Dental Management of an Adolescent with Trisomy 13 Syndrome: A Case Report

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ABSTRACT

This first fully documented dental case report reviews the medical aspects of trisomy 13, summarizes the dental literature on this topic, and describes the dental findings and treatment of a 16-year-old female long-term survivor. (*J Dent Child*. 2004;71:91-95)

KEYWORDS: TRISOMY 13, DENTAL CASE REPORT

First described in 1960, trisomy 13 (Patau) syndrome is an autosomal genetic disorder caused by an extra chromosome of the D_1 group.[1,2] Regular trisomy 13 accounts for the majority of cases, while translocations occur in 20% and mosaics are noted in 5%.[3,4] These chromosomal patterns are associated with differences in clinical presentation of the syndrome or phenotype. The prevalence for the most commonly inherited form is 1 in 12,000.[4-6] Approximately 400 live births occur annually in the United States, with a slight excess of females.[7] The mean maternal and paternal age from 3 large studies is 31.8 and 34.1 years, respectively, and, thus, is 5 years higher than for normal births.[5,6,8]

Life expectancy in regular and translocation trisomy 13 patients is generally low, with a median survival rate of less than 3 months. Approximately 38% live beyond their first year, and only 3% reach age 10.[6,8] However, isolated cases of survival into adolescence and early adulthood have been reported[6,9,10] with the oldest being 32 years of age.[11] The most common causes of death are cardiopulmonary arrest (69%), congenital heart disease (13%), and pneumonia (4%).[6] Because the prognosis for long-term survival is poor, corrective surgery is initially withheld awaiting the outcome of the first few months of life and then only performed if the individual has a reasonable chance for longer-term survival.[12]

Trisomy 13 syndrome is associated with multiple and severe congenital anomalies. Its principal features, summarized in Table 1, include variable degrees of holoprosencephaly, with anatomic defects of the brain resulting in seizures and apneic spells, cardiac malformations, cleft lip and cleft palate, eye and ear defects, abnormal genitalia, and polydactyly.[1,2,8,12]

Not all individuals with trisomy 13 survive in a vegetative state only. Documented higher psychomotor achievements include crawling, use of a walker, or walking independently, albeit considerably delayed. Some older children are able to smile responsively, recognize close adults, drink from a cup, finger-feed themselves, and understand words and phrases.[16]

Orofacial aspects of the disorder, summarized from 2 case reports[12,17] describing 5 preschool children, include delayed eruption, over-retention and/or ankylosis of primary teeth and bilateral cleft lip/palate. Another article[18] reported, in a very detailed manner, variations in dental crown morphology and noted absence of the uvula, ankyloglossia, and clefting. An analysis of 175 trisomy 13 patients[8] yielded a higher incidence of all types of clefts in regular (64%) and translocation (67%) cases, as compared to the mosaic (35%) and partial (22%) types. A high-arched palate occurred in 9% of the patients only.

This article is the first fully documented dental case report, including intraoral photographs of the permanent dentition, of a long-term survivor with trisomy 13 syndrome.

CASE REPORT

The patient, the second child of a 26-year-old mother and 29-year-old father, was born at 40 weeks gestation after an uncomplicated pregnancy. Her 2 brothers are normal. There are no known chromosomal abnormalities in their extended family. At birth, the baby was noted to have mild dysmorphic features, including aplasia cutis. Based on this, her pediatrician suspected trisomy 13, a diagnosis which was later confirmed by chromosomal analysis. The karyotype was straight trisomy 13 with no mosaicism. Recurrent respiratory infections and apneic episodes during her first year of life required multiple hospitalizations, but their frequency decreased thereafter. The most recent hospitalizations were at age

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Table 1. Common Malformations in Trisomy 13[1,4]	
CNS, cranium	Holoprosencephaly with incomplete development of forebrain, olfactory, and optic nerves; seizures, apneic spells, moderate microcephaly, severe developmental retardation
Eyes	Colombomas of iris, retinal dysplasia, micropthalmia, hypo- or hyperteleorism
Ear	Deafness, abnormal auricles, and low set ears
Craniofacial features	Lateral or median cleft lip, cleft palate, or both; aplasia of premaxilla; sequelae of holoprosencephaly: cyclopia, cebocephaly, hypoplasia or aplasia of nose, micrognathia, sloping forehead, prominent nasal bridge
Cardiac	ASD, VSD, patent ductus arteriosus, dextroposition
Extremities	Simian crease, polydactyly of hands and sometimes feet, hyperconvex, narrow fingernails, club foot
Renal anomalies, genitalia	Polycystic kidneys Male: cryptorchidism, abnormal scrotum Female: bicornuate uterus
Neck	Short neck, loose skin on the nape, aplasia cutis

6 for pneumonia, and 3 months after this dental treatment in the operating room for an infected kidney cyst and pleural effusion.

