

Dental abnormalities associated with campomelic syndrome: case report

Gordon L. Roberts, DDS Soraya Beiraghi, DDS, MS, MSD

Abstract

Campomelic syndrome is a rare autosomal recessive disease. It is characterized by short stature with angulation and bowing of the lower limbs, hypoplasia of the facial bones, and various other skeletal anomalies. The facies are unusually flat with micrognathia, frequent cleft palate, hypertelorism, and microphthalmia. Most infants with this syndrome die from severe respiratory distress within the first hours or days of life. Those who survive infancy are mentally retarded. This case report concerns the dental treatment of a 13-year-old female with campomelic syndrome.

Literature Review

The campomelic syndrome was named in 1971 by P. Maroteaux (Maroteaux et al. 1971), but earlier, similar cases had been described (Bound et al. 1952). The condition has been reported around the world in a variety of ethnic groups and appears to be caused by an autosomal recessive gene (Beluffi and Fraccaro 1982). The syndrome occurs sporadically with a definite preponderance of female cases.

The term campomelic means "curved extremities" and the syndrome has characteristic radiographic features that include bowing of the tibia, fibia, and the femur, and multiple anomalies of the vertebrae and ribs (Pazzaglia and Beluffi 1987). Other major manifestations include dwarfism, craniofacial anomalies, and severe respiratory distress (Houston et al. 1983). The respiratory distress is caused by pulmonary hypoplasia complicated by deformities of the tracheal cartilage (resulting in a narrow tracheal lumen) and in the ossification of the sternum and ribs (Hall and Spranger 1980; Houston et al. 1983). Posterior positioning of the tongue, possibly due to the micrognathia, also has been noted. The respiratory distress frequently is accompanied by pulmonary aspiration leading to death (Tokita et al. 1979).

Approximately 25% of the cases have a congenital heart defect, usually a patent ductus arteriosus, although

other defects are not uncommon (Hall and Spranger 1980; Houston et al. 1983).

The craniofacial anomalies consist of macrocephaly and dolichocephaly. The facies are flat with a prominent forehead and hypertelorism. The nasal bridge is broad and flat and the nose tends to be small with anteverted nostrils. There are frequent cleft palates along with micrognathia (Houston et al. 1983). Radiographically the skull may demonstrate a flattening of the sella turcica and the orbits. Strabismus also has been reported. Deformities of both the middle and inner ears are common, involving the ossicles of the middle ear and the cochlea of the inner ear (Tokita et al. 1979).

The syndrome has an extremely high mortality rate as reported in the literature. Of the 92 cases reviewed up to 1982, 45 died within the first 24 hr and 44 died prior to 10 months of age (Beluffi and Fraccaro 1982). The 3 reported survivors ranged in age from 7 months to 17 years. In almost all cases, death was due to respiratory failure (Houston et al. 1983).

Another unusual aspect of this syndrome is the observed sex ratio. In one series of 97 affected patients, the clinically determined sex ratio was 2.2 females to 1 male. However when karyotypes were carried out on 50 of these patients (38 females, 12 males) it was found that 21 female patients had a typically male genotype of 46XY (Houston et al. 1983). Among the reported females are a substantial number with a male karyotype and absence of the HY antigen (Bricarelli et al. 1971). Thus, if the patient's sex is determined genetically instead of clinically, the sex ratio appears to be reversed.

Case Report

A 13-year-old white female was referred to the Columbus Children's Hospital dental clinic for evaluation and treatment of her dental condition. Because she was adopted, no accurate family history was available. The child was mentally delayed. She had the characteristic

facial features of campomelia including a dolicocephalic face with a broad, flat nasal bridge. She needed glasses and wore a hearing aid in both ears. There was no apparent involvement of the internal organs. The most striking feature was the patient's size — 3 feet, 11 inches tall and 56 pounds (normal height is ~ 5 feet and normal weight is ~ 91 pounds; Fig 1). As a young child she had some deformation of her lower limbs; this improved with age and she could walk unassisted, though with some difficulty. The original diagnosis of campomelia was made at Johns Hopkins University after a genetic evaluation in 1976.

