



Orthopedic and orthodontic features in patients with Apert syndrome: review of literature

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Abstract

In the PubMed accessible literature, studies about maxillo-facial features of Apert Syndrome and about its interdisciplinary orthodontic and surgical treatment is rare. Apert's syndrome is a malformation characterized by craniosynostosis, midface hypoplasia, ocular manifestations (hypertelorism) and syndactyly of the hands and feet. These craniofacial characteristics predispose the patient to sagittal hypoplasia of the maxilla with concomitant anterior open bite (skeletal and dental) dental crowding, a pseudocleft palate. The best way to treat this pathology is using a combined approach both orthodontic and surgical. In our review of literature we want to investigate about most common orthopedic and orthodontic alterations of these patients, in order to understand which of these are most common and to evidence that the management of Apert syndrome patients include a multidisciplinary approach involving orthodontists, neurosurgeons, plastic surgeons, ophthalmologists.

Background

Apert's Syndrome, described for the first time by Eugene Apert, is a rare disease characterized by a dominant autosomal inheritance. [1]

It is characterized by craniosynostosis, bilateral syndactyly of hands and feet, and maxillo-facial alterations as by hypertelorism, proptosis, acrocephaly or brachicephaly, short and flattened nose with a decreased nasolabial angle. [2]

In the oral cavity it is possible to observe an ogival palate, maxillary transverse and sagittal hypoplasia, dental crowding, eruptive delay, ectopic position of the teeth. Even though it could seem evident a pseudoprognathism, Mandibula is usually of normal size. Abnormalities to the nervous, gastrointestinal and urogenital systems are rare as a compromission of cardiac and respiratory system, although upper respiratory tract infections, sleep apnea, and malnutrition can be seen. [3]

The mandibular prognathism is the result of the midface hypoplasia due to a retraction of the third

medium of the face, where as lips assume usually a trapezoid configuration. [4-5] The palate is arched with bilateral swelling of the palatine processes called "pseudocleft palate", although it could be present a true cleft palate. [6]

Anterior open bite, severe crowding and maxilla hypoplasia are the most frequent dentofacial alterations associated to Apert Syndrome. Therefore, skeletal class III is not the result of a prognathic mandibula, but is due to an altered sagittal development of the jaw. [6-7]

Dental anomalies include delayed and ectopic eruption, bladder incisors, crowding of teeth, especially in open bite, bilateral crossbite, mandibular prognathism and midline deviations [8].

Material and Methods

Therefore, the purpose of this review is to show to the orthodontists and to maxillo-facial surgeons the altered cephalometric and dentofacial parameters of this class of patients analyzing literature between 2000 to 2016. The systematic review of literature has been performed on the principal medical databases: PubMed (Medline).

Our aim was to investigate the literature up to now regarding the Apert syndrome and to analyze some parameters emerging from each single study, finally comparing them with each other.

Keywords used were : [apert syndrome], [orthodontic], [craniosynostosis], [midface hypoplasia].

Many articles published about Apert Syndrome are case reports, so we want to identify, for each of them, orthodontic alterations.

Review

In 2000, Ryneerson [6] described a case of a 21-year-old caucasian woman, in good health, but with typical skeletal alterations of Apert's syndrome: hypertelorism, prominent frontal skeleton and third-rate hypoplasia. It was an increase in the length of the face, ectopic teeth, anterior open bite, poor oral hygiene, severe discrepancies about the arch length. The maxilla was hypoplastic with a V shaped arched

palate. There was also a deviation of the median line.

In 2007, Hohoff [9] described three cases of patients with Apert syndrome. In all three patients the same features were highlighted: hypertelorism, excess of the lower third of the face, a trapezoid upper lip, delayed eruption with dental crowding and a pseudoprognathism, open bites with monolateral crossbite .

In 2010, Verdonck [10] described a 14-year-old woman with Apert syndrome, pointing to the presence of maxillary facial flattening of the nasal bridge, prominent frontal skeleton, maxillary hypoplasia with mid-third retraction of the face, pseudoprognathism accompanied by an anterior open bite and unilateral crossbite. The oral hygiene of the patient was good. The palate, on the other hand, showed the arched shape to V with two bilateral swellings of palatine processes.

Benmilaud [11], in 2013, studied a 10-year-old child with Apert syndrome. No parameters other than those previously reported by other authors have reported apart from the presence of mental retardation and cleft palate with bifid uvula.

Also in 2013, Bhatia [12] treated other clinical case of a 14-year-old boy with Apert syndrome who further confirmed the presence of the same altered parameters.

In 2014 Kumar [13] and in 2016 Koca [14], they still described two clinical cases of two patients with Apert syndrome and again the parameters found were the same.

An overview of maxillo-facial features in patients affected by Apert Syndrome were presented in Table 1.

(Table 1: Overview of orthopedic, orthodontic and systemic alterations in patients with Apert syndrome in the cases reported in the literature from 2000 to 2016.)

	2000 Rynearson	2007 Hohoff (1A case)	2007 Hohoff (2A case)	2007 Hohoff (3A case)	2010 Verdonck	2013 Benmilaud	2013 Bhatia	2014 Kumar	2016 Koca
Acrocephaly/Brachicephaly	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Flattened occipital bone	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Prominent frontal bone	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Proptosis and hypertelorism	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Cardiac or respiratory alterations	No	No	No	NoA	No	No	No	No	No
Flattened nose	No	No	No	NoA	Yes	Yes	Yes	Yes	Yes
Midface hypoplasia	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Increased length of the face	No	Yes	No	Yes	No	Yes	No	Yes	Yes
Pseudoprognathism/III class	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Dental crowding	Yes	Yes	Yes	NoA	No	Yes	Yes	Yes	Yes
Ectopic eruptions	Yes	No	No	NoA	No	Yes	No	Yes	Yes
Delayed eruptions	No	Yes	No	No	No	No	No	Yes	No
Anterior open bite	Yes	Yes	Yes	Yes	No	No	Yes	Yes	Yes
Monolateral crossbite	Yes	Yes	Yes	Yes	Yes	No	No	No	No
V-shaped arched palate/pseudocleft palate	Yes	No	No	No	Yes	Yes	Yes	Yes	No
Poor oral hygiene	Yes	No	No	No	No	No	No	Yes	No
Trapezoid upper lip	No	No	No	No	Yes	No	No	Yes	No
Syndactyly	No	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Mental retardations	No	No	NoA	No	No	Yes	No	No	No

Conclusions

Concluding, the maxillo-facial alterations that we can find most frequently in patients with Apert syndrome were: hypertelorism and proptosis, occiput flattening, projection of the frontal skeleton, flat nasal bridge. In the orthodontic area, the most obvious signs were the presence of anterior skeletal and dental open bite, midface hypoplasia, delayed dentition and dental crowding, monolateral cross bite, V-shaped arched palate (only one case in our review shows cleftpalate with bifid uvula). Heart and respiratory disturbances weren't compromised in the analysis of case reports of our review. Rare cases of mental retardation were present.

In the approach to this class of patients, the aim of the specialists was to provide the patient with a more acceptable face and occlusion outcome obtainable exclusively through a combined orthodontic / surgical approach.

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