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CASE REPORT

Oral Manifestation and Dental Management of CATCH 22 Syndrome

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Abstract

The deletion of chromosome 22q11.2 is described as CATCH 22, Velocardiofacial Syndrome or Di George Syndrome. The acronym of CATCH 22 stands for cardiac defect, abnormal faces, thymic hypoplasia, cleft palate, hypocalcaemia but the acronym does not express all of the symptoms of CATCH 22 syndrome. Some clinical findings of CATCH 22 relate to congenital cardiac defects, velopharyngeal insufficiency with or without cleft palate, immune problems, feeding difficulties, hypocalcaemia, learning disabilities, behavioral abnormalities and lastly characteristic facial features. A treatment protocol of a 7-year-old child with CATCH 22 syndrome who also has dental caries complaint, toothache has been presented. Dental caries treatment and prophylactic application have been done for the patient. As a result, this study basically depicts how a dental approach can be followed for those patients who have CATCH 22 syndrome.

Keywords: CATCH 22 syndrome, FISH, thymic hypoplasia

Introduction

CATCH 22 syndrome occurs as a result of the deletion of chromosome 22 in 11.2 part. Incidence is approximately 1 in 4000 live births. The incidence in females is higher than in males. CATCH 22 Syndrome is inherited in an autosomal dominant manner. This syndrome is also known as Velocardiofacial Syndrome or Di George Syndrome (1-4). There are some clinical manifestations of this syndrome such as heart defects (74%), velopharyngeal insufficiency with or without cleft palate (69%), immune problems (77%), characteristic facial features (80%), hypocalcaemia (50%), renal anomalies (37%), learning disabilities (70-90%), hearing and speech problems, skeletal disorders, behavioral abnormalities and lastly feeding problems (1, 5). Characteristic facial features are malar hypoplasia, small and down-slanting palpebral fissures, low nasal bridge, small mouth, malformed auricles and prominent nose (5-7). Oral manifestations reveal delayed eruption of permanent teeth, enamel hypoplasia, enamel hypomineralization, hypodontia, aberrant tooth shape and dental caries (common oral problem) (7, 8). CATCH 22 Syndrome is diagnosed by fluorescence in situ hybridization (FISH). In order to cure some symptoms accompanying CATCH 22 syndrome, surgery approach for heart defects, calcium support, prophylactic antibiotic use, psychiatric pursuit and symptomatic approach can be driven (1). The purpose of this report is to present a new case of CATCH 22 syndrome.

Case Report

A 7-year-old female child with CATCH 22 syndrome was referred to Pediatric Dentistry Department of Marmara University with the complaint of caries and toothache. As a result of anamnesis taken from the parents of the patient, we have obtained some general information for our case, which are as follows:

History

The patient was delivered after 40 weeks of gestation. The pregnancy was smooth. Her parents are normal both physically and mentally. She also has 4 siblings who are sanitary. Her birth weight and length were 3250 gr and 49 cm, respectively. Her parents were referred to a hospital with her tremor complaint when she was 14-days old. Biochemical and hematologic tests were performed and hypocalcaemia (blood Ca: 5.7 mg/dl) and parathyroid hormone (PTH) (6.7 mg/dl) were detected. Results of PTH test revealed that primary hypoparathyroidism was responsible for the hypocalcemia and an immunologic examination showed the presence of a T cell dysfunction. According to immune phenotyping; low CD8 (14.9%) was detected, other CD values were normal. Agenesis of the thymus was revealed by thoracic CT scan. Echocardiography demonstrated cardiovascular abnormality (peripheral pulmonary stenosis). Physical examination of the face revealed hypertolerism, a short philtrum, thick reflected lips, micrognathia, narrow palpebral fissures, prominent nose and protuberant ear (Figure 1).

