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Cohen syndrome: case report

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Abstract

A report of a child with Cohen syndrome is presented. Of particular interest to dentists are the relatively consistent findings of open mouth, short philtrum, micrognathia, and the prominent maxillary central incisors. A combination of these findings in addition to other traits such as hypotonicity, variable degrees of mental retardation, narrow hands and feet, childhood obesity, and delayed puberty, should raise the dentist's suspicion of this or related syndromes.

Cohen syndrome first was described as a unique clinical entity in 3 patients in 1973.[1] That report included histories of 3 Caucasian children of full-term gestation with decreased fetal activity and with apparently normal parents. Two of the 3 children were siblings, and the third case was an isolated occurrence in an unrelated family.

In reviewing the literature, 28 other reports[2-12] were found. The most consistent findings include obesity, mental retardation, hypotonia, maxillary hypoplasia, short philtrum, open mouth, micrognathia, and narrow hands and feet. Less frequently observed characteristics include short stature, microcephaly, antimongoloid slanting, and prominent central incisors. Although the syndrome originally was suggested to involve an autosomal recessive pattern,[1] recent analysis strongly supports an autosomal dominance process.[13]

In this report, the authors describe an individual with documented Cohen syndrome.

Case Report

A 10-year-old Caucasian female (Fig 1) was seen at the University of Louisville School of Dentistry for evaluation of malocclusion and possible orthodontic treatment. She was the product of a full-term uncomplicated pregnancy. The mother was 23 years old and $G_3P_2A_0$ (gravida 3 - third pregnancy; para 2 - two previous deliveries; and aborta 0 - no abortions) at the time of delivery. The mother smoked 1 pack of cigarettes daily and consumed alcoholic beverages occasionally. The father was 25 years old and unrelated to the mother. The couple has 2 other healthy daughters, 14 and 12 years of age. At birth, there was a nuchal cord and the infant required resuscitation in the delivery room. Birth weight was 5 lb, 9 oz, length was 21 in, no special care was given in the nursery, and she was discharged at 6 days of age. The child slept a great deal during infancy and had no feeding difficulties. She sat unsupported at 8 months, walked unsupported at 13 months, and began to use single-word utterances at 18 months. She was using short phrases by age 3 and was toilet trained at about the same time. Intellectual testing was performed utilizing the Wechsler Intelligence Scale for Children-Revised at ages 6 and 7 and revealed an intelligence quotient of 72 and 74, respectively. She has worn ventilation tubes since the age of 2 for recurrent otitis media.

Physical examination showed a shy 10-year-old Caucasian female with: occiptal-frontal circumference — 51.4 cm (30th percentile), height — 60 1/4 in (95%), and weight — 104 lb (85%). She had an elongated face with high nasal bridge, maxillary hypoplasia, mandibular micrognathia, and downslanting palpebral fissures. Facial asymmetry was noted. Pupils were equally reactive to light and accommodation of ocular muscles was intact. Funduscopic examination was normal. The ears were lowset and prominent; the nose was beaked with hypoplastic alae and milia; and the philtrum was normal except for small nevous flameous. There was a high arched palate and promi-

