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Case Report

Two interesting cases of EEC syndrome

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ARTICLE INFO

Article history:

Received 8 November 2012

Accepted 6 February 2013

Keywords:

EEC syndrome

Ectrodactyly

Cleft lip

Ectodermal dysplasia

ABSTRACT

Aim: To report two cases of EEC syndrome with oral involvement to highlight the fact that the dental surgeon may be among the first to recognise the condition.**Introduction:** The EEC syndrome, a rare congenital syndrome is characterised by ectodermal dysplasia, distal limb anomaly, cleft lip and palate and lachrymal duct anomalies although the term oligosymptomatic EEC syndrome has been suggested for cases in which ectrodactyly may be absent and that such cases should not be deprived of the EEC syndrome diagnosis. It is usually inherited as an autosomal dominant trait with variable expressivity and penetrance and all these features rarely coexist in a single individual.**Conclusion:** Both our cases had all three characteristic features of the EEC syndrome. Other syndromes with overlapping features such as the Rapp–Hodgkin syndrome and the AEC syndrome should be considered in the differential diagnosis of incomplete forms of EEC syndrome.**Clinical significance:** It is important to follow an interdisciplinary approach to attain the greatest degree of success possible in the treatment of such cases.

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1. Review of literature

The term EEC syndrome, coined by Rudiger et al,¹ stands for ectrodactyly, ectodermal dysplasia and cleft lip/palate. Although it is a multiple congenital anomaly syndrome characterised by the above and lachrymal duct anomalies,² Kuster & Majewski reported 8 members in two families with EEC syndrome with no limb defects.³ Majewski & Kuster later coined the term 'Oligosymptomatic EEC Syndrome' and suggested that ectrodactyly was not mandatory for syndrome diagnosis.⁴ Penchaszadeh & deNegrotti had, by

this time, indicated that the lack of a major component should not preclude diagnosis and later Wallis reported a four generation family with EEC syndrome without oro-facial clefting. Although not part of the characterisation, renal anomalies, choanal atresia, growth retardation, and EEG abnormalities have been reported in patients with EEC syndrome.

Although phenotypically similar syndromes such as the Rapp–Hodgkin Ectodermal Dysplasia, The Ankyloblepharon Ectodermal Dysplasia and Clefting Syndrome exist, they are believed to be separate clinical entities.² Also, the overlap

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<http://dx.doi.org/10.1016/j.jobcr.2013.02.001>

between these and the EEC syndrome make syndrome identification difficult.

2. Inheritance

Although autosomal dominant inheritance is confirmed, in familial cases, sporadic and recessive traits have been reported. It is very likely that the EEC syndrome is another example of genetic heterogeneity and determination of its

genetic mechanism or mechanisms will need clinical variability to be carefully documented and defined.

3. Case description

3.1. Case 1

A six-year-old male, presented with complaints of broken teeth and inability to eat. A product of a non-consanguineous

Fig. 1 – Images of Case 1 (A: Profile, B: Intraoral, C: Feet, D: Hands, E: Orthopantomograph).

marriage, born full term, with a normal delivery conducted at home, the parents noticed a cleft lip and missing digits and toes at birth. He was operated for cleft lip and palate at the age of 9 months. His developmental milestones and scholastic performance was adequate. The parents denied similar features in any other family members. General examination revealed ectrodactyly of the upper and lower limbs, with a typical lobster claw hand and split feet deformity (Fig. 1C & D). The skin was dry, coarse with reduced sweating and the scalp had thin sparse hair. Facial features included a prominent forehead with a receding hair line, post surgical scars on the lip, prominent nostrils and maxillary hypoplasia (Fig. 1A).

Intraorally, few teeth were present which were difficult to classify (Fig. 1B). All the present deciduous teeth showed severe attrition upto the gingival level with pulp exposures. The cuspal tips of a permanent mandibular posterior tooth were also seen.

The Orthopantomograph showed large pulp chambers and wide root canals. The permanent tooth buds of the premolars, second molars, were visible but molar identification was not possible (Fig. 1E).

