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ABSTRACT

Trisomy 18 is a disorder characterized by psychomotor disabilities, dysmorphic features and organ malformations, including mental disabilities, growth deficiency, poor motor ability, micrognathia, microcephaly, low-set and malformed ears, distinctively clenched fists with overlapping fingers, and congenital heart defects. The prognosis is poor: 90% of infants with trisomy 18 do not survive beyond the first year of life and 99% die before the age of 10. This paper reports on a 13-year-old child diagnosed with trisomy 18. The major clinical features are cleft lip/palate, high-arched narrow palate, micrognathia, anterior open bite, posterior crossbite and taurodontism. Dental care management of these patients with special needs is discussed and the dental treatment for this child with trisomy 18 is described.

KEY WORDS: trisomy 18, Edwards syndrome, dental care, special needs patients

Dental findings and dental care management in trisomy 18: Case report of a 13-year-old "long-term survivor"

Roberta R. Ribeiro, DDS;[1] Bianca M. dos Santos, DDS;[1] Adriana S. Stuani, DDS;[2] Aldevina C. de Freitas, DDS, MSc, ScD;[3] Alexandra M. de Queiroz, DDS, MSc[3]*

[1]General dentist attending an advanced degree program in Dental care for special patients; [2]Postgraduate student at the Department of Pediatric Clinics, Preventive and Social Dentistry; Clinical Assistant; [3]Professor at the Department of Pediatric Clinics, Preventive and Social Dentistry; Attending Clinical Professor — Center of Formation of Human Resources Specialized in Dental Care for Special Needs Patients, at the Faculty of Dentistry of Ribeirão Preto, University of São Paulo, Brazil.
*Corresponding author e-mail: amqueiroz@forp.usp.br

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Introduction

Trisomy 18 was first described in 1960 by Edwards and colleagues[1] and has also been called Edwards syndrome. This is a rare chromosomal disorder in which all of a critical region of chromosome 18 appears three times (trisomy) rather than twice in the cells.

The disorder is characterized by an array of severe psychomotor disabilities, dysmorphic features and organ malformations, including mental disability, neurological dysfunction, growth deficiency, poor motor activity with increased muscular tonus, abnormal craniofacial profile with microcephaly and a prominent occipital bone. The subjects have a short neck, micrognathia, pectus carinatum, ocular hypertelorism, low-set and malformed ears, distinctively clenched fists with overlapping fingers, and multiple congenital cardiac defects.[2-5] Other possible clinical manifestations include spina bifida and renal, pulmonary and uterine abnormalities. Typical intraoral signs are leporine lip and a high-arched, narrow and sometimes cleft palate.[6]

The incidence of trisomy 18, which is the most common autosomal abnormality among liveborn infants after trisomy 21, is about one in 8,000 births.[4,5,7] A strong female predilection is observed by a 4:1 female-to-male ratio.[4,5,7] No racial predominance seems to exist. The prognosis is very poor: 95% of fetuses with trisomy 18 are spontaneously aborted,[8,9] 70% of liveborn infants die within the first three months, 90% die during the first year and 99% die before the age of 10.[8,9] "Long-term survivors" in this context are usually defined as patients who survive more than one year of age.[4,5] The high mortality rate is usually due to severe cardiac and renal malformations, feeding difficulties, sepsis and apnea caused by defects in the central nervous system.[5,8,10] Severe psychomotor deficits and growth retardation are invariably present for those who survive beyond infancy. As in most other syndromes, advanced maternal age is regarded as a primary risk factor for trisomy 18.[9]

Despite the number of reports addressing the clinical features and conditions involved with trisomy 18 from a medical perspective, there is little information regarding the oral findings and dental treatment of children afflicted

with this disorder, presumably due to their very low life expectancy.[6] In fact, we found only one published case report[6] which specifically described the intraoral aspects and dental care of a 7-year-old patient with trisomy 18. Therefore, the purpose of this article is to describe a "long-term survivor," a child diagnosed with pure trisomy 18.

Case report

A 13-year-old female patient with trisomy 18 was admitted to the clinics of the Center of Formation of Human Resources Specialized in Dental Care for Special Needs Patients, at the Faculty of Dentistry of Ribeirão Preto, University of São Paulo, Brazil.