There were several interesting dental findings. The child was still in early mixed dentition, having lost the permanent maxillary and mandibular central and lateral incisors and the mandibular first permanent molars which had both been exposed surgically. All of the primary molars were present except the mandibular left first primary molar which had exfoliated. A high arched palate was noted, but there was no frank clefting. The child appeared to have a severe form of amelogenesis imperfecta with the permanent teeth affected more than the primary (Figs 2-4).

The enamel of the permanent teeth was light brown in color with a rough surface texture. The permanent teeth were small and the mandibular permanent molars had a flat occlusal anatomy. Clinical examination revealed hard enamel which could not be penetrated with an explorer. Radiographically the child had a slightly prognathic maxilla with a very steep mandibular plane. Her parents said that when her primary molars erupted, she had an anterior open bite, which decreased due to severe attrition of the posterior teeth. All teeth had



FIG 2. Frontal view showing the hypoplastic and pitted surfaces of the enamel of the permanent incisors. Note also the small size of the permanent teeth and the severe gingival overgrowth.

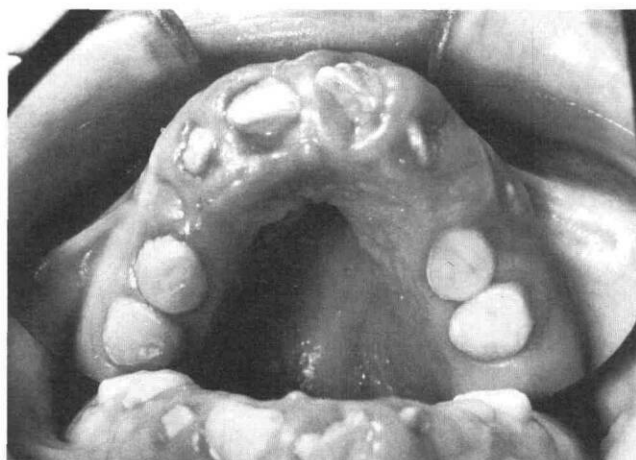


FIG 3. Maxillary occlusal view showing the thin enamel and poor anatomic contours of the primary posterior teeth.

enamel that appeared thin radiographically with a density similar to the dentin.

Although the eruption of the permanent teeth was delayed severely, root formation appeared to progress normally. Both the first and second mandibular permanent molars had essentially complete root development. However, the bone superior to the crypts of the second molars had resorbed, even though they were still deep within the body of the mandible and the teeth did not appear to have moved occlusally. Thus, there appeared to be a disturbance in the eruption process (Fig 5).

Treatment

Since the major concern of the parents was esthetics, it was decided to restore the permanent incisors with composite resin restorations. Before restoration of the teeth, it was necessary to alleviate the gingival hyper-



FIG 1. Full-body view showing the patient's bowed legs as well as evidence of both macrocephaly and a broad nasal bridge.

The enamel of the permanent teeth was light brown in color with a rough surface texture. The permanent teeth were small and the mandibular permanent molars had a flat occlusal anatomy. Clinical examination revealed hard enamel which could not be penetrated with an explorer.

Radiographically the child had a slightly prognathic maxilla with a very steep mandibular plane. Her parents said that when her primary molars erupted, she had an anterior open bite, which decreased due to severe attrition of the posterior teeth. All teeth had

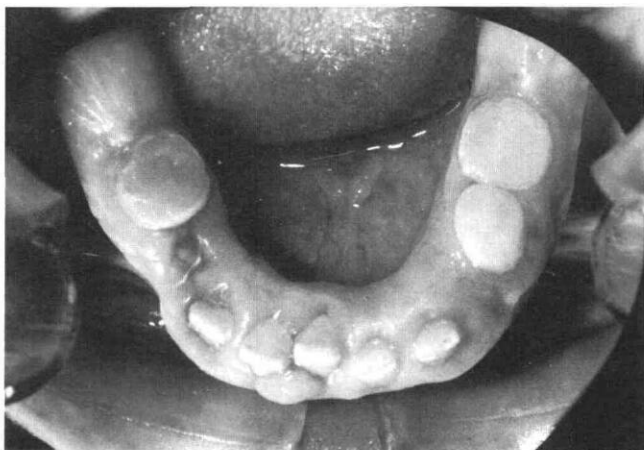


FIG 4. Mandibular occlusal view showing the delayed eruption of the permanent first molars and the poor anatomy of the primary molars.

