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Orofacial findings and dental treatment in a 19 year old patient with trisomy 18 mosaicism: A case report

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Abstract

Trisomy 18 syndromes, also called Edwards' syndrome, is a chromosomal disorder due to the presence of an extra chromosome 18.It presents as either complete, mosaic or partial forms. The complexity and severity of the phenotypic presentation is extremely variable in individuals with mosaicism of trisomy 18, ranging from severe manifestations with early mortality to apparently phenotypically normal adults. Though the condition has occasionally been reported from a medical perspective, cases describing orofacial manifestations and approach to dental care have been rare. We describe the craniofacial and dental characteristics of a 19-year-old male with both the typical and less common findings of trisomy 18 syndrome. Our case adds to the existing knowledge of the spectrum of mosaic trisomy 18 from a dental perspective. This case report will enable dental professionals to recognize the phenotypic alterations of this syndrome, facilitating timely referral for necessary multidisciplinary treatments including preventive, restorative and prosthetic rehabilitation.

Keywords: Dental Management, Edwards' Syndrome, Mosaicism, Oral Manifestations, Trisomy 18

Introduction

Trisomy 18 or Edwards' syndrome is a chromosomal disorder first described by Edwards et al in 1960. [1] The presence of an extra chromosome 18 (trisomy) leads to noticeable major and minor anomalies in the affected individual. The condition is characterized by prenatal growth deficiency, distinct craniofacial features, typical features of hands and feet (clenched fists with overriding fingers, nail hypoplasia, short hallux, club feet), short sternum in addition to marked psychomotor and cognitive disabilities. Multiple congenital defects affecting cardiac, renal, pulmonary and genitourinary systems are also common.[2][3] Characteristic orofacial features include dolichocephaly, short palpebral fissures, ocular hypertelorism, micrognathia, external anomalies of the ears and redundant skin at the back of the neck. [2] An elevated risk of mortality has also been reported with 99% of children dying before age ten. [4]

Less than 5% of patients have mosaicism of trisomy 18 which is a less severe form of Edwards' syndrome, as only some of the cells have the extra copy of chromosome 18, rather than every cell. [2] These individuals exhibit extremely variable phenotype, ranging from severe manifestations with early mortality to apparently phenotypically normal adults. [5]

Though trisomy 18 has occasionally been reported in literature from a medical perspective, there is an extreme paucity of information regarding the orofacial manifestations and approach to dental management in individuals afflicted with this condition. The current article presents a rare case of trisomy 18 mosaicism aiming to contribute new knowledge and applicability to dental practice and to draw clinically useful conclusions.

Case History

A 19 year old male patient with trisomy 18 mosaicism reported to our private dental clinic in Kerala, India with the chief complaint of generalized toothache in both upper and lower jaws. He was the only son of healthy, non-consanguineous parents (46 year old father and 44 year old mother) and was born at full term by normal delivery with a birth weight of two kilograms. The prenatal and perinatal histories were non-contributory. Cytogenetic investigation of peripheral lymphocytes using GTG-banding revealed mosaic trisomy 18 with approximately 90% of trisomic cells.

On general examination he was found to have kyphoscoliosis, hypoplastic fingernails, short sternum and club feet with prominent heels. He had a poorly nourished physique, exhibited moderate psychomotor retardation and had communication disorder.

Dysmorphic facial features included dolichocephaly, elongated face, narrow bifrontal diameter, low set ears, downslanting palpebral fissures, humped nose and a small mouth with limited opening.

Oral manifestations included incompetent lips, narrow high-arched palate, anterior open bite and steep mandible.

Intraoral examination showed poor oral hygiene; bulbous, inflamed gingiva and moderate to severe caries involving all the teeth.

Radiographic evaluation of the panoramic radiograph revealed partial anodontia with caries affecting all the teeth present, alveolar bone and root formation were within normal limits (figure 1). Vertical growth pattern with increased mandibular plane angle was confirmed from a lateral cephalogram.

A comprehensive individualized treatment plan was formulated. Parents were given dietary counseling and were instructed about home care oral hygiene methods. The initial phase of treatment aimed at addressing the chief complaint. Medicines were prescribed, oral prophylaxis followed by topical fluoride application was done. Further invasive procedures were not undertaken on the first visit considering the inability of the patient to co-operate. With the parents' consent, periodic follow up visits were scheduled to carry out necessary restorative procedures. However, they failed to report back for the scheduled visits.