The patient was the second child of healthy nonconsanguineous parents (47-year-old father and 44-year-old mother) and was born after 32 weeks of pregnancy by cesarean delivery. At birth, the child weighed 2,190g and had congenital anomalies and dysmorphic features suggestive of Edwards syndrome. Cytogenetic analysis from lymphocyte culture using GTG-banding revealed a 47,XX+18 karyotype in 100 analyzed metaphases and the child was diagnosed as having pure trisomy 18.

An overview of the patient's records showed that she was first brought to the clinic in 1994, at the age of 2. The child was breast-fed up to the age of 4 months, after which she started eating soft foods. She had delayed neuropsychomotor development, unilateral (right) cleft lip affecting the anterior third of the palate, high-arched narrow palate, low-set retroverted ears, webbed neck, pectus excavatum, first and second digits overlapping the third and the fifth digit overlapping the fourth, slightly enlarged hallux with anomalous implantation, bilateral syndactyly, hypertonia in the lower limbs and accentuated osteotendinous reflexes.

The initial phase of the treatment plan was directed at providing the parents with basic instruction in oral health care, such as information on caries, training in the mechanical removal of dental plaque and counseling on a low-sucrose diet. Dental care included professional prophylaxis, topical applications of 1.23% acidulated phosphate-fluoride gel (Sultan Topex, DFL Ind. e Com. Ltda., Petrópolis, RJ, Brazil) and sealing of the first molars' pits and fissures. Corrective surgery of the cleft lip/palate area was postponed until the age of 3 years and 3 months because of the patient's low body weight.

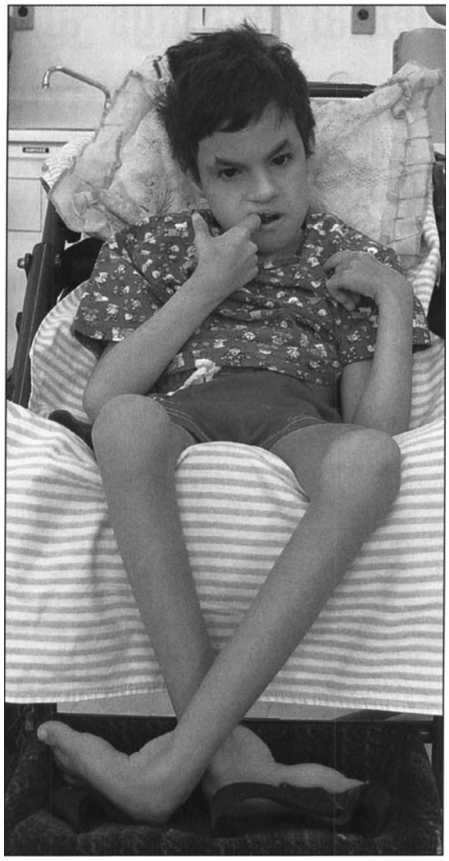

Figure 1. Clinical appearance of the 13-year-old patient.

The mother was asked to bring the child back to our service for follow-up visits and to enroll in a continuous preventive dental care program, but, regrettably, she failed to attend the scheduled visits.

Nine years later, the child was readmitted to the clinic for general dental care. She had physical characteristics typically observed in trisomy 18, such as short stature, long, thin and hyperextensible digits, hypoplastic fingernails, rocker-bottom feet with prominent heels, malformation of upper and lower limbs, joint contractures, arched and discontinuous eyebrows, and a narrowed and elongated face (Figure 1). The craniofacial findings included microcephaly, a prominent occipital region, low-set dysplastic ears, underdeveloped nose, micrognathia, and small mouth with a limited oral opening. Evaluation of the radiographic examination showed osteopenia in the pelvis and long bones. Her fingers had visible signs of self-inflicted injuries and calluses, caused by the habit of putting her hands in her mouth and biting the fingers and palms (Figure 2). Although the patient could eat semi-solid foods, she was unable to move or communicate, did not have urinary or anal sphincter control and only rarely reacted to external stimuli.

