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EEC SYNDROME, CLINICAL FINDINGS - CASE REPORT

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ABSTRACT

Introduction: Ectrodactyly syndrome, ectodermal dysplasia and cleft lip and/or palate (CEE), is a rare genetic anomaly of autosomal dominant inheritance and estimated incidence of 1.5/100 million inhabitants. The triad that names it are also its main characteristics and gravity can be expressed in different degrees of severity. The objective of this study was to report the case of a child diagnosed with EEC syndrome treated at CEAPAC (Center for Care and Research in Craniofacial Anomalies), narrating the clinical findings, with emphasis on the dental aspects found. Case report: A 10-year-old female child was born well –

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according to the grandmother's reports. Oral and limb malformations were diagnosed in the prenatal period through ultrasound. The following physical characteristics were observed: corrected complete transforamen right lip cleft, with the presence of a fistula in the anterior region of the palate, hypertelorism, dacryocystitis, prognathism, slightly rotated ears posteriorly, dry skin and hair, ectrodactyly, and bilateral syndactyly of feet and hands. In the dental evaluation, the presence of mixed dentition, dental agenesis, dental shape anomaly, class III skeletal pattern, malocclusion, caries disease and endodontic involvement of a tooth is noted. Results: Despite the malformations in the limbs, the patient had good motor coordination and was instructed about oral hygiene. Then, the treatments continued. The patient is still being monitored. Conclusions: It is necessary that patients with CSE be followed up early and periodically by a multidisciplinary team due to the malformations resulting from the syndrome.

Keywords: Congenital Abnormalities, Cleft Palate, Cleft Lip, Rare Diseases.

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INTRODUCTION

Ectrodactyly syndrome (E), ectodermal dysplasia (E), cleft lip and/or palate (C), usually identified by the acronym EEC is a rare genetic congenital anomaly, of autosomal dominant inheritance due to heterozygosis in the TP63 gene located on chromosome 3, with about 30% inherited and 70% due to de novo mutation (SUTTON & BOKHOVEN, 2021). Some older studies estimate the incidence to be 1-9/100 million individuals in the population (SHARMA et al., 2015), however, despite considering it a rare condition, Sutton & Bokhoven (2021) state that the prevalence of 63 TP63 disorders individually or collectively is unknown, despite the fact that more than 300 cases have been described in the literature (GOMES, BIELLA, NEVES, 2003). The triad that names it makes up its main characteristics, however, the disorder has variable expressiveness (BHARATI et al., 2020; PACHAJOA & HERNÁNDEZ-AMARIS, 2014) and incomplete penetrance (ORPHANET, 2011).

The following clinical manifestations may be present: anatomical or functional changes in the sweat, sebaceous, salivary and mammary glands, skin changes (hypopigmentation, dryness, hyperkeratosis, atrophy), changes in hair (thin and sparse hair and eyebrows), nail dystrophy, ectrodactyly, syndactyly, clinodactyly, facial hypoplasia, otological changes, short stature, genitourinary malformations (renal agenesis, urethral atresia, hydronephrosis), involvement of the nervous system, choanal atresia, ophthalmologic defects (tear duct defect, photophobia, corneal ulcers, keratitis, blepharitis, entropion), and endocrine anomalies (thymus hypoplasia, hypopituitarism, growth hormone deficit). Rarely, spongy white nevus, delayed psychomotor development, and malignant lymphoma may occur. Dental findings include: Complete or incomplete cleft lip and palate and dental changes (small and/or absent teeth, dysplastic and hypoplastic) (FERNANDEZ et al., 2010; PASCUAL et al., 2003; SUTTON & BOKHOVEN, 2021).

The diagnosis can be given in the prenatal period from ultrasound, or postnatally, in which case it is based on clinical examination, radiography of limbs and mandible, and if necessary, other tests (ultrasound, ophthalmological examination and genetic test) (GOMES et al., 2003; ORPHANET, 2011).

The patient affected by the syndrome must be accompanied by a multidisciplinary team that provides holistic care. Surgery may be necessary to correct orofacial anomalies and improve the function and appearance of the limbs (GONZÁLEZ-BALLANO et al., 2014; PACHAJOA & HERNÁNDEZ-AMARIS, 2014).

