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Catch 22 Syndrome dental anomalies at the earliest: A Rare Case Report

[1]Dr Joby Peter, [2]Dr Vijai S, [3]Dr Krishnakumar, [4]Dr Anashwara MS, [5]Dr Anusree

[1-5]Malabar Dental College & Research Center Mannor Chekkannor Road Edappal, Malappuram

Corresponding Author: Dr Krishnakumar, Reader, Malabar Dental College & Research Center Mannor Chekkannor Road Edappal, Malappuram

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Introduction

Di George syndrome is one of the most common multiple anomaly syndromes. The labels velo-cardio-facial syndrome, 22q11 deletion syndrome, cono-truncal anomalies face syndrome, CATCH 22, and Sedlackova syndrome have all been attached to the same disorder. [1] The incidence is reported to vary from 1:4000 – 1:5000 live births. The incidence in females is usually found to be higher than in males. CATCH 22 Syndrome is inherited in an autosomal dominant manner. Usual cause is micro-deletion of 22q11.2 sequence[2] and hence it will result in the loss of 30-40 genes and this can be detected by using specific probes in fluorescence in situ hybridization (FISH) analysis. But in a minority of cases, a mutation in TBX_1 can also be detected.[3] There are some clinical manifestations which are exhibited by this syndrome such as heart defects (74%), velo-pharyngeal 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, behavioural abnormalities and lastly feeding problems.[4] The most frequent features include mild eye (narrow palpebral fissures, small optic discs) and ear anomalies (low set ears, abnormal folded pinna, middle ear abnormalities, hearing deficits), alterations in nasal anatomy, slender hands and digits, reduced intelligence and short stature.[5,6] Oral manifestations includes delayed eruption of permanent teeth, enamel hypoplasia, enamel hypo-mineralization, hypo-dontia, aberrant tooth shape and dental caries.[7]

Little information is available in the field of dentistry about this syndrome. Information on the oral characteristics and dental features might be useful as well as the histological data of dental structures will serve as an additional data for evaluation of patients with suspected diagnosis of syndrome and to provide periodic care for their dental needs for a better living hood.

Case Report

A 5 1/2-year-old male patient presented with the complaint of pain in the lower left back tooth region since one week. Past dental history revealed frequent dental pain and swelling for which he had taken medications. But no dental treatments were carried out for the same. There was no relevant familial history. He was the second child in the order of birth born to non-consanguineous parents. The Prenatal & Post Natal history revealed that pregnancy was normal, the birth uncomplicated at term. The birth weight and length were normal. At the very next day from his birth, he had developed multiple episodes of epileptic seizures lasting for 3-5 minutes and the baby was admitted in the ICU. On Radiographic investigation it was diagnosed that thymus was not present at birth. On further investigatory test results revealed the presence of hypo-calcemia and minor cardiac defect. The tendency towards infections in combination with hypo-calcemic seizures, heart malformations (atrial septal defect) led to suspicion of Di George syndrome. A FISH analysis (Fluorescent in situ Hybridization analysis) was performed on metaphase chromosomes from cultured blood sample of this patient using LSI TUPLE1/ARSA probe from Vysis inc., USA. The result showed only one signal (red) on one of the chromosomes 22 at 22q11.2 region and the signal (red) was absent on the other chromosomes 22 in all the metaphases analysed and hence confirmed with the presence of Di George Syndrome. The developmental milestones were delayed. He started walking and talking late and has a nasal twang during speech. Parents has also noticed a slight delay in the eruption of primary dentition. His sibling (elder brother) is leading a normal life. The main therapeutic problem throughout the years, however, was recurrent serious infections, mostly of the respiratory tract.

Extra-oral examination revealed a dolichocephalic skull with a leptoprosopic face. Lateral profile of the patient showed mid-face deficiency. Face was mildly flattened, downward slanting palpebral fissures, and slightly retrognathic mandible. The ears were low-set and there were no apparent TMJ problems. He seemed to have non-competent lips, and a short mouth which is open at rest position (Figure 1).