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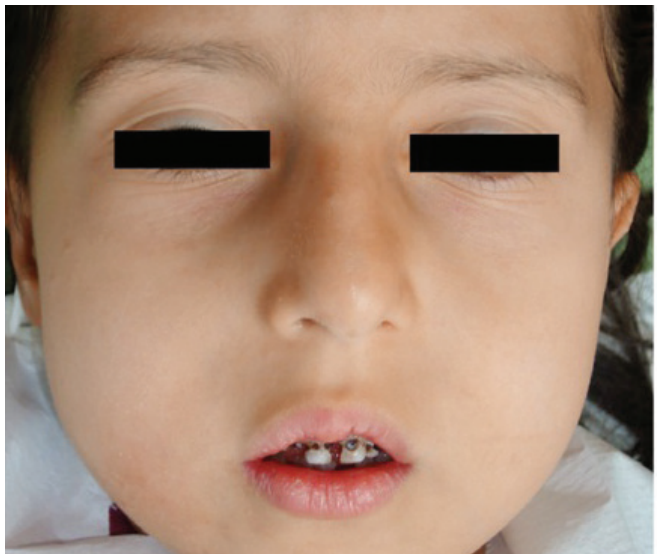
Figure 1. Facial appearance of the child.



According to cytogenetic and molecular cytogenetic analysis report, numeric or gross chromosomal abnormality in chromosomal analysis with High Resolution Bonding Technique (HRBT) was not detected. Nevertheless, 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. This result is consistent with the clinical manifestations in CATCH 22 Syndrome. Her present weight and length are 20 kg and 114 cm, respectively. She is under the supervision of a pediatrician. She also uses medication regularly.

Oral Findings and Principle of Treatment

She has poor oral hygiene. Extra oral and intraoral examinations were performed and intraoral tissues were appeared as normal except for the teeth. A panoramic radiography that shows incipient and deep caries was taken (Figure 2). Deepened palate and small mouth was observed. Oral examination of the patient reveals dental caries (all of the primary teeth) and dental abscess (#54 tooth) (Figure 3). As a result of consultation with her cardiologist, we did not use prophylactic antibiotics during the dental treatments. Dental caries of primary teeth was treated without local anesthesia and teeth were restored with compomer resin (Figure 3). The primary tooth with dental abscess (#54) was treated via root-canal treatment. All necrotic tissues were removed from the teeth using file before irrigation with normal saline. The tooth was dried with sterile paper-points. Metronidazole and cotton pellets were placed into the pulp chamber respectively and sealed with temporary restoration. 2 weeks later, clinical symptoms and signs were not observed and the tooth was filled using ZOE paste that restored with compomer resin. Pit-fissure sealant was applied to chewing surface of the permanent teeth. After the whole treatment, APF gel was applied to full mouth. At the end of one year follow-up of the patient, new carious lesions were not observed and the patient had a good oral health care.

Figure 2. Panoramic radiography.

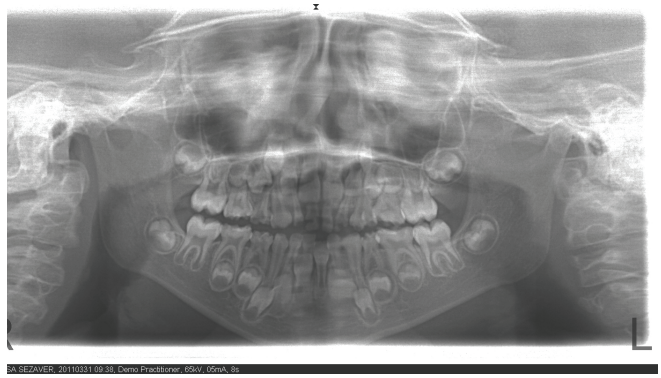
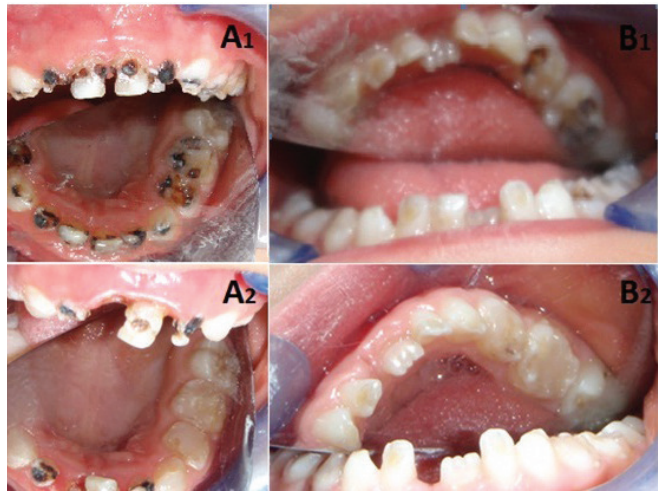


Figure 3. Intra-oral status before the treatment: A1, Maksillary teeth. B1, Mandibular teeth. A2, Maksillary teeth before the treatment (Figure out the arrow showing a dental abscess). B2, Mandibular teeth after the treatment.