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That said, the objective of this study was to report the case of a patient diagnosed with EEC syndrome, attended at CEAPAC (Center for Care and Research in Craniofacial Anomalies), narrating the clinical findings with emphasis on the dental aspects found.

METHODOLOGY

The present research is a descriptive study, in which data and images were collected during consultations with the team. Prior to the beginning of this study, the child's legal guardian (maternal grandmother) was asked to sign the informed consent form (ICF), who consented to the dissemination of the images and data for scientific purposes. The study was approved by the research ethics committee (opinion 7.107.057 and CAAE 36452320.0.0000.0107), following Resolution No. 510/2016 of the National Health Council.

RESULTS

A 10-year-old female child with EEC Syndrome attended CEAPAC accompanied by her maternal grandmother (her legal guardian), from another specialized center.

According to the grandmother's reports, the patient was born well, despite SGA (small for gestational age) and low weight in the fetal period (about 300 grams at the sixth month of intrauterine life). The type of delivery was cesarean section and the malformations were diagnosed in the prenatal period through ultrasound examination.

The patient has the following physical characteristics: corrected complete transforamen right lip cleft (Figure 1 A), but with the presence of a fistula in the anterior region of the palate (Figure 1 C), hypertelorism, dacryocystitis (Figure 1 D), prognathism (Figure 1 A and B), slightly rotated ears posteriorly, dry skin and hair (Figure 1 A and B), ectrodactyly, and bilateral syndactyly of the feet and hands (Figure 2 A and B). After the speech-language pathology evaluation, it was found that the voice was hypernasalized due to the fistula in the anterior palate region. The grandmother narrates that the surgeries to correct the lip, palate, hands and feet were performed at the specialized center where the patient was initially treated.

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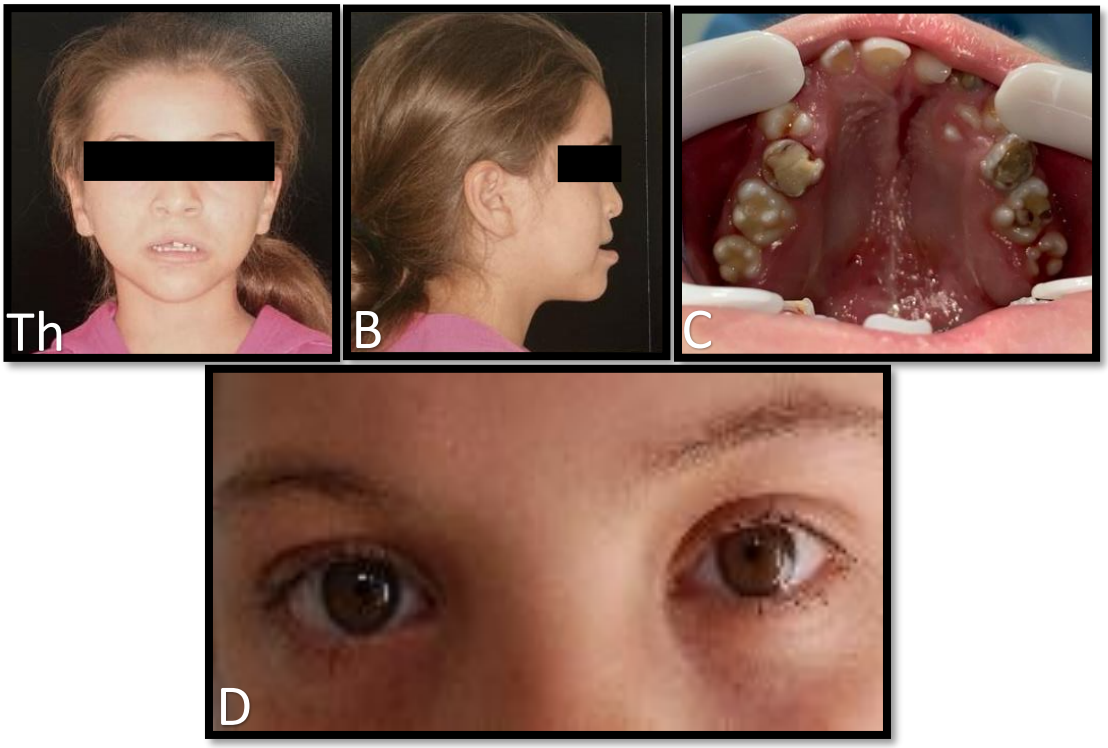
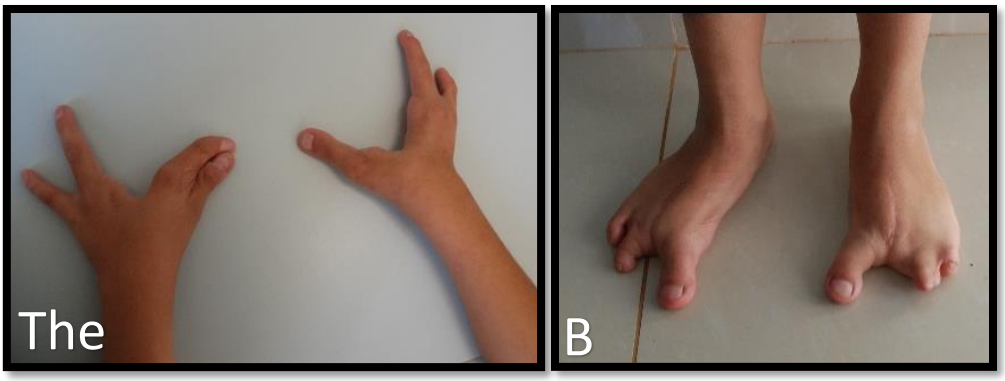
Figure 1 - In A and B - the following can be observed: Prognathism, ears slightly rotated posteriorly, skin and hair dry. In C: Fistula in the anterior region of the palate. In D: Hypertelorism, eyes with secretion.