The 16-year-old white female patient, weighing 25 kg and measuring 132 cm, is severely mentally retarded and very delayed in both mental and motor development. Although she has a 75% hearing loss, she is vocal, but nonverbal. In addition, she is legally blind as a result of bilateral colombomas, cataracts, and abnormalities of her optic nerves. However, she was responsive to the examiner. At home, she crawls around the house and has had fractures of both clavicles and femur, all from falls. She finger-feeds herself and drinks with assistance from a cup. Although she suffers from pronounced gastroesophageal reflux (GER), her parents continue to deny placement of a gastrojejunostomy tube, one treatment of GER disease. Her heart is at the upper limits of normal with no murmurs. Menarche has not yet occurred.

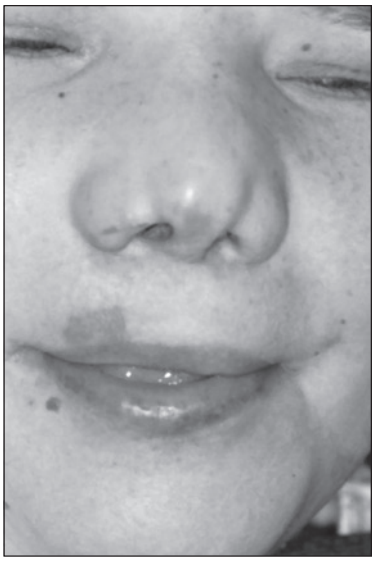

Figure 1. Photograph of midface.

Due to multiple other health problems and repeated concerns of physicians that the patient had a statistically low life expectancy, dental care was not aggressively pursued. Her primary care physician referred the patient to us because he feared that the negative effects of the chronic infections, emanating from her multiple severely decayed teeth, would have a negative impact on her overall health.

The initial extraoral exam revealed some of the facial features of trisomy 13:

1. micropthalmia;
2. low-set ears;
3. capillary hemangioma in an atypical location on the upper lip;
4. retrognathic mandible.

