opaque white-brown-yellow discoloration. The enamel is softer than normal and tends to chip from the underlying dentin. Radiographically, the affected enamel exhibits a radiodensity that is similar to dentin (Neville *et al.*, 1995).

Case 1 exhibited many of the morphological and radiographic features that of hypomaturation AI. In figures 1, 2 & 3 dentition exhibit mottled, opaque white enamel with scattered areas of brown discoloration, and areas where the enamel has chipped-off from the underlying dentin. Figure 4 shows defects in the labial cervical regions of upper anterior teeth where the softer enamel has undergone abrasion exposing the underlying dentin. Here it could be postulated that in life this individual has attempted to remove the discoloration by a vigorous oral hygiene method and accelerated the abrasive process. Radiograph (Figure 6) shows that radiodensity of enamel is reduced and appears similar to dentin.

Dentinogenesis imperfecta (DI)

This is a group of hereditary developmental conditions that affect the structure and composition of dentine. Both syndromic and non-syndromic forms of DI are known (Shields *et al.*, 1973). While their clinical presentation is highly variable, all types of DI share an opalescent blue-gray or yellow-brown discoloration of the tooth enamel. Although normal in structure, the enamel tends to fracture, especially on the incisal and

occlusal surfaces of the teeth, presumably because of an abnormal dentinoenamel junction (Shafer *et al.*, 1983). This usually leads to rapid attrition of the crown height (Gage *et al.*, 1991).

Shields and his co-workers (1973) have proposed three types of DI, based on clinical findings:

Type I occurs with osteogenesis imperfecta (a heritable connective tissue disorder characterized by skeletal fragility, growth deficiency, and a variable number of non-skeletal features), although the latter may occur without dentinogenesis imperfecta. It results from a variety of mutations in the type I collagen gene (Pallos *et al.*, 2001).

Type II is not associated with osteogenesis imperfecta unless by chance. This type is the one most frequently referred to as *hereditary opalescent dentin*. It is inherited as an autosomal dominant trait and is in fact one of the most common dominantly inherited disorders in human beings, affecting approximately one in every 8,000 persons.

Type III, reported in a southern Maryland inbred population known as the Brandywine isolate, is characterized by the same clinical appearance of the teeth as types I or II but with large pulp chambers and multiple pulp exposures in deciduous teeth. Type III is an autosomal dominant trait (Shields *et al.*, 1973).

The distinctive radiographic appearance of DI teeth is critical in establishing the correct diagnosis. The crowns are bulbous, with a constricted area at the cement-enamel junction and the roots appear shortened (Pettiette *et al.*, 1998). The enamel is not always evident in radiographs (Roberts and Schour, 1939). The most striking feature of types I and II teeth is the obliteration of the pulp chamber and root canals as a result of

overproduction of dentine. The cementum and supporting bone appear normal (Shafer *et al.*, 1983).

Dentinogenesis imperfecta type I was excluded based on the absence of any signs of osteogenesis imperfecta in this individual. Most of the morphological features of dentinogenesis imperfecta type II were apparent in case 2. For example, fracturing of enamel on the incisal and occlusal surfaces of the teeth, and attrition of crown heights (Figures 8 & 9). The only difference in the teeth of this skull, the colour of the enamel appeared distinctively grey rather than blue-grey or yellow-brown as described in the literature. It should be noted however that the description given in the literature is that of living individuals whereas our two cases consist of exhumed dry bones. The dehydration and the effects of geo-chemical changes of the skeletal material (e.g. diagenesis) may be the explanation to the above mentioned discrepancy in colour. The initial distorted radiograph (Figure 11) appeared to have some of the radiological features such as bulbous crowns, shortened roots, and obliteration of pulp chambers and root canals. But the subsequent accurate OPG (Figure 12) did not reveal any of these features apart from enamel being less opaque compared to normal teeth and the distinction between enamel and dentine being relatively vague and the radiodensity of enamel reduced to almost that of dentine. Therefore, the final diagnosis was similar to case 1 that is amelogenesis imperfecta (hypomaturation type). We included the details regarding the erroneous OPG to alert to the dangers of radiographic distortions leading to misdiagnoses.

Conclusions

We are quite sensitive to the fact that differential diagnosis in osteological materials is limited due to the absence of clinical diagnostic attributes (Ortner, 2003). Even so, we feel comfortable in diagnosing these two cases as amelogenesis imperfecta (hypomaturation type). Given the relatively high prevalence of this condition in living populations and the scarcity of reports in the paleopathology literature, we consider that these conditions have probably been missed or misdiagnosed and therefore under reported in previous archaeological studies.