Source: The author

Figure 2 – In A: Ectrodactyly and syndactyly of the hands. In B: Ectrodactyly and syndactyly of the feet.



Source: The author

In the dental evaluation, the presence of mixed dentition can be noted, with the following teeth affected by caries: 36, 53, 54 and 64 (Figure 3 A and 3 B). Shape anomalies are also observed in teeth 16, 17, 26 and 27 (Figure 3 A and 3 C). The patient has a skeletal pattern consistent with class III and dental malocclusion (Figure 3 A, 3 C and 3 E). All dental elements have been restored (Figure 3 D and 3 E) and orthodontic treatment will be initiated.

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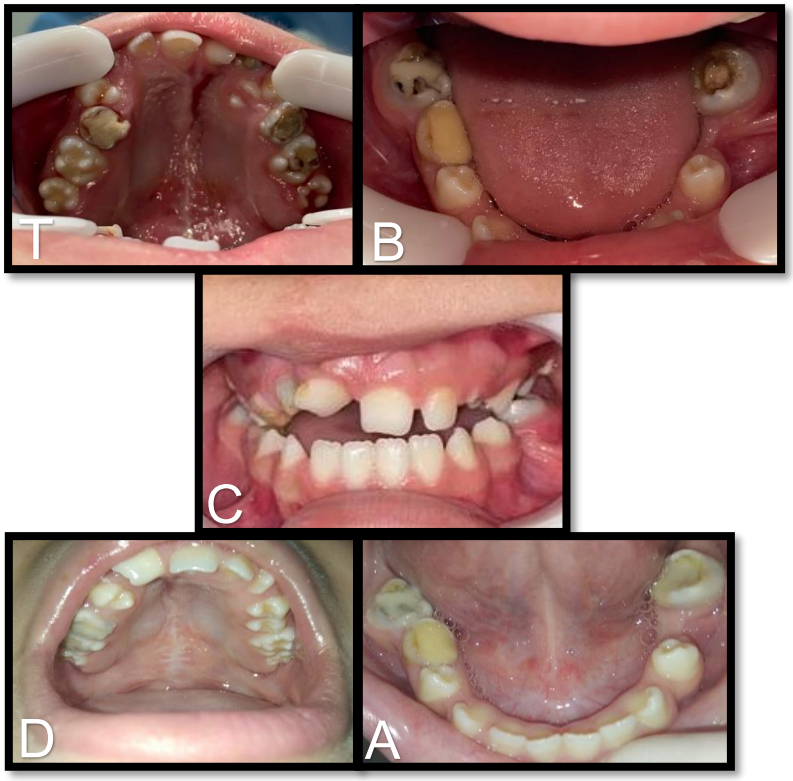
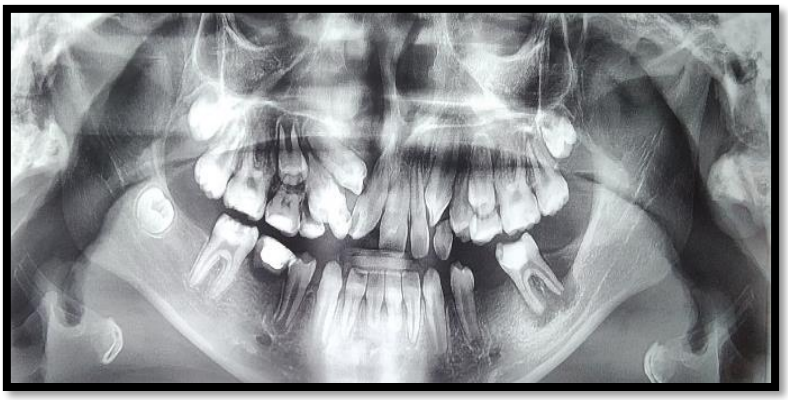
Figure 3: In A: Presence of caries lesion, shape anomaly and malocclusion. In B: Presence of caries. In C: Shape anomaly and malocclusion. In D: Upper teeth restored. In E: Restored lower teeth.



