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DENTAL MANAGEMENT IN PATIENTS WITH SYNDROMIC CRANIOSYNOSTOSIS; CASE REPORT

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ABSTRACT

Introduction: *Apert syndrome is a challenging syndromic condition for health care specialists because it affects the patient's overall physical, oral and behavioral health. Knowledge of the protocols to be followed and dental management by the operator of patients with AS are of great importance in the dental field, allowing for adequate and guided craniofacial and behavioral control of the patient.*

Objective: *The objective of this article is to present a clinical case that shows an adequate treatment plan for a patient with Apert syndrome.*

Methodology: *an objective description of the clinical case is made with analysis of a total of articles, including review and original articles, as well as clinical cases, of which 12 bibliographies were used because the other articles were not relevant for this study. The sources of information were indexed journals, as well as search engines such as PubMed, Google Scholar and Cochrane; the terms used for the information search were: Apert syndrome, acrocephalosyndactyly, craniosynostosis, dental care.*

Results: *We report a 15-year-old female patient who presented the characteristic triad of Apert syndrome: craniosynostosis, facial malformations and symmetrical syndactyly.*

Conclusions: *Knowledge of the disease by health professionals will be of vital importance in order to develop appropriate strategies and efficient care protocols for patients with Apert syndrome, resulting in a better quality of life for the patient in the short and long term.*

KEY WORDS: *acrocephalosyndactyly, craniosynostosis, dental care.*

INTRODUCTION

Patients with some type of syndromic condition are more likely to have a high morbidity and mortality rate, so the care needs and knowledge of proper management in the medical or dental office are of vital importance. For the initial clinical evaluation of patients with special needs, three important steps are required:

a. General health evaluation: in this section, the cooperation of the person in charge of their care is important, in order to know about any extra anomalies that may be of interest, since the following should be made known: medications used, diseases and eating habits(1).

b. Evaluation of oral health: the patient's current oral condition will be assessed and previous treatments will be analyzed. This includes taking X-rays, which will be useful for the diagnosis of the jaws and teeth(2).

c. Evaluation of the behavior: it is considered for the management of syndromic patients, so the behavior is analyzed by means of some scales, among them the most used is the modified Frank scale to predict the cooperation and motor skills.

After completing this first phase of clinical evaluation of the patient, preventive treatment should be started: prophylaxis, fluoride application and sealant placement. Conservative therapy includes restorative treatment in both deciduous and permanent dentition. In case there are habits or some type of malocclusion, it is when you should intervene with orthopedic treatment and orofacial exercises. In the craniofacial surgery center the analysis, diagnosis, treatment and rehabilitation of the patient are established depending on the pathology that is present, whether congenital, hereditary or genetic origin. Surgical therapy includes dental extractions, gingivectomies, biopsies, oral or maxillofacial surgery(1,2).

Apert syndrome (AS), also known as acrocephalosyndactyly, is one of the rarest and most severe craniosynostosis syndromes, accounting for approximately 4.5% of all cases of

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craniosynostosis. It is a rare congenital autosomal dominant disorder characterized by severe craniosynostosis (premature closure/fusion of multiple cranial sutures, specifically the coronal suture) and is associated with craniofacial anomalies, including symmetrical syndactyly of the 2nd to 4th fingers and toes (partial or complete); fusion of the skin and bones of the fingers/toes, with a common fingernail(2,3).

In severe cases, synostosis of the radius/humerus and shoulder and elbow joints may occur, with ocular (superficial orbits, exophthalmia, strabismus, hypertelorism and downward slanting palpebral fissures), ear (chronic otitis media, hearing loss), (obstructive sleep apnea, mouth breathing), skin (acne, excessive sweating), brain (ventriculomegaly, hydrocephalus) and malformations of the corpus callosum and/or limbic structures; in addition, some children may have mild mental/intellectual deficits, with an average intelligence quotient (IQ) of 74, pharyngeal (short stature) and internal organ (gastrointestinal, cardiovascular, genitourinary) abnormalities(3,4).

Craniosynostosis leads to a restriction of the anteroposterior growth of the facial skeleton, from the glabella to the posterior fontanelle, resulting in the characteristic conical head of AS in its variable acrobrachycephaly or turribrachycephaly. Craniofacial findings include midfacial/maxillary hypoplasia with class III malocclusion, premature fusion of the fifth and sixth cervical vertebrae, flat forehead and occiput, depressed broad nose with bulbous tip, and deviated septum(5).