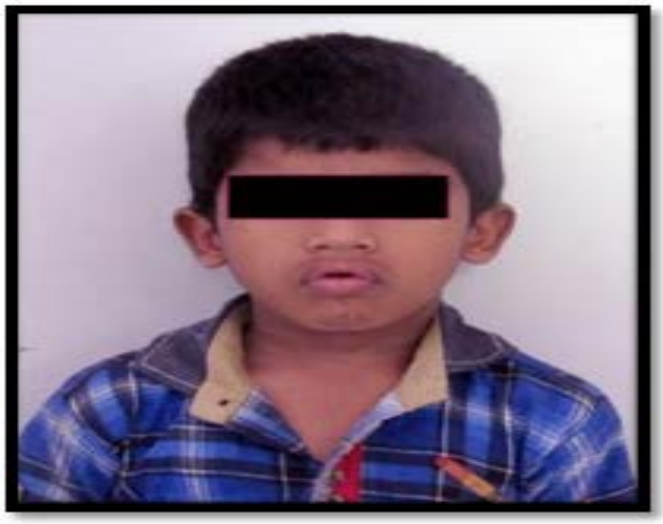

Figure 1: Profile view

Intraoral examination revealed poor oral hygiene with halitosis and slight gingival inflammation. Varying degrees of enamel hypoplasia as well as Type III ECC can be appreciated during examination (Figure 2, 3 and 4).

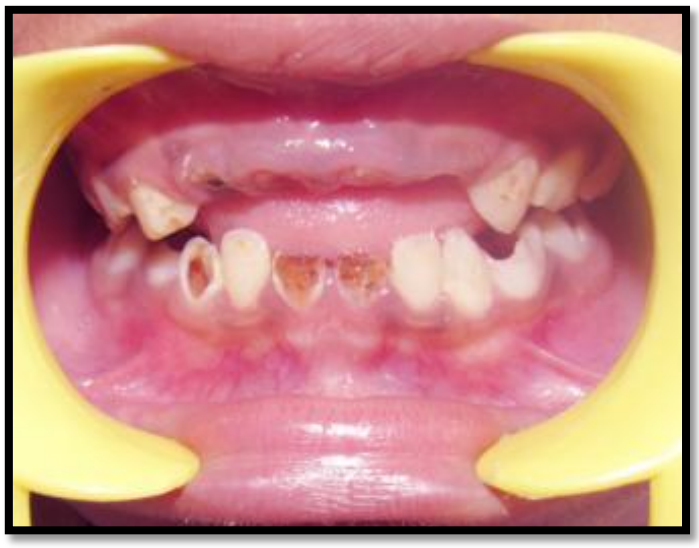

Figure 2 : Anterior frontal view

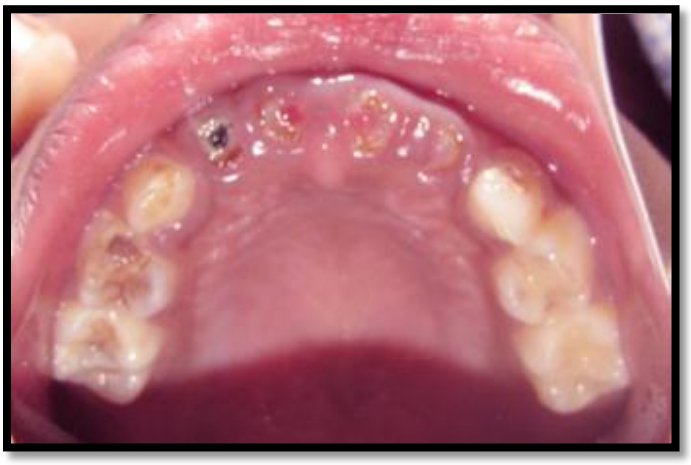

Figure 3 : Upper occlusal view

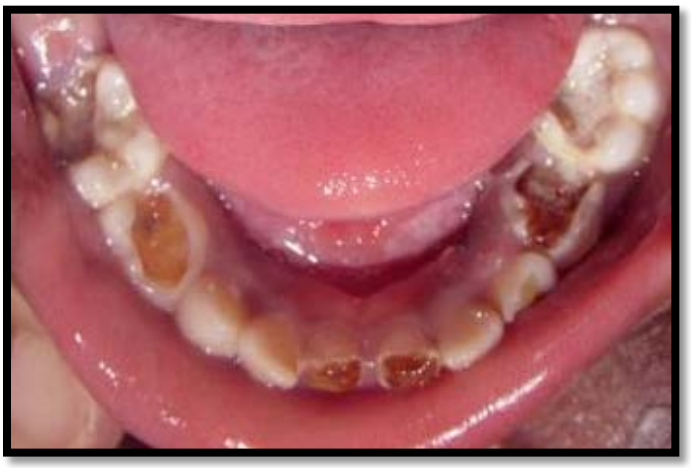

Figure 4 : Lower occlusal view

Radiographic examination revealed distinct radiolucencies in the enamel in relation to 55,54,64,65,71,74,81,83 and radiolucency involving pulp in relation to 75, 84,85 and root stumps of 51,52,61 and 62 were observed (Figure 5).

