Case Report

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Di George Syndrome: Catch 22-A Case Report

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Abstract

The deletion of chromosome 22q11.2 is described as Velocardiofacial Syndrome, Shprintzen syndrome or Di George Syndrome. CATCH 22 stands for cardiac defect, abnormal faces, thymic hypoplasia, cleft palate, hypocalcaemia. Other defects seen are velopharyngeal insufficiency with or without cleft palate, immune problems, feeding difficulties, hypocalcaemia, learning disabilities, behavioral abnormalities and lastly characteristic facial features. A high prevalence of dental caries, abnormalities of tooth shape, eruption and number, and enamel defects such as hypomineralisation and hypoplasia are also seen in these patients. A case of 7 year old child with Velocardiofacial syndrome is discussed in this article. Facial dysmorphism and common dental manifestations is typically noticeable in patients with this syndrome. Enamel aberrations related to hypocalcemia may result in a higher frequency of dental caries. The dentists need to be aware of the dental features of this condition in order to refer them to the adequate specialists.

Keywords: Digeeorge, Catch 22, Dental caries.

Introduction

DiGeorge syndrome (DGS) is a rare genetic disorder resulting from a defect involving third (III) and fourth (IV) pharyngeal pouches and arches, during embryonic development [1-3]. This 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 [4]. It is the most common micro deletion syndrome in humans with an incidence of 1:4000 in the community [5,6]. 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 whose main function is the regulation of calcium to the bone and teeth.

Dr. Angelo M. DiGeorge in 1968 described major features and clinical appearances found in four infants [7]. The main features are congenital heart disease, absence

J Den Max Surg, 2(1): 128-131 (2019)

or hypoplasia of thymus, hypoparathyroidism with consecutive hypocalcaemia.

DiGeorge syndrome phenotypic spectrum is wide as it includes heart defects, skeletal abnormalities, velopharyngeal insufficiency with or without cleft palate, immune problems, hypocalcemia, learning disabilities and behavioral abnormalities. Characteristic facial features are malar hypoplasia, small and down-slanting palpebral fissures, low nasal bridge, small mouth, malformed auricles and prominent nose [8-10]. Oral manifestations reveal delayed eruption of permanent teeth, enamel hypoplasia, enamel hypomineralization, hypodontia, aberrant tooth shape and dental caries (common oral problem) [9,10].

Case Report

A 7 year old boy reported to the department of Pediatric and Preventive Dentistry. A detailed medical, social, and dental history was obtained. The patient was diagnosed

with DGS at birth, (according to FISH study which is conducted by using D22S75 (Aquarius-Cytocell) probe specific to Di George Syndrome, one of the homologous chromosomes 22 could not be detected.)

A medical history of hypocalcaemia and vitamin D deficiency, congenitally missing thymus gland, and frequent recurrent infections was recorded. The patient was treated for isolated cleft palate (soft palate) and subsequent speech therapy at the age of 4 years. 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, a short philtrum, hooded eyes with narrow palpebral fissures, hypertelorism, broad nasal bridge with a bulbous tip, micrognathia, Class III malocclusion and lip protrusion (Figure 1a, Figure 1b).

Figure 1a: Patient shows specific extraoral features.

Figure 1b: Showing micrognathia in maxilla.

Intra-orally, the patient was in the mixed dentition phase. Multiple active carious lesions were present. Upper anterior teeth were grossly decayed with root stumps present. Only the upper right canine and lower left permanent lateral incisor were completely intact. (Figure 2,3) Patient had small mouth opening and was quite uncooperative which made it difficult to take proper photographs.

Figure 2: Showing dental decay in maxilla.

Figure 3: Showing dental decay in mandible.

Patient was highly uncooperative and therefore was referred for treatment under general anesthesia.

Discussion

DGS causes a wide variations of clinical manifestations. The severity and the extension of these manifestations remains a major problem when treating patients. Most of the extra-oral findings in the present case study are consistent with those reported in the literature [11-13].

Delayed growth and hypocalcemia were seen in our patient as a result of hypoparathyroidism, along with mild mental retardation.

Recurrent infections occurring in patients with DGS have negative impacts on providing the oral hygiene. These infections increase the risk of dental caries in children as these affect the dietary intake, resulting both in a higher frequency and increased consumption of products rich in carbohydrates to increase the child's energy intake. Oral hygiene is also adversely affected due to these infections [14]. Our patient manifested with low intelligence and highly uncooperative behavior which also lead to undesirable oral hygiene maintenance along with consumption of sugar containing cough syrups.

Some studies have claimed that feeding and speech problems have high rate incidence in CATCH 22 syndrome and speech problem is related to cleft which principally accompanies this syndrome [15,16]. Our patient also showed a medical history of soft palate cleft and was on speech therapy.

According to some authors, these patients should be followed for a long time, because depression and anxiety may precede the establishment of psychosis. Frequent infections due to dysfunction of T lymphocytes, with possible absence of cellular immune response may be attributed to thymus agenesis. In our case, patient gave history of thymus agenesis and was being treated for recurrent infections.

Many cases are not detected at birth or in childhood because DGS is a progressive disease and clinical signs that may appear later in life. At least 30% of the patients do not have heart defects, and many cardiac malformations, such as right-sided aorta, are "silent" anomalies and have no obvious clinical signs or symptoms [17].

Practical Guidelines for Managing Patients with 22q11.2 Deletion Syndrome have been framed. These provide an overview of management and the specialties commonly involved. There are recommendations for the "at diagnosis" stage and later developmental stages. These also give cautions and considerations that may be encountered by any clinician involved in the patient's care [18].

Early prevention and intervention are crucial by the dentists to provide a healthy oral cavity for these patients and hence further research and studies need to be carried out.

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