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CONGENITAL BILATERAL PERISYLVIAN SYNDROME: FIRST REPORT IN A JAPANESE PATIENT

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Summary A Japanese boy with congenital bilateral perisylvian syndrome is described. He had oropharyngoglossal dysfunction and severe dysarthria. Magnetic resonance imaging of the brain disclosed bilateral perisylvian malformations suggesting polymicrogyria. The patient also showed mental retardation, epilepsy, and poor motor skills.

Key Words congenital bilateral perisylvian syndrome, neuronal migration disorder, polymicrogyria, pseudobulbar palsy, Foix-Chavany-Marie syndrome

INTRODUCTION

Congenital bilateral perisylvian syndrome is a congenital neurological syndrome characterized by pseudobulbar palsy and bilateral perisylvian polymicrogyria (Kuzniecky *et al.*, 1993). Clinical manifestations of pseudobulbar palsy, congenital facio-pharyngo-glosso-masticatory diplegia represent developmental Foix-Chavany-Marie syndrome or bilateral anterior opercular syndrome, resulting from bilateral opercular lesions. This is the first report of a Japanese patient with this syndrome.

CASE REPORT

The patient was a 9-year-old Japanese boy with pseudobular palsy. His father, age 39, his mother, age 34, and the younger brother, age 4, were all healthy. The parents were not consanguineous. The mother had a previous spontaneous abortion at 10 weeks of gestation, and the gestation of the patient was complicated by threatened abortion with vaginal bleeding at 5 months gestation. He was born by elective Cesarean section because of bicornate uterus. The gestational period was

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35 weeks, and the birth weight was 2,642 g. Delivery and the neonatal period were uneventful.

Delayed milestones were noticed during infancy. He held his head at 3 months, but sat alone at 13 months, and walked alone at 23 months. Development of expressive language was markedly delayed. At the age of 9 years, he could vocalize only a few words which were hardly comprehensible because of dysarthria. In contrast to expressive language, his perception of language showed better development. He understood spoken commands and conveyed his intention by body language. He is currently learning sign language. His intelligence quotient was 57 on a modified Gesell scale.

His physique was normal without any dysmorphic features. G-banded chromosomes of leukocytes were 46,XY. He had slightly weak vision since he developed bilateral rhegmatogenous retinal detachments necessitating surgery at the age of 8 years. His face was expressionless with weakness of the facial musculature. The orbicularis muscle were weak, and the mouth was always half-opened. He demonstrated slight dysphagia and constantly drooled, but the gag reflex could be elicited. The most striking feature of pseudobulbar palsy was limitation of tongue movement. He could neither protrude nor laterally move his tongue. He also had poor motor skills. He was unable to hop on one leg, and used his hands clumsily.

He had febrile convulsions at the age of 1 and 2 years. He developed a few episodes of brief nocturnal generalized tonic-clonic seizures at the age of 8 years. An awake record of the electroencephalograms (EEGs) revealed occasional spikes in the left central area. The background activity was in the normal range, alpha waves around 9 c/sec. Epilepsy was controlled by carbamazepine.

Magnetic resonance imaging (MRI) taken at the age of 3 years showed bilateral perisylvian malformations suggesting polymicrogyria or macrogyria (Fig. 1). There were gyral abnormalities involving the bilateral perisylvian and perirolandic regions, extending high into the parietal regions. The cortex appeared thick and smooth, with the underlying white matter diminished.

DISCUSSION

Congenital bilateral perisylvian syndrome was named in a report of 31 similar cases in 1993 (Kuzniecky *et al.*, 1993). Clinical, electroencephalographical, and neuroradiological characteristics have been delineated (Guerrini *et al.*, 1992a, b; Kuzniecky *et al.*, 1994a, b, c). Our patient represents a typical case of this syndrome. This patient met all the essential criteria established by Kuzniecky *et al.* (1993); oropharyngoglossal dysfuction, moderate to severe dysarthria, and bilateral perisylvian malformations on imaging. He also has all of the additional criteria; delayed milestones, epilepsy, mental retardation, and abnormal EEG, although he does not have other criteria of the syndrome; arthrogryposis multiplex, other limb malformations, or infantile spasms. Patients with this syndrome have been so far