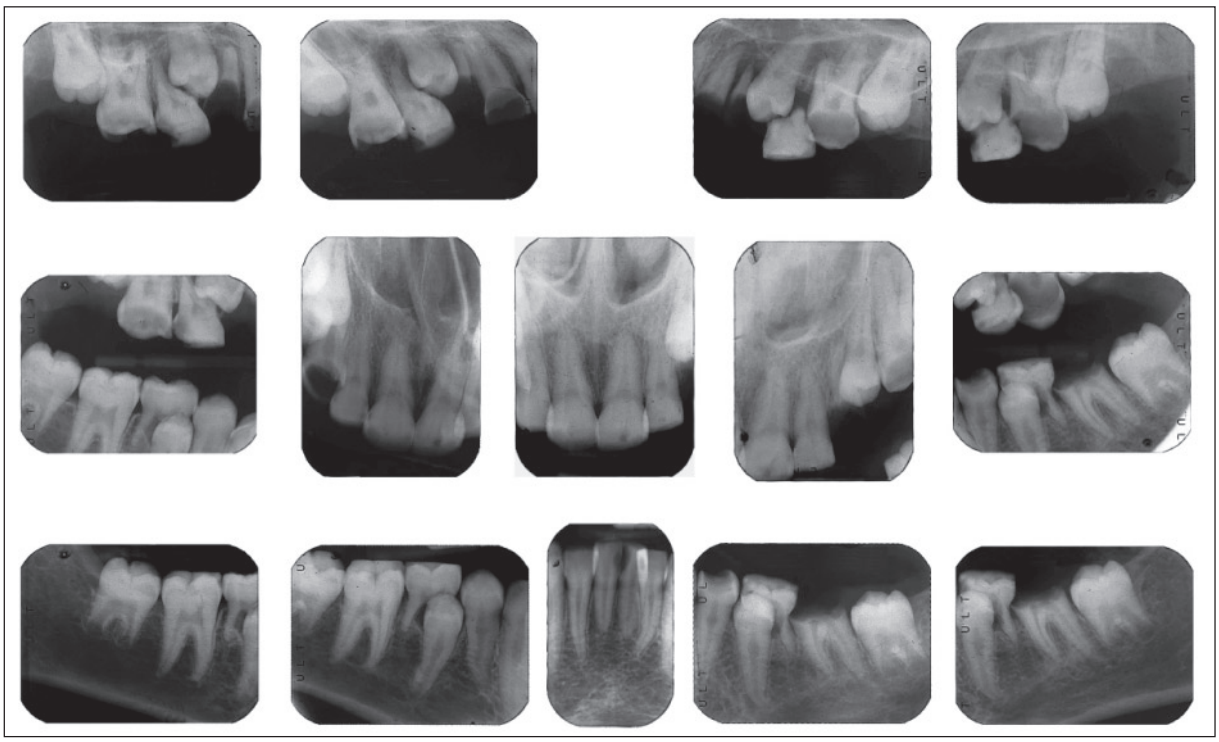

Figure 2. Full-mouth series radiographs.

Her nose had a bilateral notch near the nares, and the nares themselves were small (Figure 1). Intraorally (Figures 2 and 3), the patient showed a delayed Cl. II_1 mixed dentition (6 mm overjet, 0% overbite, unilateral posterior crossbite) with agenesis of both maxillary canines, first premolars, and third molars. All 4 primary second molars exhibited prolonged retention with intact tooth morphology, whereas the other retained primary teeth presented as severely destroyed crown (C) or root tips only (B, H, I). Multiple permanent teeth were decayed (teeth 3, 7, 8, 9, 14, 19, 21, 24, 25, 30, and 31), but of normal size, color, and shape. Teeth 2, 4, 13, 15, 18, 20, and 29 were impacted, and the maxillary permanent second molars had erupted ectopically. Bruxism caused considerable attrition of the incisal surfaces of all her anterior teeth, while enamel erosion due to her GER was mild. Moderate to severe plaque accumulation on all of her teeth was responsible for generalized gingivitis.