Two years later, when he was brought back for treatment, it was found that all teeth except four maxillary anterior teeth (11, 12, 13 and 21) had been extracted over time and the oral hygiene status remained compromised. The edentulous ridges were flabby with severe undercuts. (Figure 2, 3). The existing condition was explained in detail to the parents and a new treatment plan was devised after assurance from them regarding their role in successfully completing the treatment. Accordingly, root canal therapy was performed on the teeth present and porcelain fused to metal crowns were given. Fabrication of mandibular complete denture with maxillary removable partial denture was done in stages. (Figures, 4, 5)

Discussion

Trisomy 18 is a rare autosomal chromosomal disorder with three basic forms complete, partial and mosaic. Individuals having mosaic form carry both trisomic and euploid cell lines. [6]

Here we present a case of mosaic trisomy 18 with certain typical phenotypic features of complete trisomy 18 along with few alterations from normal, which, to the best of our knowledge, have not been previously described. This is one of the very few studies to report the specific oral features of a single patient with this condition.

Unlike individuals with complete trisomy 18, people affected with mosaicism may appear phenotypically normal with a greater probability of survival beyond childhood. [7] Our patient is an adult with moderate psychomotor and communication disorder.

Though there is no genetic basis, dental carious lesions have greater incidence in syndromic cases as reported by Gryst et al. [8]

Hermesch et al. reported nine cases of 18p deletion syndrome with high caries index which was the same as in our patient. [9]

The intraoral features presented here mostly resembled the case reported by Ribeiro et al. [10]

Unusual features like kyphoscoliosis, humped nose, downslanting palpebral fissures, narrow bifrontal diameter and communication disorder can be attributed to the chromosomal abnormality. Absence of serious systemic problems could have contributed to longevity in this case and no special considerations had to be given before or during dental treatment because of the same. Though the family was extremely caring about the general well-being of the patient, oral care was not given due importance. The family's commitment being an essential factor for maintaining adequate oral health in individuals with special needs was stressed upon during the counseling session for parents.

It is important to report such cases in literature as clinical presentation of such cases are quite variable. Early detection of cases can pave way for establishing individualized noninvasive preventive programs for these children as early as possible reducing the need for extensive dental procedures in future.