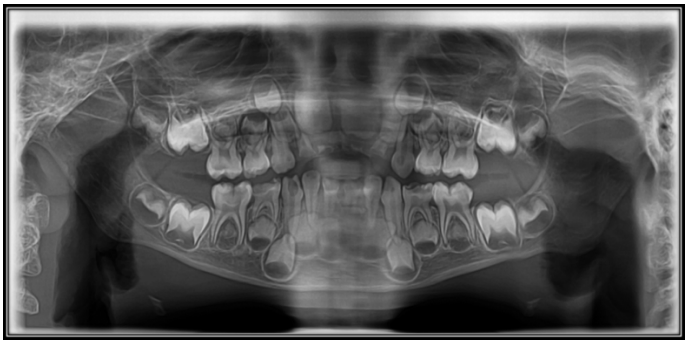

Figure 5 : Orthopantomograph showing Type III ECC

Dental Management

After conducting haematological, endocrinology and biochemistry tests and analysing present calcium (8.5 mg/dl), E.S.R (25 mm/hr), T3 (1.02 ng/ml), T4 (9.62 ug/dl), TSH (2.61 ulU/ml) and 25-OH VITAMIN D (34.12 ng/ml) results, informed consent from his paediatrician was obtained.

As part of familiarising with dental procedures, he was made comfortable by starting off with simple procedures like oral prophylaxis (hand scaling) and fluoride applications. Oral hygiene instructions were given to the parents. In the further visits simple GIC restorations for 54, 64, 65 and 74 were performed followed by pulp therapy for 75 and 85 were performed. SDF application for 55 followed by GIC restoration and SDF application for 83 followed by strip crown were performed. Root stumps i.r.t.52, 51, 61, 62 were extracted. 71 and 81 were also extracted due to physiologic mobility and a ground section of the extracted tooth (71 & 81) was performed. Extraction of 84 was also done due to its increased root resorption and mobility. Stainless steel crown were given i.r.t. 75 and 74 followed crown and loop space maintainer i.r.t. 84 (Figure 8). Groper's appliance was also given till the eruption of upper permanent incisors (Figure 9). Patient was recalled for follow-ups in the second week, third month and his improved oral hygiene was appreciated.

Histologic findings of deciduous teeth

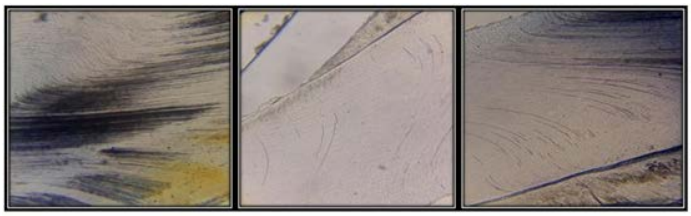

Figure 6 : Ground section performed on 71, 81

The ground section of extracted tooth 71 and 81 was performed. A mild irregularity could be appreciated in the

dentinal tubules which might be due to caries, otherwise, normal features were observed (Figure 6).