Discussion

As it is known, dental caries is generally due to diet-eating habits and lack of oral hygiene in healthy individuals (9). The dental caries lesion of the patient influencing especially the primary dentition was both incipient and deep adversely. However, CATCH 22 syndrome might also cause dental caries in patients.

It has been reported that infections occurring often in patients who have CATCH 22 syndrome have negative impacts on providing the oral hygiene. These infections increase the risk of dental caries in children (7, 9). Previous studies depict that infections concomitant to the syndrome may 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 (7, 9). Furthermore, because of these infections children cannot carry out the principles of the oral hygiene. According to a study conducted by Klingberg et al.(9) dental caries risk increases in patients who are afflicted with the CATCH 22 syndrome influencing the saliva secretion rate, buffer capacity, number of cariogenic bacteria, saliva total protein, Ig A and electrolytes concentrations.

Moreover, enamel defects occur due to hypocalcaemia, hypoparathyroid and reduced level of both calcium-phosphorus

(9, 10). Previous studies indicate that enamel defects are common findings in CATCH 22 syndrome (6, 7, 10, 11). This was not seen in our patient although she has hypocalcaemia and takes in calcium support. Furthermore, Klingberg et al. (7) states that enamel defects such as hypoplasias or hypo mineralization with disintegrated enamel may better retain dental plaque increasing the risk for dental caries. This could not be demonstrated in our clinical study despite the fact that numbers of decayed teeth were significantly high.

Tooth agenesis and supernumerary teeth are not the characteristic features of the syndrome CATCH 22 (3, 6). Klingberg et al. (7) argues that hypodontia has been rarely seen in primary and permanent dentition. Nevertheless, hypodontia has not been observed in our case according to results of panoramic radiography taken from the patient. It should be noted that a research needs to be conducted on the patients who have CATCH 22 syndrome with hypodontia.

As it is known from the previous studies, feeding and speech problems in patients are the characteristic findings of this syndrome (1, 3, 5, 12, 13). Nugent et al. (3) claims that feeding problem has high rate incidence in CATCH 22 syndrome and speech problem is related to cleft which principally accompanies CATCH 22 syndrome. On the other hand, according to Solot et al. (14), speech problems are common and they are independent of palatal defects. In fact, speech problems are closely relevant to cognitive factors and typical facial phenotype. In our study, this was the case but palatal defects were not observed in the patient, even though there was a speech problem. This illustrates that there exists a similar approach between our study and the study of Solot et al. (14), when speech problems of the patients are considered.

Thymic agenesis may lead to frequent infections due to dysfunction of T lymphocytes, with possible absence of cellular immune response (6). Immunodeficiency occurs as a result of thymic agenesis. In our case, thymic agenesis and immunodeficiency were observed.

Peripheral pulmonary stenosis was monitored by using echocardiography in our case. Poor oral hygiene causes risk of endocarditis in the cardiac patients. For this reason, oral hygiene practice ought to be given to the patient and to his/her family as a protective precaution. Bacteremia resulting from dental treatment can be fatal for the patients who suffer from CATCH 22 syndrome. It can be inferred from the study of Szczawinska-Poplonyk et al. (8), that dental treatment of the children with CATCH 22 syndrome should be directed to pediatric dentistry. Preventive dental treatment is necessary for the patients with CATCH 22 syndrome. Pit-fissure sealant and fluoride implementation have been done in our case as a preventive dental treatment so as to prevent the permanent teeth from dental caries.

Briefly, there are some oral and general findings of CATCH 22 syndrome. Appropriate dental treatment protocol should be determined in consideration of manifestations of the syndrome. Dentists should provide a healthy oral cavity for the patients who have CATCH 22 syndrome. As a consequence, this study basically depicts how a dental approach can be followed for those patients who have CATCH 22 syndrome.

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