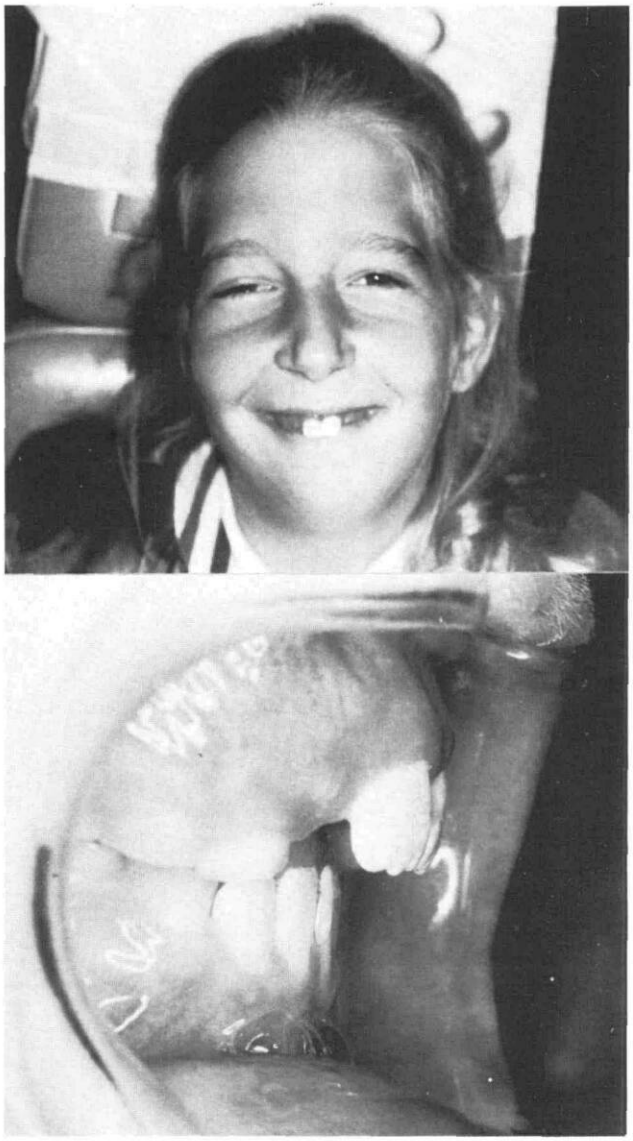

FIG 1. *(top)* A child with Cohen syndrome; note the prominent central incisors.
FIG 2. *(bottom)* Intraoral view of anterior teeth revealing large overjet, minimal overbite, and Class II relationship.

nent maxillary central incisors. The neck was supple with no lymphodenopathy, lungs were clear, and no heart murmur was detected. There was no breast development. The child demonstrated mild obesity with no organomegaly. Peripheral pulses were intact and examination of the genitalia revealed a few straight pubic hairs. The hands were long and narrow, fingers were elongated, but not tapered, and there was joint hyperextensibility. The toes were elongated with hypoplastic toenails, and the feet were long and narrow. There was an exaggerated lumbarlordosis.

Neurologic examination revealed intact cranial nerves, symmetrical tendon reflexes 2+, and flexor plantar responses with no clonus. Superficial reflexes were intact. Muscular hypotonia and weakness were present. Chromosome analysis showed a 46,XX constitution. In view of the above findings, the diagnosis of Cohen syndrome was made.

Examination of other family members revealed that the father, one of his sisters, and a niece all had clinical signs suggestive of Cohen syndrome.

The dental history was not significant. There was no history of oral trauma or toothaches, and the patient was seen previously by another dentist. The following teeth were present: 3, A, B, C, 8, 9, H, I, J, 14, 19, K, L, N, O, P, Q, S, T, 30. Occlusal caries were present in teeth A and 14. There was mild generalized gingivitis in the anterior portion of both arches and mild localized gingivitis in the posterior portion of both arches. The maxillary central incisors were prominent (Fig 2). The permanent molar relationship was Class II. There was no primate or incisor spacing, and the overbite and overjet were 5% and 8 mm, respectively. There was a mild posterior crossbite on the left side. Bilateral "popping" of the TMJ could be detected during maximal opening and closing sequences, and there was some occlusal wear associated with mild bruxism. A herpetic lesion was noted in the commissure area of the lips. A cephalometric radiograph and subsequent analysis revealed a flattening of the cranial base, small mandible, shortening of the anterior cranial fossa, short posterior facial height, and hypoplastic maxillary bone. The patient was classified as a shy, but cooperative dental patient.

Discussion

The pediatric dentist has the opportunity of being one of the first medical-health personnel to observe children who manifest uncommon syndromes or anomalies. Therefore, it is important that they be astute diagnostic clinicians and familiar with these unusual cases. The authors recently observed two unrelated children with Cohen syndrome one of whom is reported. Of particular interest to dentists are the relatively consistent findings of open mouth, short philtrum, micrognathia, and the prominent maxillary central incisors. A combination of these findings, in addition to other traits such as hypotonicity, variable degrees of mental retardation, narrow hands and feet, childhood obesity, and delayed puberty, should raise the dentist's suspicion of this or related syndromes.

The patient in this report did not exhibit any unusual caries pattern for children of her age and this geographic region. Since the patient was relatively cooperative, restorative treatment was provided with patience, and standard tell-show-do behavioral modification techniques. Sedatives and restraints were unnecessary; however, the patient was sensitive to many dental stimuli. The patient's psychomotor skills

were not well refined for her age. The parents also were informed of the importance of establishing a good oral hygiene program at home since patients with Cohen syndrome exhibit variable degrees of mental retardation. The child has been referred to an orthodontist for treatment of the malocclusion.

Often, related syndromes have several characteristics in common, as is true of Cohen syndrome. Patients with this syndrome present with obesity, mental retardation, limb and genital anomalies, all of which are also found in the Prader-Willi and the Lawrence-Moon-Biedel syndromes. However, it has been suggested that these other disorders can be distinguished easily from Cohen syndrome based on differences in characteristic craniofacies, limb findings, and the timing and character of obesity.[3]

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Quotable Quote: xerophthalmia

Xerophthalmia blinds more than half a million children annually in parts of Asia, Africa, Latin America, and the Middle East. It is caused by a lack of Vitamin A or, more precisely, for want of such foods as liver, eggs, carrots, and dark green leafy vegetables in diets.

Yet just 2 capsules of Vitamin A annually — at a cost of 4¢ (U.S.) — can save sight and in most cases lives as well.

WHO: Zeroing in on xerophthalmia
with Vitamin A. World Health.
June, 1985 pp 30–31.