



Maxillofacial features in patients with Noonan Syndrome

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Introduction

Noonan syndrome(NS) is a congenital disease characterized by low stature, chest deformity showing a pectus excavatum, cardiac abnormalities, in particular pulmonary stenosis, facial dysmorphism, hearing problems and dysmorphism of the ears , haematological alterations, cryptorchidism, brachydactyly, mental retardation (1).

In most cases the diagnosis of this disease is clinical, sometimes supported by genetic abnormalities. It is transmitted with dominant autosomal mode and the mutation associated with this pathology is a point mutation of the PTPN11 gene located on the long arm of chromosome 12, but as evidenced by other dominant pathologies, a percentage of cases occurs as a result of a new mutation. (2)

Incidence is between 1 out of 1,000 and 1 out of 2,500 born alive in absence of race, ethnicity and gender preference. (3)

Clinical diagnosis is not always possible in the infant, so it is carried out at 6-7 years of age. (4)

Many pathologies have phenotypic characteristics similar to those present in NS, such as Turner's syndrome with which it shares epicanthus, cardiovascular alterations and skeletal anomalies. However, the Noonan's syndrome may also be associated with other pathologies such as LEOPARD Syndrome, Costello's Syndrome, Cardiopagocytic Syndrome and Type 1 Neurofibromatosis. (5)

The quality of life of a patient suffering from NS is normal, although always subjected to specific attentions for the presence of several concomitant pathologies. The management of these patients therefore requires a multidisciplinary approach, with a close collaboration of different specialist. (6)

Characteristics of NS-affected patient's oro-facial district:

There are few studies in literature that explain the orthopedic-orthodontic management of NS patients in the odontostomatologic field; most of them are represented by case reports.

However, a literature systematical analysis reveals interesting data about the characteristics of oral district.

According to several studies the oro-facial region is characterized by triangular facial shape, posteriorly rotated ears, eyelid ptosis, flat nose root, high forehead. These aspects are unclear at birth, become more accentuated during childhood, and are attenuated during adulthood. (7-8)

In the oral cavity, there is a greater incidence of caries, even in areas not generally affected, because of a greater aptitude for the consumption of sugary foods and a lower propensity to oral hygiene due to psychomotor deficits. (9)

A smaller percentage of cases show a tendency to the formation of odontogenic keratocysts (10) and giant cell lesions. (11-12)

A series of dento-skeletal alterations emerge from the systematic analysis of case reports in the literature.

Methods

Our aim was to investigate literature about Noonan Syndrome and to analyze some parameters emerging from each single study, finally comparing them to each other.

The studies included were conducted between 1994 and 2015 with a total of 10 NS patients.

The keywords used in the PubMed search engine for literature analysis were: [noonan syndrome]; [orthognathic surgery]; [pediatric surgery].

Review

We have subsequently established orthodontic parameters used to compare the cases. Specifically, we have defined the skeletal class, divergence, the interincisal angle amplitude, the protrusion/retraction of the jaws, the presence of ogival palate.

Sugar, in 1994, illustrated a case report of a 22-year-old male patient, single son without NS family history. Clinically, his appearance was characteristic, he showed a triangular face and a flat nose root. Clinically there was an open front bite, a Class III malocclusion, mandibular prognathism and maxillary hypoplasia. Therefore, according to Sugar, this patient was not an ideal candidate for orthodontic treatment,

so he opted for a surgical approach that included Le Fort I osteotomy on the upper jaw, bisagittal osteotomy of the branch on the mandible and autologous bone graft taken from the iliac ridge to overcome the poor size of the forehead.(13)

Okada, in 2003, studied an 8-year-old Japanese boy with NS. Orthodontically there was an ogival palate and an open front bite. Cephalometric analysis had shown a tendency to hyperdivergence as the gonial angle was larger in size, with a dolicho-facial typology. (14)

Buccheri and Ceccano, in 2006, published a clinical case of two brothers affected by Noonan's Syndrome with skeletal open bite, hyperdivergence, ogival palate and monolateral cross bite. Even functional alterations such as night snoring and atypical swallowing were found in both cases. (15)

Asokan, in 2007, studied a 13-year-old girl with NS, highlighting the presence of skeletal open bite and a III skeletal class. (16)

Ortega, in 2008, intercepted two clinical cases of patients with NS, highlighting each of their facial and dental features. Both affected by Å II skeletal class, the patients described by Ortega also showed an increased gonial angle and for one of them an increase in the interincisal angle. Both patients showed dental crowding and one of them also showed the presence of monolateral cross bite. (17)

Emral, in 2009, described a 13-year-old male patient with NS. During clinical examination, although the cephalometric measures showed a tendency to hyperdivergence, it was found a 9 mm deep bite, together with dental Agensis and ogival palate (18).

Ierardo, in 2010, described an 8-year-old patient with low-grade NS and heart problems. Cephalometric analysis and facial typology were standard, there was a first-class skeletal relationship and a deep bite. Agenesies of elements 4.5 and 3.1 were present. (19)

Uloopi, in 2015, studied a 13-year-old boy affected by NS. Cephalometric analysis showed I skeletal class relationships with horizontal jaw growth patterns and lingualization of lower incisors. In the mandibular anterior region there were three supernumerary teeth. Orthopantomography showed agensis of upper incisors 1.2 and 2.2 and clinically it was possible to observe ogival palate and deep bite. (20)

Conclusions

The systematic review of literature from 1994 to 2015 for various cephalometric and clinical parameters

analyzed by different authors allows us to state that some of them appear continuously in patients affected by NS. In particular, patients with Noonan Syndrome have a greater tendency to hyperdivergence and to skeletal open bite, although it is frequent deep bite due to lingualization of lower incisors, the presence of permanent teeth agensis and supernumerary dental elements. There is no particular preference for skeletal class malocclusion, reflecting a development trend similar to the incidence observed in the normal population.

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