Acknowledgements

The authors would like to thank the Museum of Anthropology of the University of Coimbra for permission to study the identified osteological collections. The authors are also grateful to *Departamento de Medicina Dentária da Faculdade de Medicina da Universidade de Coimbra*, to *Serviço de Radiologia dos Hospitais da Universidade de Coimbra*, and to Dr Cristina Pedroso, Dr J. Azenha Cardoso and Dr Joaquim Reis of *Clínica de Estomatologia e Medicina Dentária* for producing the radiographs of the skulls. The authors also acknowledge the editor of the journal and the anonymous reviewers whose valuable comments and suggestions allowed us to improve the manuscript.

References

Clarke NG and Hirsch RS. 1991. Physiological, pulpal, and periodontal factors influencing alveolar bone. In *Advances in Dental Anthropology*, Kelley MA, Larsen CS (eds). Wiley-Liss: New York; 241-266.

Cunha E and Wasterlain SN. The Coimbra identified osteological collections. *Documenta Archaeobiologiae* (In press).

Gage JP, Symons AL, Romaniuk K, Daley TJ. 1991. Hereditary opalescent dentine: variation in expression. *Journal of Dentistry for Children* **58**: 134-139.

Hillson S. 1996. *Dental Anthropology*. Cambridge University Press: Cambridge.

Hillson S and Bond S. 1997. Relationship of Enamel Hypoplasia to the Pattern of Tooth Crown Growth: A Discussion. *American Journal of Physical Anthropology* **104**: 89-103.

Neville BW, Damm DD, Allen CM, Bouquot JE. 1995. *Oral & Maxillofacial Pathology*. W.B. Saunders Company: Philadelphia; 79-84.

Ortner DJ. 2003. Background data in paleopathology. In *Identification of pathological conditions in human skeletal remains*, Ortner DJ (ed.). Academic Press: San Diego; 37-44.

Pallos D, Hart PS, Cortelli JR, Vian S, Wright JT, Korkko J, Bruononi D, Hart TC. 2001. Novel *COL1A1* mutation (G599C) associated with mild osteogenesis imperfecta and dentinogenesis imperfecta. *Archives of Oral Biology* **46**: 459-470.

Pettiette MT, Wright JT, Trope M. 1998. Dentinogenesis imperfecta: endodontic implications: case report. *Oral Surgery Oral Medicine Oral Pathology Oral Radiology and Endodontics* **86**: 733-737.

Pinto R, Cristovão E, Vinhas T, Castro MF. 1999. Teor de fluoretos nas águas de abastecimento da rede Pública, nas sedes de concelho de Portugal Continental. *Revista Portuguesa de Estomatologia, Medicina Dentária e Cirurgia Maxilofacial* **40**: 125-142.

Robb ND, Cruwys E, Smith BGN. 1991. Regurgitation erosion as a possible cause of tooth wear in ancient British populations. *Archives of Oral Biology* **36**: 595-602.

Roberts E and Schour I. 1939. Hereditary opalescent dentine (dentinogenesis imperfecta). *American Journal of Orthodontics and Oral Surgery* **25**: 267-276.

Rocha MA. 1995. Les collections ostéologiques humaines identifiées du Musée Anthropologique de l'Université de Coimbra. *Antropologia Portuguesa* **13**: 7-38.

Santos SA and Coimbra F. 2004. Resumo periódico. Divulgação dos resultados de acordo com o Decreto Lei n.º 236/98. Laboratório Controlo Qualidade. Águas de Coimbra.

Shafer WG, Hine MK, Levy BM. 1983. *A textbook of oral pathology*. Philadelphia: W. B. Saunders Company.

Shields ED, Bixler D, El-Kafrawy AM. 1973. A proposed classification for heritable human dentine defects with a description of a new entity. *Archives of Oral Biology* **18**: 543-553.

Wasterlain SN. 2006. '*Males da boca*': *Estudo da patologia oral numa amostra das Coleções Osteológicas Identificadas do Museu Antropológico da Universidade de Coimbra (finais do séc. XIX/inícios do séc. XX)*. PhD dissertation. Coimbra: Universidade de Coimbra.

Witkop CJ Jr. 1998. Amelogenesis imperfecta, dentinogenesis imperfecta and dentin dysplasia revisited: Problems in classification. *Journal of Oral Pathology & Medicine* **17**: 547-553. DOI: 10.1111/j.1600-0714.1988.tb01332.x