Ectodermal dysplasia—an unusual dental presentation

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Ectodermal dysplasia (ED) is a rare group of disorders affecting the hair, teeth, nails and sweat glands to a variable degree. There is a wide range of clinical presentation of ED. Missing teeth or abnormal tooth form may be the first indicator of the presence of the disorder. There is typically hypodontia with microdontia. We present an unusual case of ED with severe hypodontia and macrodontia affecting all first permanent molar teeth. We also consider the classification and presentation of this disorder.
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INTRODUCTION

Ectodermal dysplasias encompass a large and diverse group of diseases affecting tissues of ectodermal origin, e.g. hair, nails, teeth, and sweat glands. ED was first described by Thurnam in 1848.[1] Now there are over 170 different, clinically distinct subtypes.[1-4] EDs are rare. The incidence has been estimated at 7 in 10,000 births.[5] ED follows all possible modes of Mendelian inheritance, i.e. autosomal dominant or recessive or X- linked dominant or recessive and there have also been sporadic cases reported.[3]

Pathogenesis is thought to involve an altered epithelial- mesenchymal interaction.[2,3] About 30 causative genes have been isolated. These are involved in intracellular communication and signaling, cell development, cell survival and differentiation among other things.[5]

CLASSIFICATION

Several classifications exist and can be confusing. Some are based on clinical features[6,7] and others on genetic components of the disorder.[2] Clinically, there are two broad groups. Group A includes those cases in which there is a defect in at least 2 of the 'classic' structures, i.e. hair, nails, teeth, and sweat glands with or without other defects. Group B encompasses those cases in which only 1 of the 'classic' structures is affected, but there co- exists at least another ectodermal defect.[4] All cases can further be subdivided into pure ED- in which there are only ectodermal signs- and ED syndrome- in which there are ectodermal features as well as other malformations.[4] The most common variant is X-linked hypohidrotic dysplasia, which consists of hypotrichosis, hypohidrosis and hypodontia.[8,9]

CLINICAL FEATURES

Craniofacial clinical features can include an old- looking face, frontal bossing, prominent supraorbital ridge and antimongoloid palpebral slant. The ears may be prominent and pointed, the nose short and wide, with a dished-in face and reduced facial height. Additional features are thin, protruded upper lip and everted lower lip, small and pointed chin and deep labio-mental fold. Dental features include natal teeth and missing primary and permanent teeth. Tooth form may be microdont, taurodont and conical. Hypoplastic enamel and caries have also been reported. Hair is sparse or absent. Sweating is decreased or absent and skin is dry and hyperkeratotic.[10]

CASE REPORT

A 6 year old boy was referred to the dental department at Great Ormond Street Hospital for Children by the dermatology department. Dentally, he complained of a retained primary incisor tooth and occasional pain from the permanent molar teeth. The patient was born at 35 weeks with 6 natal teeth. These were removed soon after birth.

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Medical history included total alopecia, which began at 18 months. The hair on the scalp and eyebrows grew back sparsely over the next few years (Figure 1). The patient also suffered with dry skin and eczema. He had a slightly conical shaped head, and pointed prominent ears. He had a prominent labio-mental fold. The patient exhibited no abnormality of nails or sweat glands. There was no family history of related features or missing teeth.

The patient was seen by a clinical geneticist who made a provisional diagnosis of ectodermal dysplasia, based on the abnormalities of hair and teeth. He was doing well at school and not getting teased.

On dental examination the following teeth were fully erupted

R 16 55 53 11 | 61 63 65 26 L
46 83 | 72 73 36

Radiographic examination revealed severe hypodontia (Figure 2). The only other tooth that was present, apart from those erupted, was 2.1 (Figure 3). This was impacted in the line of the arch and appeared abnormal in size and form. Tooth 11 was notched and deeply invaginated palatally. The enamel on all four primary canines was pitted and hypoplastic. The first permanent molar teeth were considerably larger than average and were concave occlusally (Figure 4). Caries was evident in the following teeth

