

MAXILLOFACIAL RADIOLOGY

Ectodermal dysplasia

SADJ September 2023, Vol. 78 No.8 p420-421

L Merbold¹, Chané Smit²

CASE

A 9-year-old female patient presented to the clinic with a main complaint of abnormally shaped and missing teeth. Upon clinical examination, the patient had a repaired cleft lip and palate and sparse hair of her eyebrows. Intraoral and radiological examination revealed generalised enamel hypoplasia, multiple conical-shaped teeth and multiple congenitally absent teeth.



Figure A

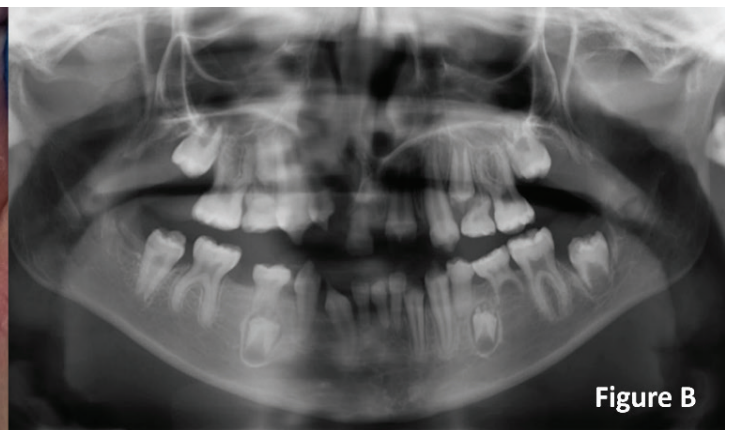


Figure B

Figure A: Clinical image showing the enamel hypoplasia (white arrow). Figure B: Panoramic radiograph showing congenitally missing teeth, enamel hypoplasia and conically shaped teeth.

INTERPRETATION

Ectodermal dysplasia (ED) characterises a group of disorders where two or more structures derived from the ectoderm exhibit developmental disturbances.¹ These include teeth, skin, hair, nails and sweat gland defects.² In some cases, ED may cause problems with hearing and vision and might also be associated with cleft lip and/or palate.² ED is a rare, diffuse, non-progressive, hereditary disorder³ with more than 150 subtypes.¹ There are different forms of ED – the first form occurs as X-linked recessive and the second form as autosomal dominant. In the X-linked recessive form males more commonly develop ED, while females only present as carriers of the abnormal gene. The autosomal dominant form is clinically present if either of the parents has ED, thus there is a 50% chance of ED transmission to their child.⁴

The head and neck manifestations of ED include the following:¹⁻³

- Enamel hypoplasia
- Malformed teeth (teeth that are smaller and pointed)
- Anodontia or hypodontia
- Malocclusion or ectopically-positioned teeth
- Cleft lip and/or palate
- Reduction in salivary flow
- Thin or sparse hair
- Reduced density of eyebrow/eyelash hair
- Nail dystrophy
- Abnormal functioning of the sweat glands
- Frontal bossing
- Saddle nose
- Periocular skin wrinkling and pigmentation

Diagnosing ED requires a multidisciplinary approach. ED is usually diagnosed during early childhood but, due to the wide range of clinical symptoms, some individuals are only diagnosed in adulthood.⁴ Many children with ED are diagnosed after a dental examination when the primary teeth have a delayed eruption pattern or when the teeth in the oral cavity are atypical in shape.⁵ Agenesis of teeth is a common finding – this includes anodontia (complete absence of permanent teeth), oligodontia (if six or more teeth are absent) and hypodontia (if less than six teeth are absent). The most common absent teeth are the maxillary lateral incisors and first premolars, followed by the mandibular central incisors and first premolars.⁵ Dental agenesis can affect jaw growth of the alveolar bone and favours a tendency for skeletal class III development.⁶ A thorough clinical and radiological

Authors' information

1. Lené Merbold, BChD, PGDipDent (Radiology), MSc (Maxillofacial Radiology), Department of Oral and Maxillofacial Pathology, School of Dentistry, Faculty of Health Sciences, University of Pretoria, South Africa.
ORCID: 0000-0003-4041-127X
2. Chané Smit, BChD, MSc (Maxillofacial Radiology), Department of Oral and Maxillofacial Pathology, School of Dentistry, Faculty of Health Sciences, University of Pretoria, South Africa.
ORCID: 0000-0003-4047-6356