Based on the clinical and radiological findings, EEC syndrome was diagnosed. The patient was lost to follow up.

3.2. Case 2

A 13-year-old male presented with a cleft on the upper lip (Fig. 2B). The patient gave a history of early shedding of the deciduous teeth, reduced sweating and heat intolerance. The intra uterine, neonatal and family history of the patient was not contributory. The patient denied consulting any other doctor for his complaints in the past. General examination revealed dry skin with normal hair. Examination of the limbs revealed ectrodactyly of both hands with a typical lobster claw hand and split feet deformity of both feet. There was a deep cleft of the hands with the central ray being deficient causing the absence of 2nd and middle phalanges. The feet also showed a similar deep cleft with missing 2nd and 3rd toes. There was also syndactyly of the patient's left 4th and 5th toes (Fig. 2C).

The upper lip was thick with an incomplete left sided hare lip. The palate was normal. Intraoral examination revealed a

Fig. 2 – Images of Case 2 (A: Intraoral, B: Cleft lip, C: Feet).

malposed left maxillary central incisor and a missing left maxillary lateral incisor (Fig. 2A). The complement of teeth was normal with respect to shape, size and colour. He was diagnosed as a case of EEC syndrome and referred for reconstructive surgical repair.

4. Discussion

The acronym, EEC, was first proposed by Rudiger et al, characterised by ectrodactyly, ectodermal dysplasia, cleft lip and/or cleft palate.¹ Since then, numerous reports in the literature have broadened its clinical spectrum. The transmission appears mostly by autosomal dominant inheritance, with variable expressivity and reduced penetrance.^{5,6} Some authors believe that the classical cases of EEC syndrome are mostly due to p63 gene mutations.⁷ The variability in the phenotypic expression is explained by the anomalies of ectodermal/mesodermal interactions.² The skin, nails, teeth, salivary & lacrimal glands and limbs have similar embryonic development, pointing to the inclusion of variations in EEC syndrome.

The involvement of skin includes hypopigmentation, dryness and hypohidrosis. Hair may be dry, coarse, sparse and hypopigmented. Nails may show dystrophy with transverse ridging and pitting. The ophthalmologic findings reported are blepharitis, conjunctivitis, corneal scarring and spontaneous perforation, reduced Meibomian & lacrimal gland secretion and dacryocystitis secondary to abnormalities of lacrimal gland apparatus.^{2,5,8} Limb anomalies include ectrodactyly (84%), as in our cases or syndactyly, clinodactyly, with the deficiency of central ray being most common.

Cleft palate with or without the involvement of lip appears to be present in nearly 68% of cases.⁹ Dental findings include partial or total hypodontia, malformed conical teeth, enamel hypoplasia, severe caries. Chranowska et al, have reported a case with anodontia as the sole feature of EEC.¹⁰

According to Buss et al, a diagnosis of EEC can be made based on ectodermal dysplasia with one of the following features – ectrodactyly, cleft lip or palate and lacrimal duct abnormalities.² Both our cases had ectodermal dysplasia, ectrodactyly and cleft lip, thereby fitting into the diagnosis. However, other syndromes with overlapping features, such as the Rapp–Hodgkin syndrome and the Ankyloblepharon, Ectodermal dysplasia, Cleft lip/Palate (AEC) syndrome should be considered in the differential diagnosis of incomplete forms of EEC syndrome. Dental management should include preservation of the primary dentition with the use of fluoridated mouthwashes & salivary substitutes, restoration of carious teeth and replacement of missing teeth. Treatment of EEC syndrome involves an interdisciplinary approach.

5. Clinical significance

Whenever cases of EEC syndrome are encountered, it is important to follow an interdisciplinary approach, depending on its phenotypic manifestations, to reduce complications, minimize untoward sequelae and provide the best possible medical care.

Conflicts of interest

All authors have none to declare.

Acknowledgments

The authors gratefully acknowledge the contribution made by Dr. Siddhartha Gupta towards this paper.

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