OROFACIAL FINDINGS IN NOONAN SYNDROME: A CASE REPORT

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Abstract

Noonan syndrome is an autosomal dominant multisystem disorder, associated with cardiac anomalies and a distinctive facial appearance, characterized by genetic heterogeneity. Noonan syndrome affects both females and males, and has an estimated incidence of 1 per 1,000-2,500 live births. The present report aims at presenting the cranio-dento-facial findings in a case of Noonan syndrome in a 6 year-old male.

Keywords: Noonan syndrome, dental and facial features, orthodontics.

INTRODUCTION

Noonan syndrome was first described by Noonan and Ehmke in 1963 as a multisystem disorder. They defined a specific group of nine patients with pulmonary stenosis, short stature, mild mental retardation, hypertelorism, low-set ears and unusual but similar faces [1]. Other general aspects reported in literature are: broad or webbed neck, a peculiar chest deformity – pectus carinatum superiorly and pectus excavatum inferiorty, congenital heart diseases, swallowing difficulties, lymphedema, mental retardation and poor coordination, learning disabilities, speech delays, joint or muscle pain and coagulation deficiencies [2-5]. Noonan syndrome was often correlated to Turner syndrome, because some common features of theirs, such as epicantus folds, right-sided congenital heart disease and various skeletal malformations [6]. In Noonan syndrome, no chromosomal anomaly has been found [5,6]. Tartaglia et al. [7] reported in 2001 the molecular basis of this syndrome: mutation in the PTPN11 (protein tyrosine phosphatase non-receptor type 11) gene, reported in about 50% of the individuals with Noonan syndrome.

The estimated incidence is of 1 per 1,000-2,500 live births [8]. Although the frequency of sporadic cases appeared to be high in early reports, more recent surveys show direct transmission from parent to child in 30%-75% of cases. Maternal transmission of the gene is far more common than paternal transmission (3:1) [6]. Many cases have been reported in the medical literature, but only a few of them describe some details of the cranio-dento-facial features: a high arched palate, dental malocclusion and articulation difficulties [9-13]. Teeth anomalies are also seen with late eruption, increased risk of dental decay and above average need for orthodontic treatment [14]. Therefore, the aim of the present article is to present the oro-facial findings in a case with Noonan syndrome in a 6 year-old male.

CASE REPORT

The 6-year-old male patient was referred to the Orthodontic Dental Clinics for orthodontic examination. A detailed medical and dental history was obtained from the patient and from his parents. The mother had the same syndrome, with a history of cardiac valve surgery.

Medical history

The patient had been diagnosed as having Noonan syndrome at 2 months. He presented cryptorchidism, pulmonary stenosis and persistent ductus arteriosus. At 2 years, he underwent an orthidexy surgical treatment, followed by surgery for a catheterization procedure for pulmonary stenosis. The patient is also suffering from hypothyroidism and he follows treatment
with thyroid and growth hormones. Physical examination revealed a relatively short stature (height:107 cm, weight: 17.5 kg), low neck-hair line and low set, posterior rotated ears and a short neck. He exhibits a very active behavior and the level of cooperation was satisfactory, appearing to be cognitively normal.

**Facial findings**

The patient presented a facial dimorphism with bilateral telecanthus, hypertelorism, ptosis and a tendency for exophtalmos. Facial shape was of an inverted triangle, with a relatively large nose and full lips. He had a tendency for lip incompetence and he reported the habit of mouth breathing. (Fig. 1)

![Figure 1. Facial aspects](image)
Intra-oral examination

Oral examination revealed the primary dentition with extensive and multiple profound caries lesions, most of them with atypical locations and defects in teeth mineralization and dystrophies. The lower primary central incisors were exfoliated and the primary upper incisors had baby bottle tooth decay. The patient presented a narrow high-arched palate with prominent rugae. The presence of an important skeletal anterior open bite can be clinically correlated with the thrust tongue habit with abnormal swallowing pattern, although no nasal obstruction was found. The postlacteal plane was in mesial step.

Radiographic examination

Hand-wrist radiography demonstrated a slightly retarded bone age (5 years and 6 months) according to the standards of Greulich and Pyle [15].
All permanent teeth, except for the third molars, were present on the orthopantomography. Slight signs of crowding can be observed in the upper canine-premolar area. The sequence of eruption seems to be inverted in the canine-premolar mandibular region (Fig. 4).

Treatment plan

A multidisciplinary approach, involving pedodontic, orthodontic and endocrinologic treatment, was planned. In the first stage, the active lesions have been treated by the pediatric dentist and then the orthodontic treatment was started. Patient’s growth and hormone level was closely monitored by the endocrinologist. The patient received a myofunctional appliance, in order to correct the tongue thrust habit and to control the anterior skeletal open bite during the period of upper and lower incisors eruption. The frequency of control visits is at 2 moths and the eruption needs to be supervised.

DISCUSSION

The present case report reinforces the statement that Noonan syndrome is variable in phenotype, due to its genetic heterogeneity. The patient had many common features, such as: cryptorchidism, pulmonary stenosis, short stature, low neck-hair line and low set, posteriorly rotated ears and a short neck. A relatively rare associated characteristic is thyroiditis, reported in literature just in few cases. The oro-facial features frequently reported and observed in this patient were: hypertelorism, ptosis, a relatively large nose, full lips, a high arch palate and anterior skeletal open bite [10-14]. He also exhibits less reported features - such as multiple caries [9, 13, 14, 16]. The patient received a myofunctional appliance, in addition to a proper oral hygiene and a caries reduction protocol. The parents were informed that, depending on the growth pattern, an additional orthodontic treatment with frequent visits and good cooperation will be necessary.

References