plasia that had occurred in the anterior segments. One month after the surgical procedure there was still an excess of fibrous tissue covering the permanent incisors. A reverse bevel gingivectomy was performed around the maxillary incisors to expose the crowns fully. After initial healing occurred (2 weeks), the teeth were restored with composite resin restorations. This procedure improved their contours as well as color and surface texture. The mandibular incisor region was treated with a reverse-bevel gingivectomy and the tissue healed uneventfully.

Treatment of the posterior dentition to date has been limited to surgical exposure of the mandibular left and right first permanent molars to encourage eruption of these teeth. Both teeth were small with the same rough enamel exhibited by the permanent incisors. Since the primary molars had begun to exfoliate, it was decided to wait and see if the premolars would erupt. Orthodontic therapy to force the permanent molars to erupt is not being considered at the present time.

Discussion

A number of unique features were observed in a patient afflicted with a syndrome that is usually fatal in infancy. Consequently, there is almost no information in the literature concerning the condition of the orofacial structures or any aspects of dental treatment for such patients.

One of the most striking features of this child's dentition is the abnormal appearance of permanent tooth enamel that demonstrated one of the forms of amelogenesis imperfecta. In the general population, 1:14,000 are affected with amelogenesis imperfecta and it also has been associated frequently with other hereditary syndromes that have more generalized defects (Witkop and Sauk 1976). Many of these syndromes are

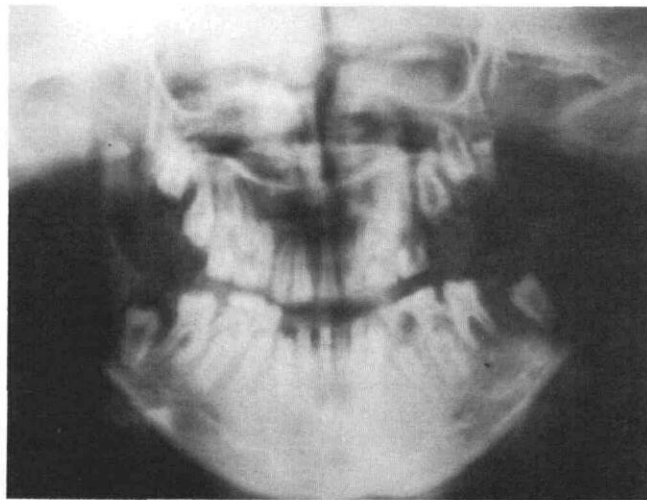


FIG 5. Panoramic radiograph showing the completed root formation of the permanent molars even though they have not erupted. Note bone resorption superior to the mandibular permanent second molars

extremely rare, as in this case, and it is difficult to determine whether the enamel defect is actually a component of the larger syndrome or a separately inherited condition.

Mucopolysaccharidosis (MP) is one of the syndromes whose features are similar to those of campomelia. These patients also suffer from dwarfism and, in type IV, also exhibit a short, broad nose and hearing loss. All types of MP are caused by an autosomal recessive gene with the exception of type II, (Witkop and Sauk 1976). The enamel is affected somewhat differently than in the present case. In MP both the primary and the permanent teeth are affected, resulting in a thin enamel of normal hardness. The enamel is often pitted or covered with a vertical pattern of dimpling, especially on the buccal surfaces (Witkop and Sauk 1976). In contrast, this patient had a more generalized, granular enamel that was more noticeable in the secondary dentition. Also, the tooth eruption was delayed, whereas this is not true of the MP-type syndromes.

The appearance of the teeth of the patient in our report most resemble closely the description of autosomal recessive rough amelogenesis imperfecta. In this condition newly erupted teeth have a rough, granular surface and are yellow, while there are multiple unerupted, impacted teeth (Witkop and Sauk 1976). However, in contrast to our case report both dentitions are equally affected, there is almost no enamel present on any of the teeth, and the unerupted teeth seemed to undergo resorption in their crypts (Witkop and Sauk 1976).