Source: The author

From the panoramic radiograph, it was possible to observe agenesis of elements 12, 35, 37, 38, 45 and 47 and periapical lesion in the first permanent molar on the left side, evidencing the need for endodontic treatment (Figure 4 A).

Figure 4 – In A: Panoramic radiograph, in which agenesis and periapical lesion can be observed.



Source: The author

The endodontic treatment of the tooth was started, however the patient was very agitated, requiring a lot of conversation and prior conditioning. The filling of the tooth in

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question has not yet been completed, due to the presence of suppuration, and it was decided to change intracanal medication until this condition ceases.

The patient is being monitored by teams of speech therapists, doctors, dentists, physiotherapists, geneticists and social workers according to their planning. Periodic returns will be scheduled in order to monitor the patient's oral health.

DISCUSSION

The clinical findings found in the reported patient led to the clinical diagnosis of EEC Syndrome. Similar characteristics were reported in the works of Fernández et al. (2010); Guzmán-Cerda et al., (2017); Rosa et al. (2017); Sutton & Bokhoven (2021).

Some authors have shown in their studies that patients with EEC syndrome should be monitored in a multidisciplinary manner, starting treatment with surgical corrections, in order to improve the functionality of the systems involved in the malformation (BRAVO et al., 2016; GUPTA et al., 2017). The patient in this report is cared for by a multidisciplinary team and underwent surgical correction of the cleft lip and palate, as well as surgery to improve the appearance of the feet and hands, prior to entering this center.

Bharati et al. (2020) report that individuals with EEC syndrome are more predisposed to diseases such as caries and gingivitis, possibly due to the difficulty of performing proper hygiene, due to ectrodactyly of the upper limbs. The minor reported has good motor coordination in the hands, but active caries were found in some teeth. Thus, the work of the dental team aims to reestablish the patient's oral health, through restorations, endodontic and orthodontic treatment. It was not possible to prevent oral diseases and maintain the health of the oral cavity, as indicated in the studies by Bravo et al. (2016) and Gupta et al. (2017), due to the age at which the child arrived at the center. However, the patient was advised to improve her toothbrushing technique, and motivated to maintain daily oral hygiene habits, as well as healthy eating to avoid future caries.

CONCLUSION

In view of the malformations resulting from EEC syndrome, it is necessary for the patient to be monitored by a multidisciplinary team capable of providing him with a better quality of life. As for dental treatments after cheiloplasty and palatoplasty surgeries, they should be started as early as possible, so that, unlike the case reported, in which the patient developed numerous cavities, the use of preventive methods and the restoration of

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oral health are efficient. In view of the scarcity of studies on the subject, since it is a rare syndrome, it is hoped that this report will contribute to the professionals of multidisciplinary teams and dentists knowing the management of patients with EEC.

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