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Case Report

THE COHEN SYNDROME: REPORT OF A CASE

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Summary We report on a sporadic case satisfied with a proposed diagnostic criteria for Cohen syndrome. This 10 year-old Japanese boy had truncal obesity, short stature, mild mental retardation, hypotonia, maxillary hypoplasia, micrognathia, narrow hands and feet, high-arched palate, prominent upper central incisors, high nasal bridge, but no pigmentary retinopathy. Autosomal recessive manner of inheritance was suggested by the pedigree.

Key Words Cohen syndrome, MCA/MR syndrome

CASE REPORT

The propositus is 10-year-old Japanese boy whose parents denied consanguinity. However, grandparents of both sides were born in the same narrow district of their home town. The mother is 142 cm tall, and her two sisters also have short stature as well as obesity and infertility. In addition, two paternal aunts have short stature. He was referred to us because of mental retardation and short stature. He was the second child, when the father was 40 and the mother was 34 years of age. The first female child was suddenly died of unknown cause at one year of age. The pregnancy was uneventful. He was delivered at term by cesarean section due to cephalopelvic disproportion. His birthlength was 47 cm (−1.0 SD), birthweight 2,851 g (−0.6 SD), and OFC 36 cm (+2.2 SD). His early development was slow. He had neck control at 12 months, and walked alone at 19 months of age. Generalized hypotonia was pointed out. He had been included in a training program for developmentally retarded children through his childhood. He entered in a special class of elementary school. Obesity was first noted at age 8 years. At 10 years of age (initial visit), his height was 126.7 cm (−2.2 SD), weight 32.6 kg (−0.3 SD), and OFC 50.6 cm (−1.8 SD). His physical findings consisted of hypotonia, short stature, mild truncal obesity (degree of obesity 23%), dolichoce-

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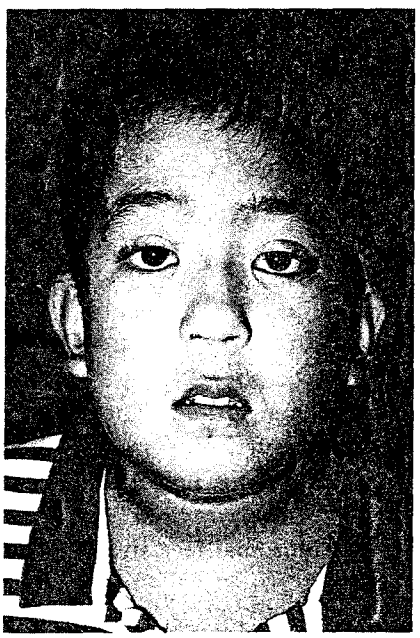

Fig. 1. Facial appearance at age 10 years. Note mild ptosis, epicanthal folds, high nasal bridge, open mouth, prominent maxillary incisors, and prominent ears.

phaly with mild frontal bossing, epicanthus, ptosis, downward slanting of the palpebral fissures, prominent ears and antihelices, high nasal bridge with bulbous tip of the nose, open mouth, prominent maxillary central incisors with malaligned teeth, small mouth, high-arched palate, micrognathia, and maxillary hypoplasia (Fig. 1). Other findings are tapered fingers, bilateral Dubois' sign, hyperextensible finger and toe joints, phimosis, concealed penis, and cryptorchidism. He had narrow feet with a wide gap between first and second toes. His IQ was evaluated around 60. His karyotypes are normal male. Thyroid test and other routine laboratory examinations were normal. Ophthalmological examinations revealed no pigmentary retinopathy or bull's eye lesion and refractive errors. A bone survey revealed normal bone age and small bilateral middle phalanges of 5th fingers. Dermatoglyphics showed 7 ulnar loops, 3 whorls, and normal atd angle.

DISCUSSION

The Cohen syndrome was first reported as an obesity/hypotonia syndrome associated with mental retardation, narrow hands and feet, and characteristic facial appearance consisting of downslanting palpebral fissures, short philtrum, open mouth, prominent upper central incisors, maxillary hypoplasia, and micrognathia

(Cohen *et al.*, 1973). To date, more than 80 cases have been reported under the designation of Cohen syndrome. In Japan, only two sporadic cases (Sameshima *et al.*, 1982; Nambu *et al.*, 1988) and one sib case with two affected brothers have been reported (Kondo *et al.*, 1990). Because it is difficult to diagnose Cohen syndrome when the case is sporadic (Gorlin *et al.*, 1990), Escobar (1990) has proposed a diagnostic criteria for the Cohen syndrome that at least five of nine major findings should be present in an individual patient: *i.e.* obesity, short stature, mental retardation, hypotonia, maxillary hypoplasia, short philtrum, micrognathia, narrow hands and feet, and narrow or high-arched palate. Our patient shared eight out of nine proposed major findings except short philtrum. In addition, he had prominent central incisors, high nasal bridge, and downward slanting of the palpebral fissures. His phenotype and pedigree suggesting autosomal recessive manner of inheritance strongly support the diagnosis of Cohen syndrome.

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