The prevalence of the syndrome has been estimated to be between 1 / 65,000 and 1 / 200,000 newborns, with no gender predilection. AS has an autosomal dominant pattern of inheritance, associated with advanced paternal age, maternal infections, maternal drug use and cranial inflammatory processes. More than 98% of cases are caused by specific mutations in the fibroblast growth factor R (FGFR2) gene on chromosome 10q25-10q26, which are exclusively paternal in nature. FGFR is a family of mitogenic signaling molecules that play an important role in the control of cell proliferation and survival; thus, in AS, fibroblasts are unable to produce the essential fibrous material in various craniofacial tissues, including bone sutures and cartilage, and during tooth formation and regeneration; therefore, the mutated FGFR2 gene may influence the dental abnormalities observed in AS (2,6,7). Molecular genetic mapping of specific FGFR gene mutations or prenatal ultrasonographic detection of structural abnormalities is recommended to confirm the diagnosis(2,7).

Therefore, the present study aims to expose and identify the treatment protocol aimed at children with AS in order to establish in an orderly manner the procedures to be followed to achieve the proposed results.

PRESENTATION OF THE CASE

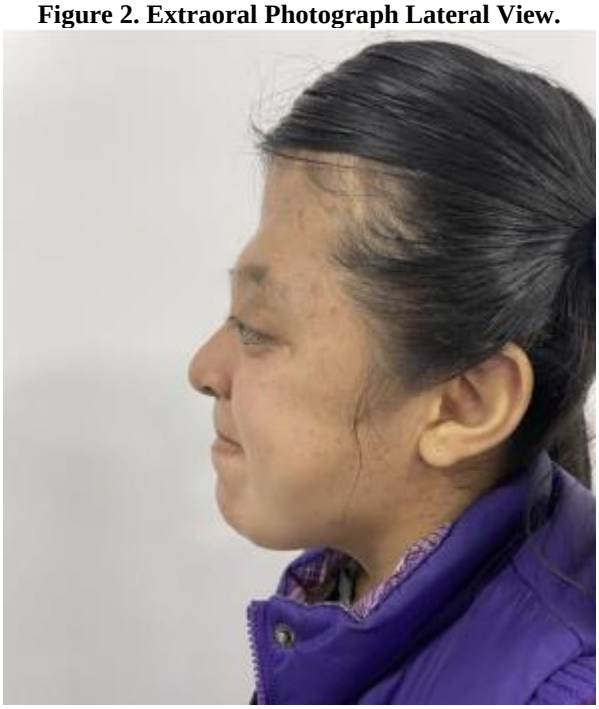
A 14-year-old female patient attended the Cleft Lip and Palate Clinic of the Catholic University of Cuenca. Physical examination shows that the patient presents craniosynostosis, macrocephaly and facial asymmetry. In the upper facial third we can see scars at frontal level caused by previous surgeries, low ocular implantation, hypertelorism, hypoplasia of the upper third, turri-brachycephaly, low capillary insertion. In the middle third of the face there is evidence of short nasal pyramid, low auricular implantation and middle third hypoplasia. In the lower third there is a transverse and anteroposterior deficiency of maxillomandibular growth (Figure 1 and 2). (Figure 1 and 2) At the level of the upper and lower limbs there are mitten-shaped hands and feet.

Figure 1. Extraoral Photograph Frontal View.



Source: The Authors.

Figure 2. Extraoral Photograph Lateral View.



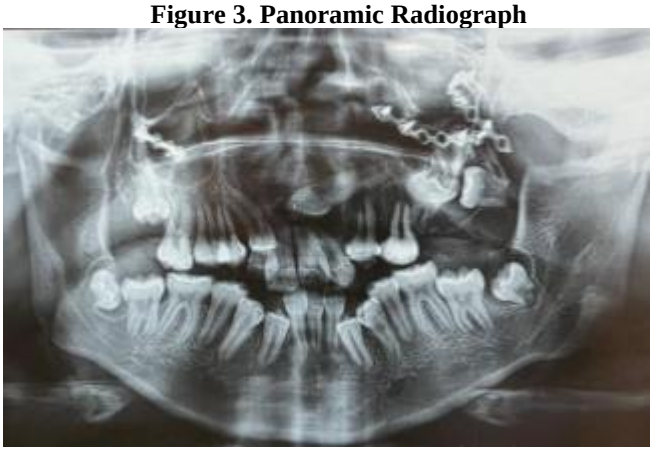
Source: The Authors.