Corresponding author

Name: Lené Merbold
Email: Lmerbold@gmail.com

Author's contribution

1. Lené Merbold (primary author) 70%
2. Chané Smit (secondary author) 30%

examination should be performed and, if ED is suspected, the patient should be referred for genetic testing to confirm the diagnosis.²

Treatment often depends on the case's complexity and the patient's age, with some treatments only commencing after growth is completed.² Treatment may include preventive oral care, including good oral hygiene with regular dental visits. Functional and aesthetic rehabilitation should be performed via a multidisciplinary team approach. The goal of early treatment is to resolve the problem of multiple missing teeth and to enhance jaw growth to achieve better function.⁷ The initial dental treatment should mainly focus on prevention of caries and restoring tooth alterations to improve the patient's occlusion.⁷ Rehabilitation of the occlusion includes partial or complete removable dentures, orthodontic expansion devices, dental implants and/or crowns.¹ Together with this approach, a psychologist and speech therapist should aid the patient to improve social acceptance.⁸ After growth is completed, the specialities of maxillofacial surgery together with orthodontics and prosthodontics will be required.⁵ This case report highlights the importance of dentists identifying the clinical and radiological features of ED as they may be the first line of diagnosis.

AUTHORS' DECLARATION

Funding

This research did not receive any specific grant from funding agencies in the public, commercial or not-for-profit sectors.

Conflict of interest

The authors declare that they have no conflict of interest.

Ethics approval

This study was approved by the University of Pretoria Ethics Committee (Reference no 360/2023). All procedures followed the ethical standards of the Helsinki Declaration of 1975, as revised in 2008.

REFERENCES

1. Neville BW, Damm DD, Allen CM, Chi AC. (2016) Oral & Maxillofacial Pathology. 4th Edition, WB Saunders, Elsevier, Missouri, 741-743
2. Visinoni Átila F, Lisboa-Costa T, Pagnan NAB, Chautard-Freire-Maia EA. (2009). Ectodermal dysplasias: clinical and molecular review. American Journal of Medical Genetics Part A, 149a(9), 1980-2002. <https://doi.org/10.1002/ajmg.a.32864>
3. Halai T, Stevens C. (2015). Ectodermal dysplasia: a clinical overview for the dental practitioner. Dental Update, 42(8), 779-80
4. Kariya R, Kadota T, Okawa R, Ochiai M, Okuda M, Nakano K. (2023). Two cases of ectodermal dysplasia diagnosed based on congenital absence of multiple teeth. Pediatric Dental Journal, 33(2), 147-151. <https://doi.org/10.1016/j.pcdj.2023.03.001>
5. Cerezo-Cayuelas M, Pérez-Silva, A, Serna-Muñoz C, Vicente A, Martínez-Beneyto Y, Cabello-Malagón I, Ortiz-Ruiz AJ. (2022). Orthodontic and dentofacial orthopedic treatments in patients with ectodermal dysplasia: a systematic review. Orphanet Journal of Rare Diseases, 17(1). <https://doi.org/10.1186/s13023-022-02533-0>
6. Bondarets N, Jones RM, McDonald F. (2002). Analysis of facial growth in subjects with syndromic ectodermal dysplasia: a longitudinal analysis. Orthodontics & Craniofacial Research, 5(2), 71-84. <https://doi.org/10.1034/j.1600-0544.2002.01159.x>
7. Wimalaratna AAK, Weerasekara WBMCRD, Herath EMUCK. (2020). Comprehensive management of ectodermal dysplasia with interceptive orthodontics in a young boy who was bullied at school. Case Reports in Dentistry, 2020, 6691235-6691235. <https://doi.org/10.1155/2020/6691235>
8. Celli D, Manente A, Grippaudo C, Cordaro M. (2018). Interceptive treatment in ectodermal dysplasia using an innovative orthodontic/prosthetic modular appliance. A case report with 10-year follow-up. European journal of paediatric dentistry, 19(4), 307-312. <https://doi.org/10.23804/ejpd.2018.19.04.11>

Online CPD in 6 Easy Steps 

The Continuing Professional Development (CPD) section provides for twenty general questions and five ethics questions. The section provides members with a valuable source of CPD points whilst also achieving the objective of CPD, to assure continuing education. The importance of continuing professional development should not be underestimated, it is a career-long obligation for practicing professionals.