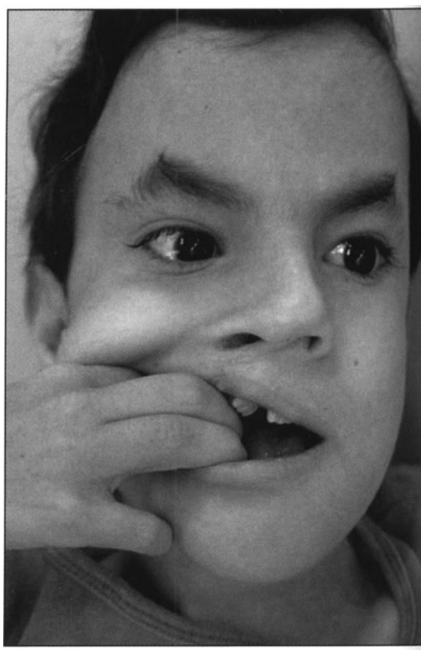

Figure 2. Patient biting her fingers. Note the arched discontinuous eyebrows.

An intraoral clinical examination revealed dental plaque deposits throughout the mouth, with gingival inflammation, multiple moderate to severe carious lesions, a deviated midline, a narrow high-arched palate, bilateral posterior crossbite, anterior crossbite and anterior open bite with the tongue positioned at the incisive papilla during rest (Figure 3). Evaluation of panoramic (Figure 4) and periapical radiographs (Figure 5) revealed agenesis of a mandibular permanent incisor, maxillary left and right premolars and mandibular

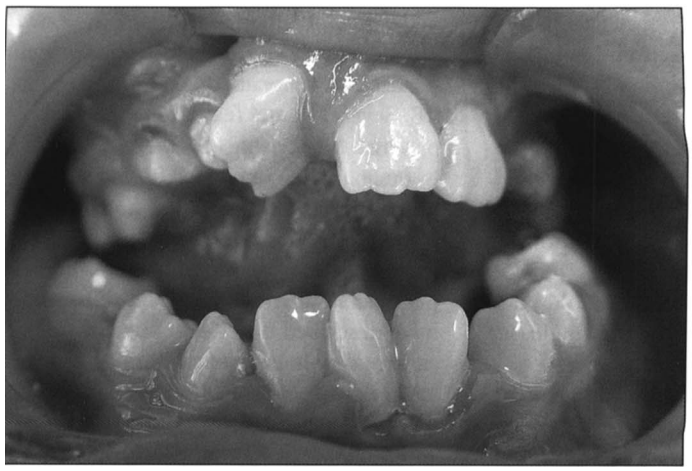

Figure 3. Intraoral frontal view showing gingivitis, deviated midline, ectopically erupted right permanent central incisor, bilateral posterior crossbite and anterior open bite with the tongue positioned at the incisive papilla during rest.

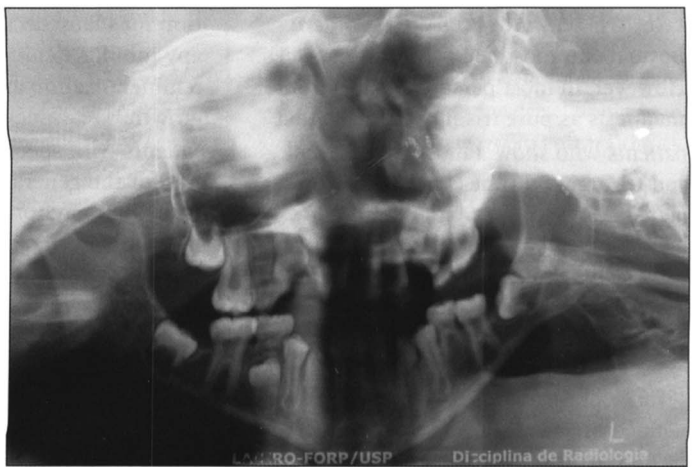

Figure 4. Panoramic radiographic image showing the presence of several dental anomalies: maxillary left and right premolars and mandibular left second premolar are missing, the permanent first molars are taurodontic, the mandibular left permanent second molar is partially impacted and horizontally displaced in the mandible.

left second premolar. All permanent first molars were taurodontic in shape with elongated coronal pulp chambers and shortened roots. The maxillary right permanent central incisor was ectopically erupted and the mandibular left permanent second molar was partially impacted and horizontally displaced in the mandible. The maxillary right permanent lateral incisor was small and conically shaped, while the maxillary left permanent lateral incisor had macrodontia. The chronology of tooth eruption was consistent with the patient's age.