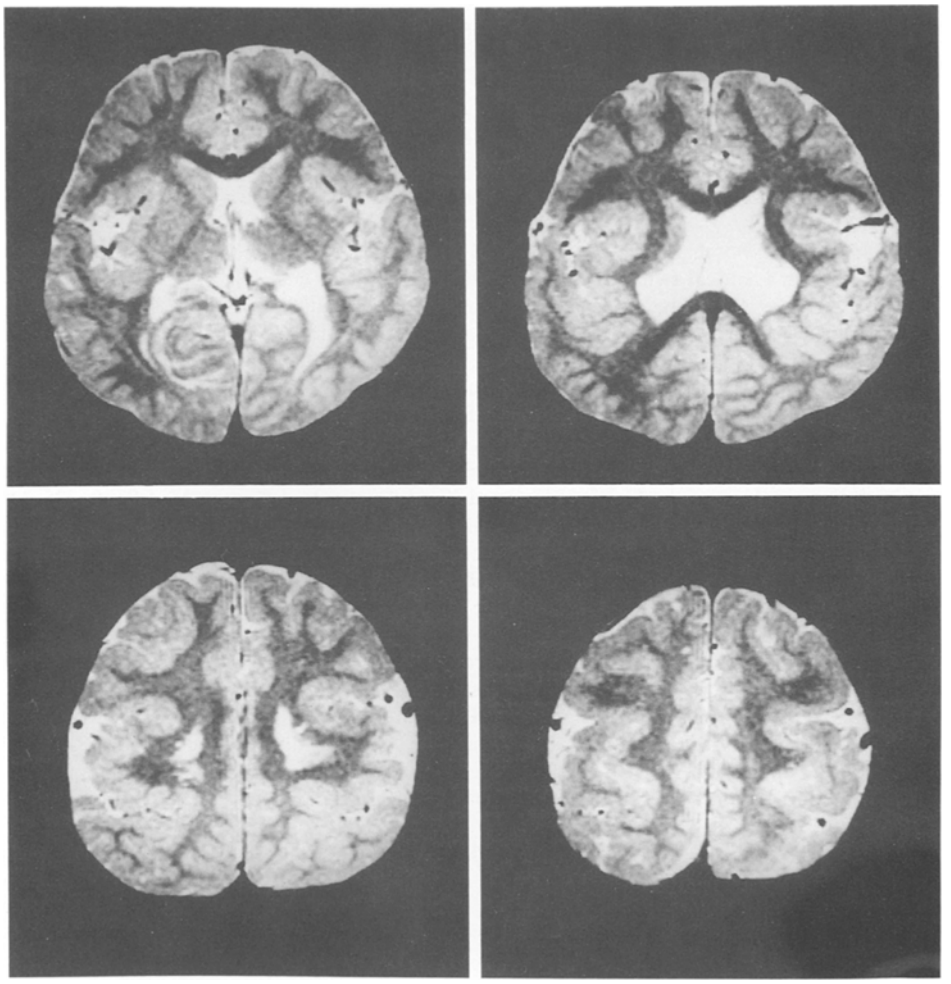

Fig. 1. T2-weighted images (TR=3,000 ms, TE=80 ms) of cranial MRI.

reported in North America, Western Europe, and Korea (Kim *et al.*, 1994). To our knowledge, this syndrome has not previously been reported among Japanese.

The neurological manifestations of this syndrome reflect the neuronal migration disorder of the bilateral perisylvian and perirolandic regions including the operculum. Although MRI cannot precisely differentiate between polymicrogyria and macrogyria (Kuzniecky *et al.*, 1989), autopsy findings of patients with this syndrome demonstrated that the underlying abnormality is polymicrogyria (Becker *et al.*, 1989; Shevell, 1992; Kuzniecky, 1994a).

The etiology of this syndrome remains unknown, although restricted ischemic injury during fetal brain development is postulated for the development of perisylvian polymicrogyria (Kuzniecky *et al.*, 1993). A genetic factor should be

contributory, since this syndrome was reported in identical twins (Graff-Radford *et al.*, 1986) and in siblings (Kuzniecky, 1993).

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