DiGeorge Syndrome: A Case Report

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Abstract

DiGeorge syndrome (DG) is a genetic disorder associated with 22q11 deletion. It involves various phenotypes, including craniofacial abnormalities, congenital heart disorders, endocrine dysfunction, cognitive deficits, and psychiatric disorders. DG cases commonly involve multiple anomalies. [1]

This case report presents the etiology, symptoms, dental characteristics and implications of DiGeorge syndrome and the treatment rendered to a 3-year-old male patient with DiGeorge syndrome who was referred to Tufts University School of Dental Medicine for treatment of dental caries.
At his first dental visit after a limited examination and diagnosis of Severe Early Childhood Caries, Silver Diamine Fluoride (SDF) was applied for dental caries management[5] and full mouth dental rehabilitation under general anesthesia was planned and completed a few months after due to his significant limitations in intellectual function and adaptive behaviour . The full mouth dental rehabilitation consisted of a comprehensive oral exam, 2 bitewing and 6 periapical radiographs, oral prophylaxis, composite resin restoration on tooth #D, extractions of non-restorable teeth #E,#F,#O,#P and topical fluoride (5%NaF varnish) application.

Introduction

DiGeorge syndrome is a common chromosome 22q11.2 deletion syndrome known as velocardiofacial syndrome.
Demographics:

- 1:4000 births, males and females are equally affected. one-half of the children of affected adults will have the deletion. Therefore, the prevalence is anticipated to rise over time. Currently, the figures are 6%-10% of new cases are familial.

Disease Mechanisms:

- Deletion arises via an unequal meiotic exchange facilitated by asynchronous replication at the deletion site and development of a TBX1 gene which affects cardiac development and the formation of the fourth branchial arch.[2] (Fig1, Table1)

Intra and extra oral aspects:

- High prevalence of dental caries; abnormalities of tooth shape, eruption and number; and enamel defects such as hypomineralization and hypoplasia.
- Skeletal malformations, velopharyngeal insufficiency with or without cleft palate, small mouth, hypotonic orofacial musculature, impaired salivary flow and high frequency of dental caries. [3]

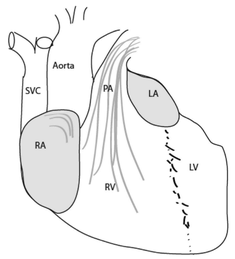


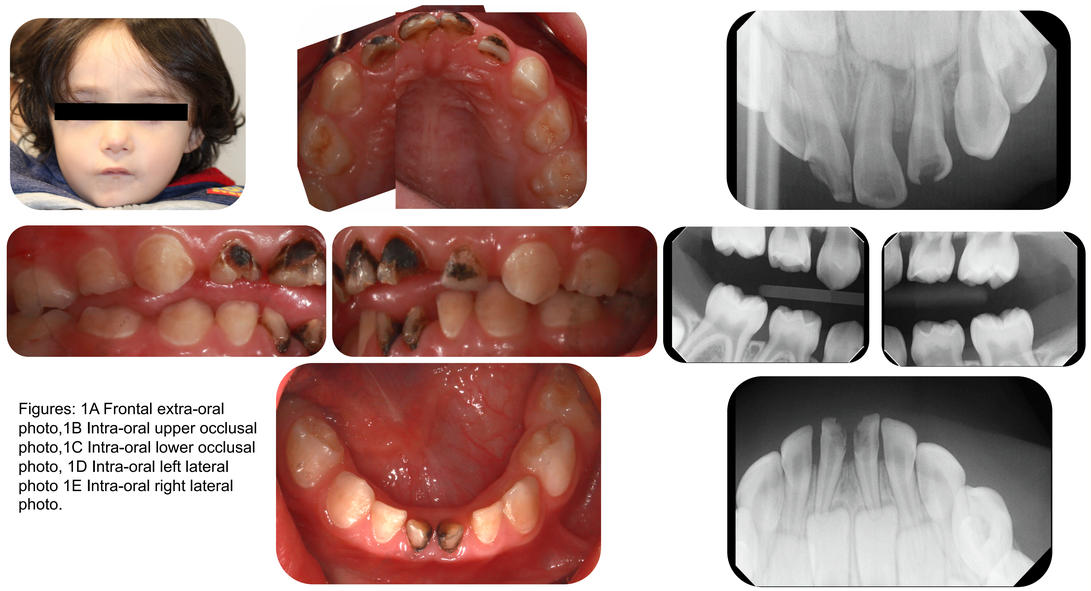
Fig1: TBX1 contributes to the right ventricle and outflow tract as well as the right atrium. The gray lines indicate the regions of the heart most consistently populated by cells derived from the anterior heart field that express TBX1. The aorta itself is infrequently populated, but the ductus arteriosus is almost completely derived from anterior heart field cells. Abbreviations: LA = left atrium, LV = left ventricle, PA = pulmonary artery, RA = right atrium, RV = right ventricle, SVC = superior vena cava.

