Cohen syndrome: case report

Stephen Wilson, DMD, PhD  Victor Escobar, DDS, PhD
Joseph H. Hersh, MD  Bruce S. Haskell, DMD, PhD

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. 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 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, recent analysis strongly supports an autosomal dominance process.

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 G3P2A0 (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 occipital-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.

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

Dr. Wilson is an assistant professor, pediatric dentistry, The Ohio State University School of Dentistry, Postle Hall, 305 W 12th Ave, Columbus, OH 43210-1241. Dr. Escobar is an associate professor, diagnostic sciences; Dr. Hersh is an assistant professor, pediatrics; and Dr. Haskell is a clinical professor, orthodontics, the University of Louisville Health Sciences Center, Louisville, KY. Reprint requests should be sent to Dr. Wilson.


---

**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.