On intraoral examination we can observe that the patient presents ogival palate with presence of cleft palate, velopharyngeal insufficiency, Angle class III, macroglossia, dental defects such as enamel hypoplasia, dental crowding, caries, generalized gingival hyperplasia, absence of dental pieces. In the radiograph multiple dental inclusions of the dental

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organs can be observed (17, 23, 26, 27, 38 and 48) (Figure 3, 4 and 5).

Figure 3. Panoramic Radiograph



Source: The Authors

Figure 4. Lateral Cephalic Radiograph



Source: The Authors

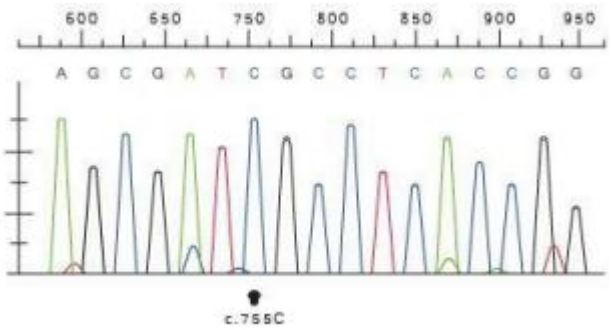
Figure 5. This image shows dental crowding, poor dental position and occlusion, dental caries, gingival hyperplasia with a change in the color of the gingiva, enamel hypoplasia type 2 classified by the International Dental Federation, where the tooth can be seen with white, opaque, yellow and brown tones.



Source: The Authors.

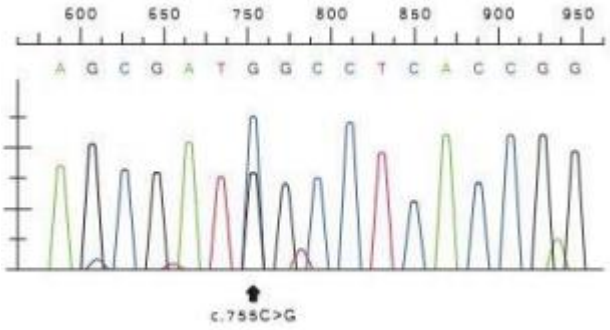
To certify the diagnosis, genetic tests were performed to prove Apert syndrome (Figure 6 and 7).

Figure 6. Electropherogram with normal FGFR2 gene sequence, the presence of homozygous cytosine at position 755 of c.DNA was observed.



Source: The Authors.

Figure 7. Electropherogram determines a heterozygous variant at position 755 of the c.DNA, a variant in the above mentioned Cytosine by a Guanine is observed. A change is detected at the level of the FGFR2 protein (p.Ser252Trp).

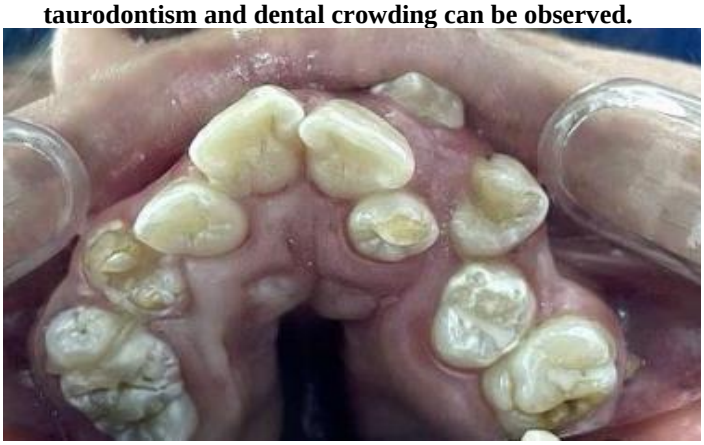


Source: The Authors.