Figures

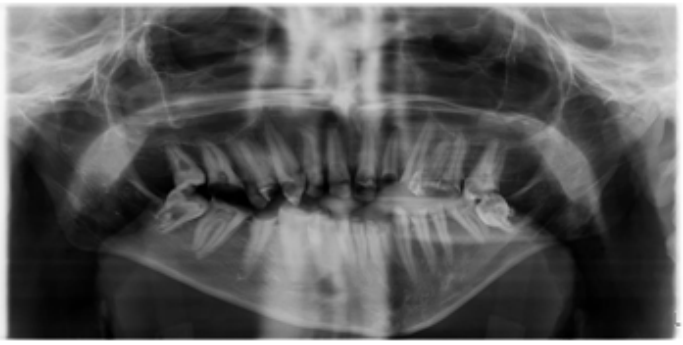

Figure 1: Orthopantomogram

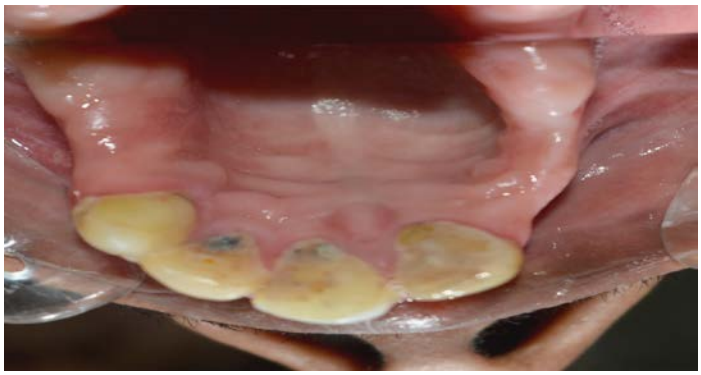

Figure 2: Partially edentulous maxillary arch

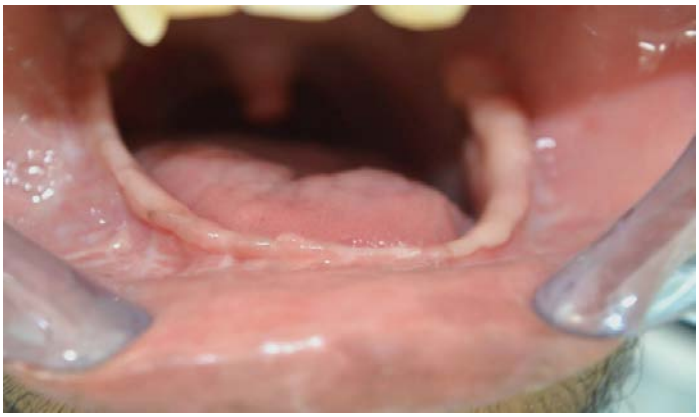

Figure 3: Completely edentulous mandibular ridge

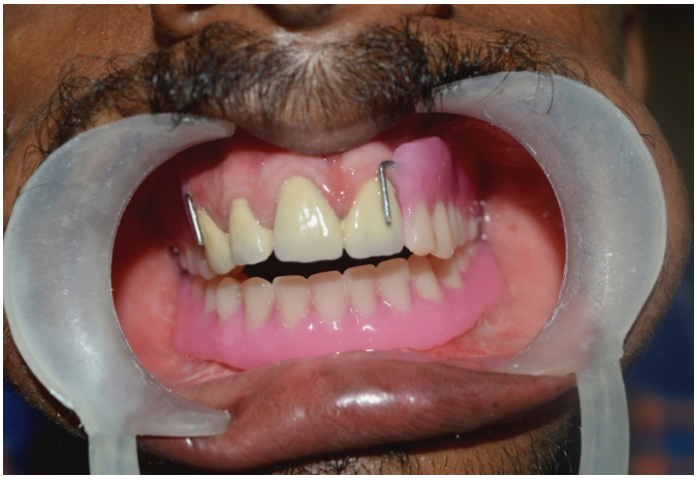

Figure 4: Mandibular complete denture with maxillary removable partial denture and porcelain fused to metal crowns on maxillary anteriors

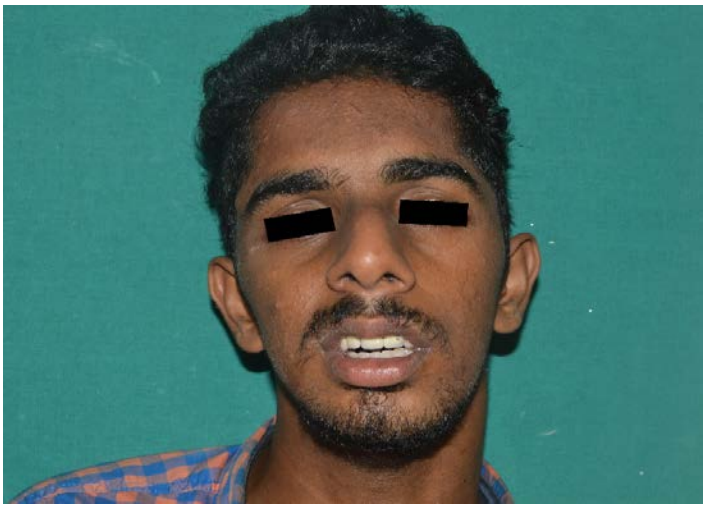

Figure 5: Extraoral appearance after full mouth rehabilitation

References

1. Edwards JH, Harnden DG, Cameron AH, Crosse VM, Wolff OH: A new trisomic syndrome. Lancet 1960, 1:787–789.
2. Cereda A, Carey JC. The trisomy 18 syndrome. Orphanet journal of rare diseases. 2012 Dec;7(1):81.
3. Crider KS, Olney RS, Cragan JD: Trisomies 13 and 18: population prevalences, characteristics, and prenatal diagnosis, metropolitan Atlanta, 1994-2003. Am J Med Genet 2008, 146A:820–826.
4. Nussbaum RL, McInnes RR, Willard HE Thompson and Thompson Genetics in Medicine. 6th ed. Philadelphia: WB Saunders Co 2001: 304-06
5. Greve G, Waaler PE, Rosendahl K**.** Low frequency mosaicism of normal cells in a 16-year-old girl with trisomy 18. Clin Genet 1993;43:83-7.
6. Tucker ME, Garringer HJ, Weaver DD. Phenotypic spectrum of mosaic trisomy 18: two new patients, a literature review, and counseling issues. American Journal of Medical Genetics Part A. 2007 Mar 1;143(5):505-17.
7. Ukita M, Hasegawa M, Nakahori T: Trisomy 18 mosaicism in a woman with normal intelligence, pigmentary dysplasia, and an 18 trisomic daughter. Am J Med Genet 1997, 68:240–241
8. Gryst ME, Mount GJ. The use of glass ionomer in special needs patients. Aust Dent J 1999 Dec;44:268-74
9. Hermesch CB, Cody JT, Cody JD. Dental caries history in nine children with chromosome 18p deletion syndrome. Spec Care Dentist 2000;20:53‑5.
10. Ribeiro RR, dos Santos BM, Stuani AS, deFreitas AC, deQueiroz AM. Dental findings and dental care management in trisomy 18: case report of a 13-year-old “long-term survivor”. Spec Care Dentist 2006;26:247-51.