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Case Report
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Unusual Craniofacial Manifestations in a Patient with DiGeorge Syndrome

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Abstract

A 10-year-old male with DiGeorge syndrome was referred to Kuwait University Dental Center with a chief complaint of generalized extensive carious lesions. Upon examination, the patient had poor oral hygiene, severe chronic generalized gingivitis, and gross carious lesions on all permanent teeth with the exception of the lower left first premolar. The caries also included the lower permanent central incisors. The medical history identified that the patient was previously treated for surgical correction of Tetralogy of Fallot. The purpose of this case report was to discuss the oral and maxillofacial findings observed in a patient with DiGeorge syndrome.

Introduction

Dr. Angelo M. DiGeorge in 1968 described major features and clinical appearances found in four infants. Those findings led to a remarkable discovery of the role of the thymus gland in the human immune system as well as a syndrome that was later understood to be caused by a deletion in a specific chromosome [1]. DiGeorge syndrome (DGS) is the result of micro deletion of chromosome 22q11.2 during the 4th to 6th week of formation, resulting in the abnormal development of the third (III) and fourth (IV) pharyngeal pouches. It is the most common micro deletion syndrome in humans with an incidence of 1:4000 in the community [2,3]. As a result of this deletion, there is absence or hypoplasia of the thymus, which is responsible for the maturation of T-cells. It is also responsible for the absence of parathyroid glands which main function is the regulation of calcium to the bone and teeth. In addition, failure of proper growth of III-IV pharyngeal pouch complex results in the failure of cardiac neural crest cells to infiltrate the pharyngeal pouches properly, thus leading to neural crest defects and subsequently congenital heart problems. DiGeorge syndrome phenotypic spectrum is wide. This includes heart defects, skeletal abnormalities, velopharyngeal insufficiency with or without cleft palate, immune problems, hypocalcemia, learning disabilities and behavioral abnormalities [4]. Common facial features of 22q11.2 syndrome are narrow palpebral fissures, hypertelorism, malar hypoplasia, micrognathia, broad nasal bridge with bulbous tip, small shaped ears, short philtrum, and abnormal facial height [5-7]. The oral cavity involvement has been highly overlooked, despite its relation with the facial region and the pharyngeal pouches formation [8,9].

Case Presentation

Medical and Social History

A 10 year old male was presented to Kuwait University Dental Center (KUDC) for dental treatment. A detailed medical, social, and dental history were obtained. The patient was diagnosed with DGS at birth. A medical history of hypocalcaemia (1.54 mmol/L) and vitamin D deficiency, congenitally missing thymus gland, surgical correction for Tetralogy of Fallot, and frequent recurrent infections was recorded. Social history showed that the parents were consanguineous and the patient has poor social skills. The parents also reported that the patient has learning difficulties, and attention problems.

Clinical Findings

A complete clinical examination was conducted. Extra-orally, the patient had superior frontal bossing, broad face with decreased vertical height, malar hypo deficiency, small cup-shaped ears,

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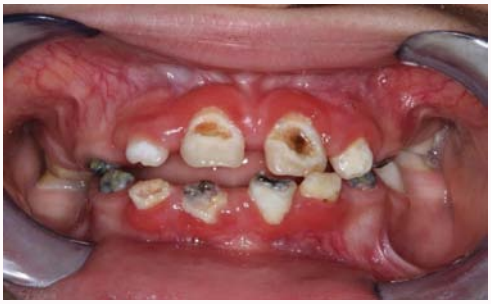

Figure 1: Pretreatment intraoral frontal photograph at the first appointment showing gross decay, enamel defects, and severe chronic generalized gingivitis.

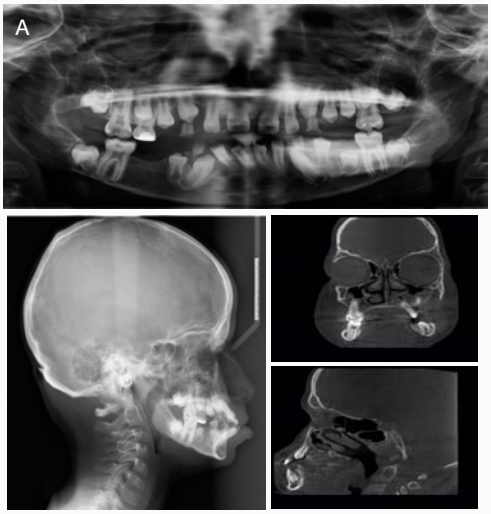

Figure 2: Pre-treatment radiographs of patient demonstrating unusual findings: (A) Panoramic radiograph showing close proximity of the apices of both lower first molars with the inferior border of the mandible. Loss of cortical definition was noted and the mandible appeared hypoplastic with a decrease in both the vertical dimension of the body of the mandible and the horizontal dimension of the ramus of the mandible. The mandibular right 2nd premolar was congenitally missing. (B) Lateral cephalometric radiograph showing evidence of mid facial and gonial angle deficiency together with reduced vertical dimensions of the mandible. (C and D) Coronal and sagittal cone beam CT images showing bilateral increase in the thickness of the posterior lateral wall of the right and left maxillary sinuses which gave the appearance of a minified right and left maxillary sinus and there was also an increased in thickness of the soft palate.