Post-Operative Radiograph And Profile

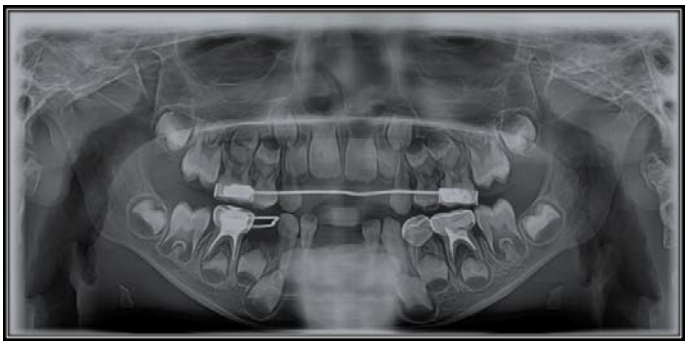

Figure 7 : Post - treatment Orthopantamograph

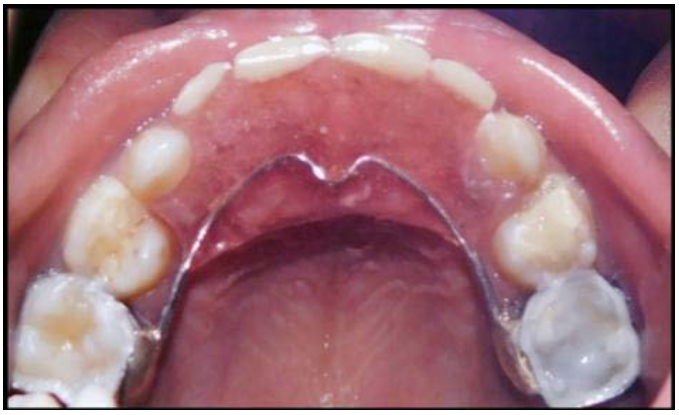

Figure 8 : Upper occlusal view after rehabilitation

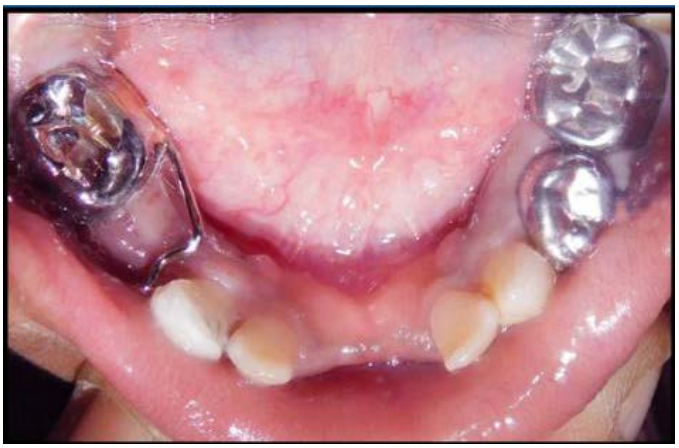

Figure 9 : Lower occlusal view after rehabilitation

Discussion

Di-George Syndrome was first described as a human thymic deficiency by Di George [8] and was established as an immunodeficiency caused by thymic aplasia accompanying defective parathyroid glands.[9] Since both thymus and parathyroid develops from the 3rd and 4th pharyngeal arches, any disturbance in its development in the 4 -7 week of gestation may lead to this syndrome. In hypo-parathyroidism occurring in infancy and early childhood, the possibility of the Di George syndrome makes investigation of immune competence mandatory. The management of patients with chromosome 22q11.2 deletion syndrome is highly dependent on age and phenotype. Patients with the chromosome 22q11.2 deletion syndrome might present at any age, although most patients receive their diagnosis shortly after birth because of the presence of a cardiac anomaly. In new-born babies, a thorough physical and radiographic examination should seek medical problems that are likely to need immediate intervention, such as cardiac anomalies, hypocalcaemia, severe immune-deficiency, or intestinal mal-rotation. The prognosis of this syndrome depends on the severity of the heart disease and the residual function of the incomplete thymus. The cause of death is infection, heart failure, or sudden death.[10]

While analysing patient's features in this case, a prominent forehead, hyper-telorism, anti-mongoloid facies, slanting of the eyes, low set ears, a short philtrum were able to be appreciated in accordance with previous descriptions of the Di-George syndrome[11].

Mild form of hypoplasia was observed and Type III ECC was confirmed in this case. Enamel defects present in this case may be due to hypocalcaemia, hypo-parathyroid and reduced level of both calcium-phosphorus in the childhood. According to a study conducted by Klingberg et al. (2007) 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, IgA and electrolytes concentrations.

Appropriate dental treatment protocol was determined giving consideration to the manifestations of this

syndrome. As it is known, dental caries is generally due to diet-eating habits and lack of oral hygiene in healthy individuals. So children with these syndrome find it difficult to maintain adequate oral hygiene, thus appropriate age related preventive and supportive measures were taken along with diet modification and proper guidance for oral hygiene. Periodic examination and treatment measures are necessary and crucial for maintaining good oral health. The anterior aesthetic Groper's appliance was given until the permanent erupts which offered him with a confident smile on his face.

Conclusion

Di George otherwise CATCH 22 Syndrome is a most common micro deletion syndrome. The prenatal diagnosis and the genetic counselling are recommended in choosing a responsible management of CATCH 22 syndrome cases. This case report attempts to throw some light on this syndrome with its oral manifestations.

Declaration Of Patient Consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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