In the management of this case we started with the pediatric dentistry area performing a prophylaxis, then we proceeded to seal completely with a luminoactivation resin sealant the affected pieces 36 and 46 classified 03 by the International Caries Detection and Diagnosis System (ICDAS) where the localized enamel rupture due to caries without visible dentin is located. In these cases the use of materials such as glass ionomer, which releases fluoride and is useful as a preventive and therapeutic approach, is also indicated. The preventive part of pediatric dentistry begins, which includes the placement of sealants in pits and fissures in teeth 35, 24 and 26. 5% fluoride varnish is placed for caries prevention and enamel renewal. The maintenance will be done every 3 months since the patient presents a high risk of dental caries. When the preventive phase is completed, the patient is referred to the periodontics area (Figure 8 and 9).

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Figure 8. Upper arch. Palatal fissure, teeth with taurodontism and dental crowding can be observed.



Source: The Authors.

Figure 9. Restored Lower Arch.



Source: the authors.

In the periodontics area, the first step was to motivate the patient to take care of his oral health on his own. Then a prophylaxis was performed to reduce the infectious focus of the teeth. The appropriate brushing technique was taught to the patient and periodic controls were performed to evaluate the improvement of the patient.

Figure 10. Plaque control with the use of discloser.



Source: The Authors.

Figure 11. Periodontal control after 1 month.



Source: The Authors.

PROPOSED SURGICAL TREATMENT

In order to achieve an adequate intermaxillary relationship and an acceptable occlusion, the following is proposed:
1.Maxillary and mandibular transverse distraction
2. Anteroposterior distraction of the middle third.
3. Surgical-orthodontic repositioning of included teeth .
4. Orthognathic and aesthetic facial surgeries.

DISCUSSION

Apert syndrome, being a congenital pathology with no cure and with a very low prevalence of 15.5 per 100,000 live births, becomes a challenge for professionals according to Torres and Lattanzi, since there is no standardized care protocol for these patients(8,9).

According to Shin, the main objective of care for patients with AS is to improve the quality of life and their optimal development(10).

According to Torre and Shin, the prenatal diagnosis of these pathologies can be determined mostly at the 19th week of gestation and thus important decisions can be made at the time of the child's birth, where the comprehensive treatment should begin with a team of professionals such as pediatricians, maxillofacial surgeons, neurologists, plastic surgeons, anesthesiologists, dentists, geneticists, otolaryngologists, who should work together to achieve a multidisciplinary team of care throughout the patient's life(8,10).

Wilkie reports that patients with AS can present upper and lower extremities of three types: type I (spade), type II (mitten) and type III (rosebud)(7).

The present case presented type II. Specialists should keep in mind that patients with syndromic craniosynostosis are more susceptible to oral diseases, so starting always with preventive treatments will be the best protocol of care for these patients. Similarly, multiple studies by Mathews and Calandrelli indicate the importance of evaluation and management of respiratory pathologies. Transverse deficient jaws will cause the patient to develop as a mouth breather, which is associated with a risk

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factor for developing gingival and periodontal diseases (10,11,12).

The patient in the case study presented a triad that is characteristic of AS where we can observe: craniosynostosis, facial malformations and symmetrical syndactyly. It was also determined that she is a mouth breather and has an atypical swallowing, so these pathologies at the beginning of her treatment were reflected in her oral cavity. In order to maintain adequate oral health, the patient was instructed in oral hygiene techniques and the use of a conventional toothbrush combined with an electric toothbrush was recommended to obtain better results. Likewise, depending on the case, fluoridation sessions could be performed. In the maintenance phase, the progress of the patient's periodontal health, how he/she has managed the techniques taught, and the results of the same will be analyzed.

CONCLUSION

The lack of knowledge or experience on the part of the operator when starting treatment of a syndromic patient can lead to late or incorrect treatment, so it is important that the operator follows a management protocol and thus provide the best quality of life for these patients. In the management of patients with Apert syndrome, a multidisciplinary treatment is necessary, where the medical and dental areas (pediatric dentistry, periodontics, orthodontics and surgery) should work together to detect the disease at an early age and provide the appropriate treatment plan in the short and long term, thus achieving a total and stable rehabilitation of the patient.

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Conflict of Interest Statement

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