hooded eyes with narrow palpebral fissures, hypertelorism, broad nasal bridge with a bulbous tip, and lip protrusion. Intra-orally, the patient was in the mixed dentition phase. The patient had severe chronic generalized gingivitis. Plaque accumulation appeared to cover all the teeth (Figure 1). There was excessive bleeding from the gingival upon gentle probing. Multiple active carious lesions were present. Enamel opacities were noticed on all upper and lower permanent incisors. The patient suffered from teeth sensitivity in his lower incisors. Only the lower left first premolar was completely intact. In addition, generalized spacing and marked bimaxillary proclined incisors were observed.

Radiographic Findings

A panoramic radiograph (Figure 2A), a full mouth series, a lateral cephalometric radiograph and a cone beam CT study (160mm x 160mm field of view kV 90 mA10) were obtained for the patient. There was evidence of generalized osteopenia with large marrow spaces and scanty trabeculation. There was also generalized loss of cortical definition in the craniofacial area. Evidence of mid facial and gonial angle deficiency was seen in the lateral skull view (Figure 2B). The mandible appeared hypoplastic with a decrease in both the vertical dimension of the body of the mandible and the horizontal dimension of the ramus of the mandible. The mandibular right 2nd premolar was congenitally missing. The skull appeared slightly dolichocephalic. There was bilateral increase in the thickness of the posterior lateral wall of the right and left maxillary sinuses which gave the appearance of a minified right and left maxillary sinus (Figure 2C). In comparison, the sphenoid sinuses appeared large. There was evidence of enlarged adenoid glands and the right and left tonsils. There was also an increased in thickness of the soft palate (Figure 2D). The teeth had large pulp chambers and multiple dilacerations in the lower anterior incisors. Orthodontic measurements revealed that both jaws were prognathic with skeletal class I relationship. There was generalized spacing (+15 mm in the mandible and +8.4 mm in the maxilla) and the anterior teeth were proclined and in bimaxillary occlusion (Max1 to SN =128, Man1 to MP = 127).

Discussion

Unexplained deletion of chromosome 22q11 causes a wide variations of clinical manifestations. The severity and the extension of these manifestations remains a major problem when treating patients with DGS. Moreover, little is known about the oral and maxillofacial complications of DGS. Most of the extra-oral findings in the present case study are consistent with those reported in the literature [7-9]. However, this is one of the few case reports that focuses on the dental aspect of DGS. Several dental manifestations were involved in this patient including teeth with enamel opacities, generalized active carious lesions, and hypodontia in accordance with previous studies [5,8-12]. In the present case study, the patient presented with a wide deficient gonial angle and bimaxillary proclination of incisors, in contrast to a small intact gonial angle and only maxillary proclination of incisors identified in a previous case report from the literature [5]. New oral findings were observed in the current case report of DGS: root dilacerations, severe spacing, malformed mandible, defective mineralization of the skull, increased thickening of soft palate, and bimaxillary proclination. Other associated oral and maxillofacial abnormalities that were reported by other case reports and not in the current report include: irregular teeth shape, hypoplastic masseter muscle, asymmetric perioral muscles, class II/III malocclusion, geographical/fissured tongue, and cleft palate [8,10,12].

Early prevention and intervention are crucial for this patient group to prevent tooth decay and early tooth loss. Evaluating the level of psycho-social difficulties for patients with DGS by a psychiatrist is highly recommended since the development of schizophrenia is very common during adulthood for these cases [4]. It may also help guide the treating dentist in setting up an appropriate oral hygiene maintenance intervention for patients that suffer from these psycho-social disabilities. Attention problems and poor social skills, which were manifested in our case, made undesirable oral hygiene maintenance results which were aggravated by unsupervised teeth brushing. Analysis of dental radiographs should be performed as early as possible to assess atypical development of teeth and bone. Hypocalcemic attack in infancy causing teeth with opacities is very common in DGS patients, thus frequent recalls for such patients are required. Further research is needed to further understand the etiology, manifestations, and management of these rare syndromes.

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