A comprehensive individual-targeted program was developed based on the patient's high risk for caries. Hygiene and dietary counseling were reinforced and the parents were instructed and trained in mechanical removal of dental plaque. The child's dental condition was explained to the parents, who agreed to comply with treatment this time. We invested considerable time and effort to explain that oral homecare must support the professional care carried out in the clinic. We also stressed that the family commitment was a partnership and was essential for maintaining adequate oral health.

Dental treatment included prophylaxis, scaling and root planing of both maxillary and mandibular arches, topical applications of 0.12% chlorhexidine gluconate (Periogard; Colgate-Palmolive, New York, NY, USA) and 1.23% acidulated phosphate-fluoride gel (Sultan Topex, DFL Ind. e Com. Ltda., Petrópolis, RJ, Brazil), extraction of severely carious primary teeth (J and T), restoration of the extensively carious teeth (3, 4, 13, 19, 30, K) with composite resin, glass ionomer cement and/or amalgam and placement of sealants on teeth 14, 21 and 28.

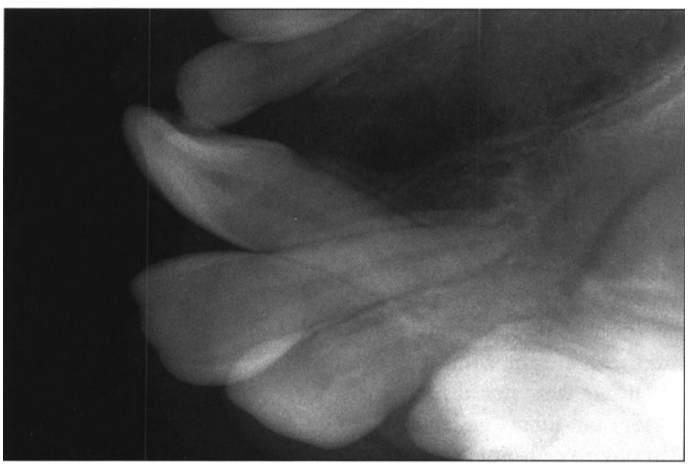

Figure 5. Periapical radiograph of the anterior region showing microdontia of tooth 7 and macrodontia of tooth 10.

The dental treatment was performed under local anesthesia at sequentially scheduled appointments in an ambulatory environment, with the patient seated in the dental chair, using pillows, foam pads and rolls for support. Neither conscious sedation nor professional physical restraint was necessary. The mother was present during the treatment sessions and assisted with the procedures, as necessary. The cavity preparation and the restorative procedures were realized under absolute isolation, with the use of a rubber dam and saliva ejector.

After four months of treatment, the patient's oral health status improved remarkably. She has attended two three-month follow-up recalls to monitor plaque and periodontal health and caries. At each recall visit, prophylaxis and topical chlorhexidine gluconate and fluoride were applied.

Permission to publish this case history was received from our local Research Ethics Committee and the child's parents.

Discussion

In some persons, trisomy 18 may be present only in a limited percentage of

cells, whereas other cells may contain normal chromosomal pairs (mosaicism). However, in most persons the syndrome manifests as pure trisomy. It seems that patients who show a mosaic of normal and trisomic cell-lines have a much better chance of surviving their childhood than those with pure trisomy.[10,11]

The child described in this paper was diagnosed as having pure trisomy 18. Therefore, considering the average life expectancy of such children and the severity of the disease,[8,9] this child can be defined as a "long-term survivor." The factors contributing to her longevity and life quality may include the absence of congenital cardiac problems or any severe systemic alterations as well as the care of her parents and supportive family.

The self-inflicted injury observed in our patient is common among children with mental disabilities.[12] The parents reported that the patient inserted her fingers into her mouth and kept "biting" or "picking" her teeth. A case report[6] in 2000 documented self-inflicted injuries that were the main cause of severe gingival recession in the primary dentition of a patient with trisomy 18. However, in our patient, there was no evidence of gingival trauma due to her finger sucking.