R 16 55 11 | 65 26 L
46 | 36

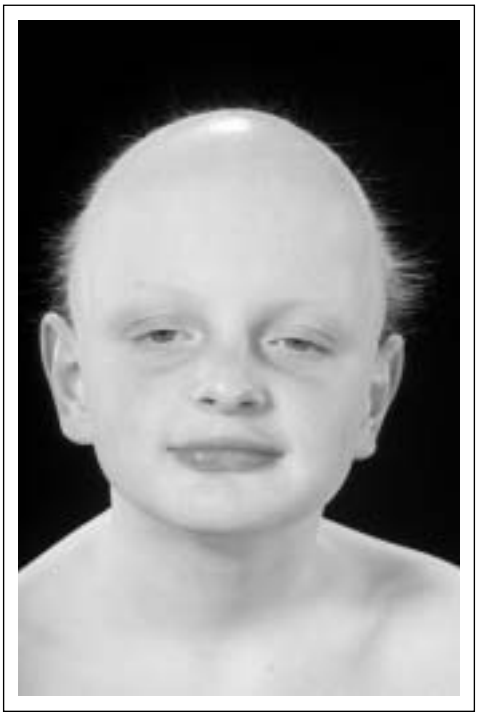

Figure 1. Clinical appearance typical of ectodermal dysplasia

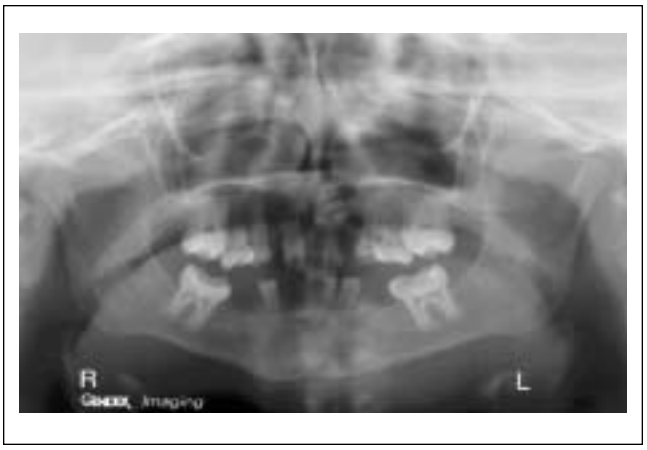

Figure 2. DPT showing severe hypodontia

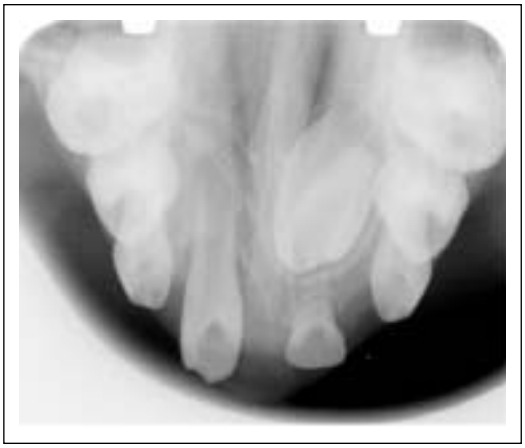

Figure 3. Upper anterior occlusal showing 2.1 impacted and abnormal in form

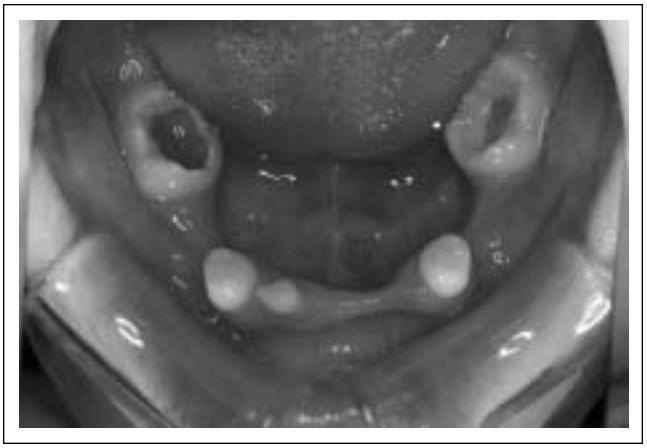

Figure 4. Photograph demonstrating macrodont form and concave occlusal surface of first permanent molars