The patient in our report had facial features and dental findings that do not correlate well with the de-

scriptions of the various amelogenesis imperfecta type disorders. It must be reasoned, then, that the error in amelogenesis is linked directly in some way to the more generalized syndrome with which this patient is afflicted.

The treatment of this patient will have to proceed slowly and will vary depending on her growth and development. Currently, treatment is oriented toward encouraging eruption without orthodontic intervention. If the permanent posterior teeth erupt fully, then she will have to be followed closely since her thin, rough enamel puts her at greater risk for caries (due to increased plaque retention) and abrasion (due to the thin enamel).

Dr. Roberts is a second-year resident and Dr. Beiraghi is an assistant professor, pediatric dentistry, The Ohio State University. Reprint requests should be sent to: Dr. Soraya Beiraghi, 3001-C Postle Hall, College of Dentistry, The Ohio State University, 305 W. 12th Ave., Columbus, OH 43210.

Beluffi G, Fraccaro M: Genetical and clinical aspects of campomelic dysplasia. *Skeletal Dysplasias*. New York; Alan R. Liss Inc 1982 pp 53-68.

Bound JP, Findlay HVL, Rose FC: Congenital anterior angulation of the tibia. *Arch Dis Child* 27:179, 1952.

Bricarelli FD, Fraccaro M, Lindsten J: Sex-reversed XY female with campomelic dysplasia are XY negative. *Human Genet* 57:15, 1981.

Hall BD, Spranger JW: Campomelic dysplasia. *Am J Dis Child* 134:285-89, 1980.

Houston CS, Opitz JM, Spranger JW, Macpherson RI, Reed MH, Gilbert EF, Herrmann J, Schinzel A: The campomelic syndrome: review, report of 17 cases, and follow-up on the currently 17-year-old boy first reported by Maroteaux et al. in 1971. *Am J Med Genet* 15:3-28, 1983.

Maroteaux P, Spranger J, Opitz JM, Kucera J, Lowry RB, Schimke RN, Kagan SM: Le syndrome campomelique. *Presse Med* 79:1157, 1971.

Pazzaglia UE, Beluffi G: Radiology and histopathology of the bent limbs in campomelic dysplasia: implications in the aetiology of the disease and review of theories. *Pediatr Radiol* 17:50-55, 1987.

Tokita N, Chandra-Sekhar HK, Daly JF, Becker MH, Aleksic S: The campomelic syndrome. *Arch Otolaryngol* 105:449-54, 1979.

Witkop CJ, Sauk JJ: Heritable defects of enamel, in *Oral Facial Genetics*, Stewart RE, Prescott GH, eds. St Louis; CV Mosby Co, 1976 chapter 7.

Origins of the Aesculapius staff

Thanks to *The Aesculapiad*, the official publication of the New Mexico AGD, for explaining the origins of the use of the staff of Aesculapius by dentistry.

"From 1200 BC to 500 AD, Greeks were developing medical techniques similar to those in use today. The secrecy of the past was replaced by thought and discussion, resulting in more rational medicine.

"Greek mythology played a part in the beginnings of medicine: Apollo married Coronis and had many children. One child Asklepios (Aesculapius in Roman), was taught by Chiron, a centaur. Hygieia (personification of health) was the daughter of Aesculapius, Panacea (personification of the universal remedy) was a daughter of Aesculapius who, with Hygieia, assisted in the temple rites and tended the sacred serpents.

In Greece there temples and sanctuaries for healing the sick that were dedicated to Aesculapius. Prayers, massage, bathing and "temple sleep" (ataraxia—a tranquilizer) were methods they used for healing. A sacred snake would "lick" disease and help with the cure (psychosomatic healing).

The snake, *Coluber longissimus* (Aesculapius), was nonpoisonous with a yellow band. The book of Numbers in the *Bible* formed a basis for the staff by stating, "Make thee a fiery serpent, and set it upon a pole," thus evolving the staff of Aesculapius.