Hermesch *et al.*[13] reported that children with chromosome 18p deletion syndrome were not at higher risk for caries than their unaffected siblings. Although syndromic conditions do not seem to have a genetic influence on the onset of caries,[13,14] an increased risk for dental caries has been observed in affected children.[15] This is often due to frequent intake of fermentable carbohydrates (sucrose-rich, soft foods offered several times a day) combined with deficient physiological oral self-cleansing and inadequate daily oral hygiene.[13,15] In many children, there is also prolonged use of nursing bottles.[14] Therefore, to decrease caries risk, these patients need to be enrolled early in individual-targeted preventive programs. Such preventive programs should include instructions for the parents or caregivers, professional prophylaxis, use of fluorides, anti-microbial agents, and sealants.[16] Topical fluoride has been used for caries prevention for many decades[17,18] and this therapy inhibits demineralization,[18] enhances remineralization and inhibits bacterial growth.[17] Placement of restorations for patients with special needs can be difficult for the general practitioner. Access to the oral environment is often limited and patient tolerance and cooperation may be reduced to rather brief periods of time.[15] Therefore, the dentist should plan for the shortest procedures and the least clinical chair time possible to complete the treatment adequately and prevent the occurrence of any kind of accident or unexpected situation related to the medical condition.

Patients with trisomy 18 usually have an extremely short life expectancy and hence rarely have dental treatment. Dental treatment is complicated by the patients' severe intellectual deficit, motor disabilities, limited mouth opening and associated cardiac disorders. In our patient, these difficulties did not hinder treatment in an ambulatory environment unlike the two previous reports[6,14] of patients with chromosome 18 abnormalities. Their dental treatment was carried out with either conscious sedation using nitrous oxide and physical restraint[14] or general anesthesia.[6]

It has been shown[11] that 90% of patients with trisomy 18 have congenital cardiac abnormalities associated with the disease. However, our patient did not have any cardiac or systemic problems that required special consideration before or during dental treatment, which included prophylaxis, placement of sealants and restorations, scaling, root planing, and even extraction of severely damaged primary teeth. Dentists treating individuals with trisomy 18 should be aware of this condition and its potential complications, which include the risk of bacterial endocarditis and cardiorespiratory arrest. Dental treatment of individuals with cardiac disorders associated with trisomy 18 may need prophylactic antibiotic coverage. Depending on the extent of the patient's systemic problems, consultation with the attending physician is required to prevent complications prior to doing invasive procedures.

There is only one other report[6] on the dental/oral findings in a patient with autosomal abnormalities of chromosome 18. Several dental features reported in patients with trisomy 18 were observed in our patient, such as cleft lip/palate;[13,14,19] high-arched, narrow palate;[6,10,14,20] anterior open bite;[14,20] micrognathia;[5,7,10] taurodontism;[6] and vertical facial growth.[5,10,20] In addition, we have described other oral findings such as dental agenesis, macrodontia, microdontia, ectopic eruptions, bilateral posterior crossbite, anterior crossbite and midline deviation. Delayed dental eruption was not observed in our patient, which was consistent with other reports.[6,14]

The advances in science and use of aggressive medical therapies should lead to an increase in the number of long-term survivors of trisomy 18, especially if they do not have life-threatening congenital anomalies at birth.[5] These children and adolescents will require dental care along with lifelong medical assistance. It is important that dentists and dental hygienists are included within the team of health professionals caring for patients with trisomy 18 who survive infancy. Care for these children should emphasize individualized noninvasive preventive programs as early as possible.

Families should be informed that daily home care will reduce the incidence and/or severity of caries. Health professionals should discuss the importance of maintaining good oral health for patients who present with complex medical histories. Coping with the problems of parenting a child with a debilitating and life-threatening disorder is a challenging, lifelong experience. The physical disabilities, limitations and medical problems of these children are so demanding that, sometimes, oral health care is not regarded as a priority. However, health professionals must make sure the parents understand that inadequate oral hygiene can lead to dental problems that will bring additional and avoidable discomfort and pain for their children.

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