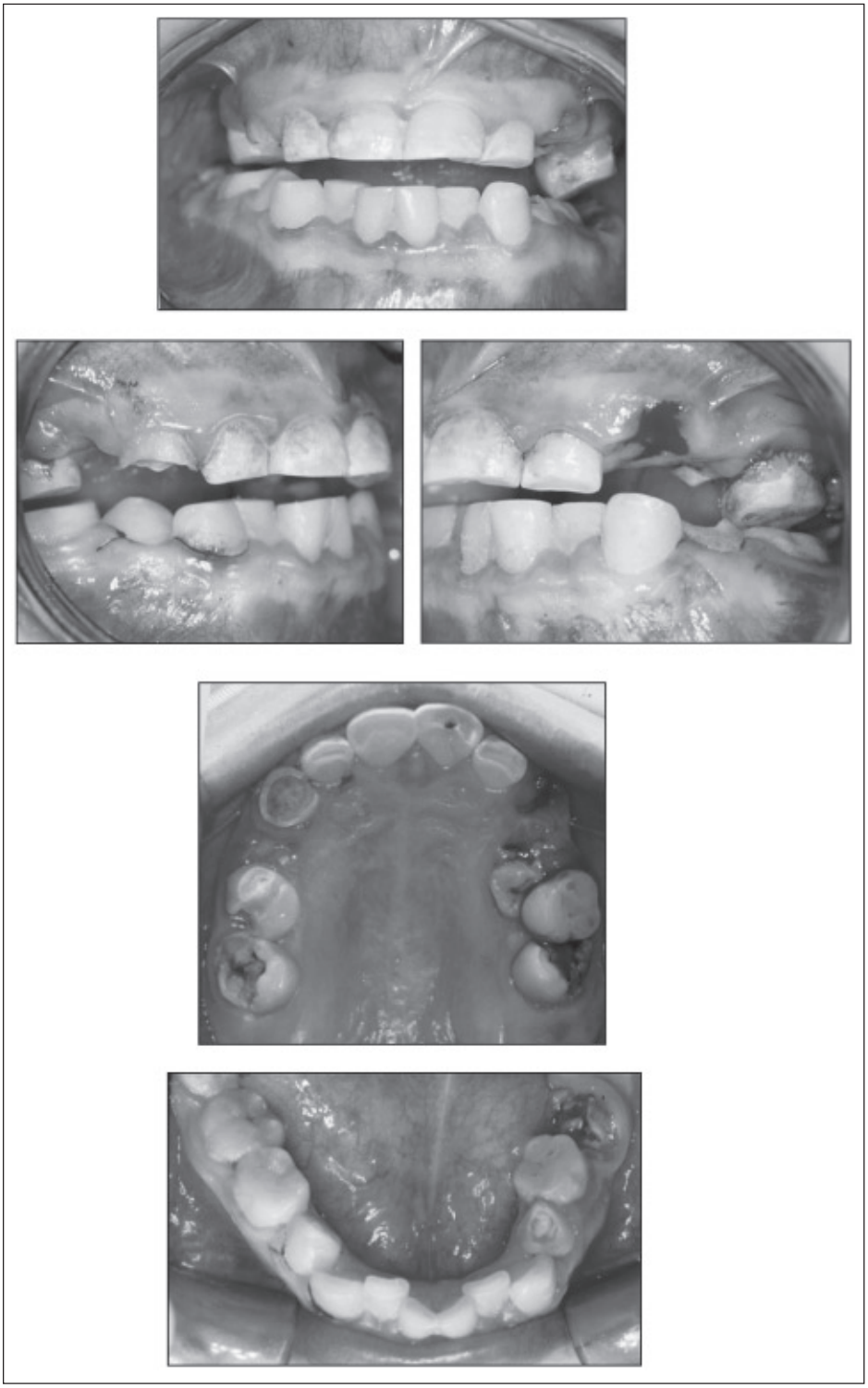

Figure 3. Preoperative intraoral photographs.

Due to her medical diagnoses, the necessity of complex dental treatment, and her inability to cooperate in the traditional dental setting, it was decided to treat her in the operating room under general anesthesia.

TREATMENT

Nonrestorable permanent (teeth 3, 14, and 19) and retained primary teeth/root tips (A, B, H, I, J, K, T) were extracted, and amalgam or composite restorations were placed in remaining carious teeth (teeth 7, 8, 9, 21, 24, 25, 30, and 31). At the postoperative visit a few weeks later, it was noted that her oral hygiene had improved. The restorations placed were intact, all extraction sites had healed well, and the cusp tips of teeth 4, 13, 20, and 29 were visible on the alveolar ridges.

When the patient was seen for her recall appointment 6 months later, these second premolars as well as teeth 2, 15, and 18 were fully erupted. With some coaxing and mild parental restraint, the exam could be performed in the traditional dental setting, and the authors were able to take a maxillary intraoral mirror photograph (Figure 4).

DISCUSSION

Long-term survival of individuals with trisomy 13 syndrome is rare and dependent on the severity of the malformations. In the first few months of life, treatment consists mainly of conservative management of the congenital anomalies. In general, surgeons postpone procedures until 12 months of age, awaiting hope for a longer life,[12] while some address life-threatening malformations earlier to increase the chances for survival. Case reports describe

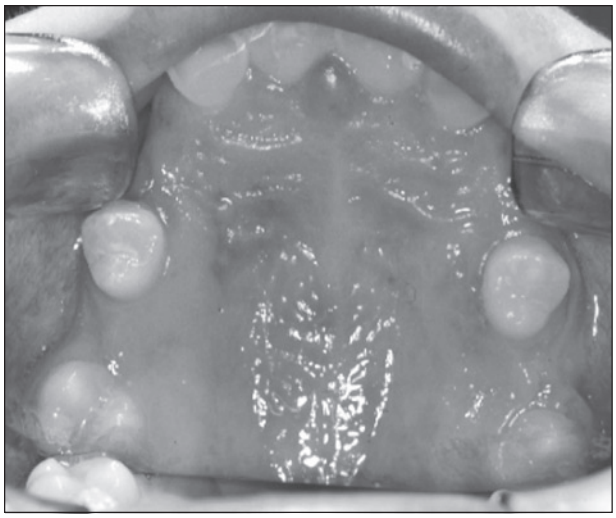

Figure 4. Maxillary intraoral photograph at recall appointment.

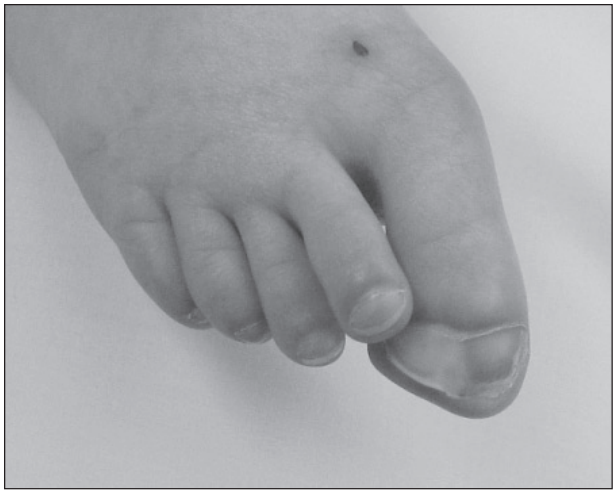

Figure 6. Photograph of right big toe.