Finding	Frequency (%)
Cardiac anomaly	
Interrupted aortic arch Type B	52%
Truncus arteriosus	35%
Tetralogy of Fallot	16%
Polyhydramnios	16%
Polydactyly	6%
Cleft palate	11%
Cleft lip	2%
Diaphragmatic hernia	1%
Spina bifida	<1%
Craniosynostosis	<1%

Table 2: Prenatal Ultrasound Findings in Patients with Chromosome 22q11.2 Deletion

Phenotypic Feature	Frequency of Deletion (%)
Any cardiac lesion	1
Conotruncal cardiac anomaly	7–50
Interrupted aortic arch	50–60
Pulmonary atresia	33–45
Aberrant subclavian	25
Tetralogy of Fallot	11–17
Velopharyngeal insufficiency	64
Velopharyngeal insufficiency postadenoidectomy	37
Neonatal hypocalcemia	74
Schizophrenia	1–25

Table 1 Frequency of the Chromosome 22q11.2 Deletion in Various Patient Populations



Figures: 1A Frontal extra-oral photo,1B Intra-oral upper occlusal photo,1C Intra-oral lower occlusal photo, 1D Intra-oral left lateral photo 1E Intra-oral right lateral photo.

Figures; 2A Right bitewing radiograph (BW), 2B upper occlusal PA, 2C lower occlusal PA, 2D left BW,

Case Report

A 3-year-old male with DiGeorge syndrome, Tetralogy of Fallot (TOF), and pulmonary insufficiency was referred to Tufts University School of Dental Medicine for treatment of caries. Limited dental examination revealed S-ECC and a submucous cleft palate (Fig1A,1B,1C,1D,1E).
Past Medical and Surgical History:
Patient was born at 41 weeks with birth weight 3.642 Kg (41.7 percentile), diagnosed with DiGeorge syndrome after birth and transferred to Tufts Medical Center (TMC) due to cyanosis. Echocardiogram obtained shortly after admission revealed TOF with a large ventricular septal defect (VSD), overriding aorta, dysplastic pulmonary valve, and dilated right coronary artery, with a right aortic arch with an aberrant left subclavian artery.(Fig 1)
Patient lives with both parents. The father has a history of (TOF).

Management

At his first dental visit after a limited examination and diagnosis of Severe Early Childhood Caries, Silver Diamine Fluoride (SDF) was applied for dental caries management[5] and full mouth dental rehabilitation under general anesthesia was planned for the patient due to his significant limitations in intellectual function and adaptive behaviour as well as due to the extent of necessary treatment.

Full mouth dental rehabilitation under general anesthesia was performed and consisted of a comprehensive oral exam, 2 bitewing and 6 periapical radiographs (Fig 2A,2B.2C.2D,2E,2F,2H), oral prophylaxis, composite resin restorations on tooth #D, extractions of non-restorable teeth #E,#F,#O,#P and topical fluoride (5%NaF varnish) application.

Post-op follow-ups were conducted via Teledentistry and subsequent clinical visits.

Conclusion

DiGeorge is a multisystem syndrome with wide variability in its severity and extent of expression in affected patients. 22q11.2DS could contribute to poor oral health. Thus, dental practitioners should consider early management to provide preventive and therapeutic interventions and thereby achieve optimum oral health. In addition, cooperation among and experience with different specialties is mandatory to improve quality of life for patients with 22q11.2DS.

Acknowledgement

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