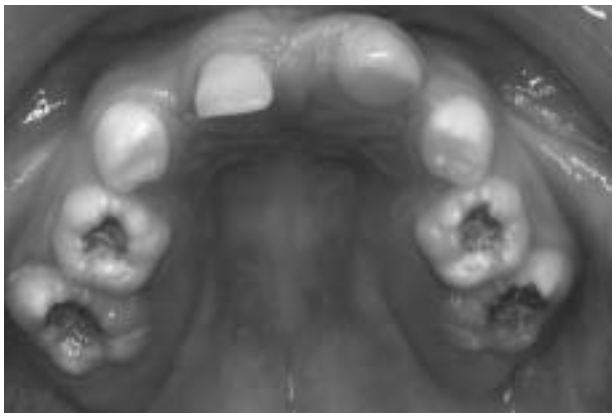

Figure 5. Post-operative view of restored upper arch

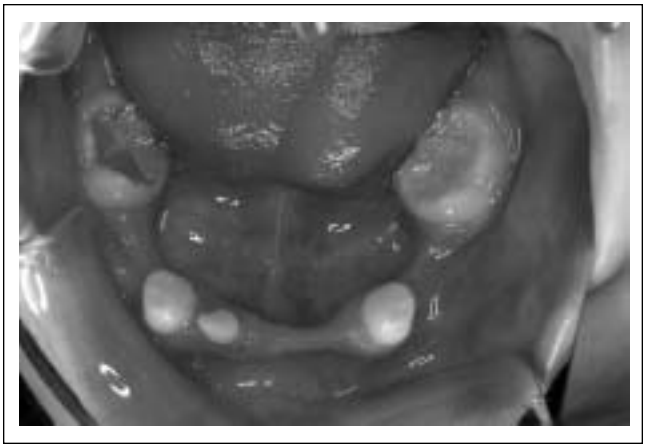

Figure 6. Post-operative view of restored lower arch

TREATMENT

Treatment included oral hygiene instruction and diet advice. An orthodontic opinion was obtained regarding the unerupted incisor and a decision made to surgically explore the area and decide what to do with the tooth at time of surgery. Following discussion with the patient and parents, treatment as a day case under general anaesthesia was planned. At operation, an orthodontic opinion was again sought, and it was decided to leave the 21 *in situ* at this time, due to its proximity to the 61 and the risk of losing the permanent incisor. Also it would not have been possible to pull the 21 into the line of the arch adequately and the abnormal morphology would preclude it on grounds of aesthetics. The 61 was built up with composite, the invagination in 11 was restored with composite, all permanent first molar teeth were restored with either amalgam or a posterior composite (Figures 5, 6). The primary molar teeth were restored with compomer or amalgam.

The patient and parents were pleased with the result in appearance anteriorly. This temporary solution will serve to delay provision of a removable prosthesis until later. The abnormal incisor will require surgical removal in the future prior to definitive prosthetic replacement. His restored dentition will serve functionally and aesthetically for the time being. However, he is likely to request aesthetic improvement prior to commencing secondary school.

Dental treatment for these patients is a particular challenge. Treatment needs are life long, and should aim to improve function and aesthetics.[11] A multidisciplinary team approach is often needed, including Pediatric Dentist, Orthodontist, Prosthodontist and Oral Surgeon. Options to be considered are fixed or removable prosthesis, cosmetic modification of existing teeth and osseointergrated implants.[12]

The widespread caries in this case highlights the importance of preventative advice, as the few teeth that are present are precious.

CONCLUSION

Ectodermal dysplasia is a rare group of genetic disorders affecting tissues of ectodermal origin. Typically hair, nails, teeth and sweat glands are affected. Dentally, the most common findings are hypodontia and microdontia. We present an unusual case of ED with large, 'saucerised' teeth and severe hypodontia. Clinicians should be alert to the fact that ED has many, varied presentations, and should be considered as a differential diagnosis in any patient with anomalies of tooth form, structure or number. Although the restorative management may be challenging, the importance of basic prevention should not be underestimated.

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