successful anesthesia for cardiac surgery at 3 months and 5 years of age,[13,14] as well as cleft lip/-palate repair at 8 months.[15]

Because of the complexity of the patient's medical problems, dental care was not a high priority for her caregivers. She had many features characteristic of trisomy 13: Micropthalmia (Figure 1), bilateral iris colombomas, hearing loss, thoracic kyphoscoliosis, short neck, polycystic kidneys, hyperconvex fingernails (Figure 5), apneic episodes, and seizure disorder. The absence of heart defects may have contributed to her long survival. Her right big toe was broad and deeply notched in the midline (Figure 6), a finding that can be interpreted as an incomplete polydactyly. Notches on both sides of her nose indicated a disturbance during nose formation.

Cleft lip and/or palate are the most commonly reported oral anomalies,[1,8] followed by micrognathia and a high-arched palate.[8,14,15] In contrast, the authors' patient had a flat palate, a bifid uvula but no oral clefting, no ankyloglossia or abnormal freni and a delayed tooth eruption pattern. Her mandible was not parti-cularly small and was rather retrognathic, a finding for which a supportive literature account exists.[18] It is conceivable that retrognathic mandibles were by some researchers misinterpreted to be smaller (micrognathic), when in fact they were only more retropositioned. Sometimes a rudimentary or nonexistent premaxilla makes bilateral clefts of the lip and palate appear as one wide median cleft only.[12]

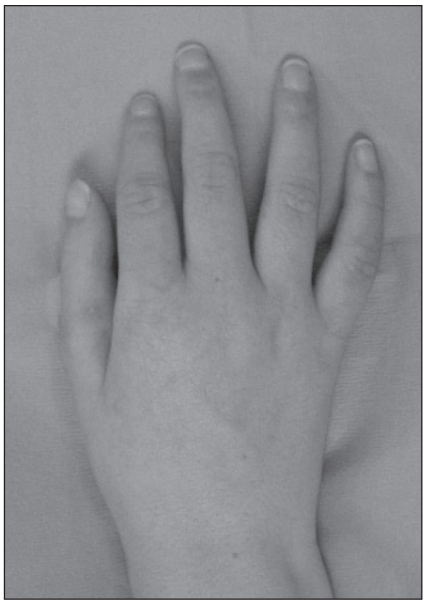

Figure 5. Photograph of right hand.

Higher parental age as an etiologic factor is unlikely, as the age of either parent equaled the mean age for all births in the United States' population and 2 healthy brothers support findings of a low sibling recurrence risk of 0.55%.[6] Approximately one fifth of all patients with trisomy 13 have a gastrojejunostomy tube,[6] but this patient is able to finger-feed herself and drink from a learning cup. Although her GER is only poorly controlled with Prilosec 30 mg/day, her tooth enamel is not significantly affected by acid erosion. Her seizures are so infrequent that she does not take seizure control medication.

Our patient belongs to a small group of individuals with higher psychomotor achievements[16]: she responds to external stimuli, recognizes and interacts with close family members, understands words and short phrases, crawls around the house, walks with support, drinks from a learning cup, and finger-feeds herself with assistance. She likes to curl up in the fetal position.

Agenesis of both permanent maxillary canines and first premolars and a small notch on either side of the nose near the nares indicate a disturbance in the area where the palatal shelves fuse with the premaxilla. Both these findings and the bifid uvula are microsymptoms of clefting. Findings of distinct anomalies in crown morphology[18] could not be verified in the authors' patient's existing dentition.

General anesthesia of patients with trisomy 13 is often difficult due to cardiac malformations and other congenital anomalies. Reduced lung volume due to spine and chest anomalies, combined with retrognathia, results in a potentially difficult airway.[14,15] GER presents a significant risk for aspiration. Despite a short neck, kyphoscoliosis, and retrognathia, intubation of our patient was easily accomplished.

CONCLUSIONS

Only a small fraction of individuals with trisomy 13 reach an age when primary teeth erupt. Survival until the mixed and/or permanent dentition is extremely rare. Patients without oral clefting are less likely to be seen by a dentist because physicians and parenting adults focus preferentially on multiple

and severe congenital anomalies and developmental problems. Nevertheless, regular dental visits from an early age on should be encouraged to prevent extensive dental disease.

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