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Congenital bilateral perisylvian syndrome as a rare clinicoradiological entity: a case report

Nadir bir klinikoradyolojik durum olarak konjenital bilateral perisilviyan sendrom: olgu sunumu

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Abstract

Congenital bilateral perisylvian syndrome (CBPS) is a recently described syndrome that includes developmental delay, variable cognitive deficits, prominent cortical pseudobulbar symptoms, and variable pyramidal signs. Seizures are common, and imaging studies are characteristic examinations. The underlying pathology is polymicrogyria. Polymicrogyria may have a focal or regional distribution or involve the whole cortical mantle. Females are affected more often than males. The sylvian fissures often extend more vertically at their posterior extent into the parietal lobes. The abnormality is usually symmetric. In this paper, we present a case of CBPS, and discuss the clinical and radiologic characteristics of this rare condition.

Keywords: Perisylvian polymicrogyria, developmental abnormalities, magnetic resonance imaging

Özet

Konjenital bilateral perisilviyan sendrom (KBPS), gelişme geriliği, değişik bilişsel bozukluklar, belirgin kortikal psödobulber semptomlar ve piramidal bulgular ile karakterize, son yıllarda tanımlanmış bir durumdur. Nöbet sık görülen bir bulgu olup görüntüleme çalışmaları karakteristiktir. Altta yatan patoloji, polimikrogiridir. Polimikrogiri, fokal veya bölgesel dağılım gösterebilir veya tüm kortikal mantoyu etkileyebilir. Kadınlar, erkeklerden daha sık etkilenmektedir. Silviyan fissürler daha vertikal seyirli olarak pariyetal loblara doğru uzanmaktadır. Anomali sıklıkla simetriktir. Bu yazıda, KBPS'li bir olguyu klinik ve radyolojik özelliklerini tartışarak ortaya koyuyoruz.

Anahtar sözcükler: Perisilviyan polimikrogiri, gelişimsel anomaliler, manyetik rezonans görüntüleme

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Introduction

Congenital bilateral perisylvian syndrome (CBPS) is an extremely rare neurological disorder that may be apparent at birth (congenital), infancy, or later during childhood. It is characterized by partial paralysis of muscles on both sides (diplegia) of the face, tongue, jaws, and throat (pseudobulbar palsy); difficulties in speaking (dysarthria), chewing (mastication), and swallowing (dysphagia); and/or sudden episodes of uncontrolled electrical activity in the brain (epilepsy). The term CBPS describes a structural

malformation of the brain. The underlying anomaly is polymicrogyria, a malformation of the cerebral cortex (outer layer of the brain) [1–3]. In this report, we present the clinical and imaging findings in a child patient with this rare condition.

Case report

An 11-year-old boy was admitted with mental retardation, dysarthria, difficult swallowing, and longstanding seizures. He was the product of a term pregnancy and was delivered by cesarean section. The patient's birth weight was 3100 g. He had bilateral club foot which were treated and she walked at 2 years of age. His siblings are normal: two girls and one boy; all the other children are healthy. Our examination included the following: psychological assessment, neurologic examination and neuroimaging evaluation. Our patient had low verbal and performance IQ levels. Global IQ was estimated to be 68. He had generalized tonic-clonic seizures. The cranial nerve functions, including eye movements and ophthalmoscopy, were normal. No signs of bulbar paralysis were present. He responded to visual and acoustic stimuli. The deep tendon reflexes were brisk. Obvious signs of mental retardation were present. He is right-handed. Speech development was strongly impaired by dysarthria. Mental development was delayed and required special education. At 8 years of age, he had a first seizure. Electroencephalography (EEG) monitoring captured bilateral, independent temporal interictal activity. Brain auditory-evoked responses were within normal range. Toxoplasma, rubella, cytomegalovirus and herpes virus (TORCH) screenings were negative. In addition, testing for myotonic dystrophy, urine and serum amino acids, urine organic acids, Watson-Schwartz, serum ceruloplasmin and heavy metals in urine were negative. Chromosomal analysis was normal. Electrocardiogram, echocardiogram, and abdominal ultrasonography were normal. Dysmorphic features were not observed. Anticonvulsant therapy was started.

To elucidate the underlying pathologic abnormality, cranial magnetic resonance imaging (MRI) examination of the patient was performed on a 1.5 Tesla magnetic resonance (MR) imager. T1-weighted spin-echo (SE) (repetition time [TR] =440msec/ echo time [TE] =15msec) and T2-weighted fast spin-echo (FSE) (TR=4400msec/TE=100msec), fluid attenuated inversion recovery sequence (FLAIR) (TR=8800msec/TE=140msec, TI=2200msec) images in axial, coronal and sagittal planes were obtained. The slice thichness was 3 mm. No gadolinium-based contrast material was administrated. Bilateral symmetrical dysplastic cortices were observed in the region of the sylvian fissures, giving normal signal on T1- and T2-weighted images; the underlying white matter was also thought to be normal (Figures. 1A-C). The sylvian fissures extended more superiorly and posteriorly than normal. The corpus callosum, basal ganglia, internal capsules, cerebellum, brain stem and pituitary gland were normal. The subarachnoid spaces were enlarged. Large cortical veins were present in the vicinity of the polymicrogyria. In our patient, the clinical and imaging findings were consistent with a syndrome labelled as CBPS.

Discussion

The term "CBPS" describes a structural malformation of the brain. CBPS is synonymous with perisylvian syndrome, perisylvian polymicrogyria, Worster- Drought Syndrome and bilateral opercular syndrome. The underlying anomaly is polymicrogyria, a malformation of the cerebral cortex (outer layer of the brain). The term polymicrogyria designates an excessive number of small and prominent convolutions spaced out by shallow and enlarged sulci (grooves), giving the surface of the brain a lumpy aspect. Although it may be difficult to recognise mild forms of polymicrogyria on a MRI scan, infolding of the outer layer of the brain and secondary, irregular, thickening due to packing of microgyri (small folds) represent quite distinctive MRI characteristics [1-4]. This syndrome was described by Kuzniecky et al. [2] as a congenital pseudobulbar palsy associated with cognitive deficits in the majority, and with bilateral perisylvian abnormalities consisting

of polymicrogyria on radiological imaging, confirmed by autopsy; radiological imaging is of particular importance in establishing the diagnosis.

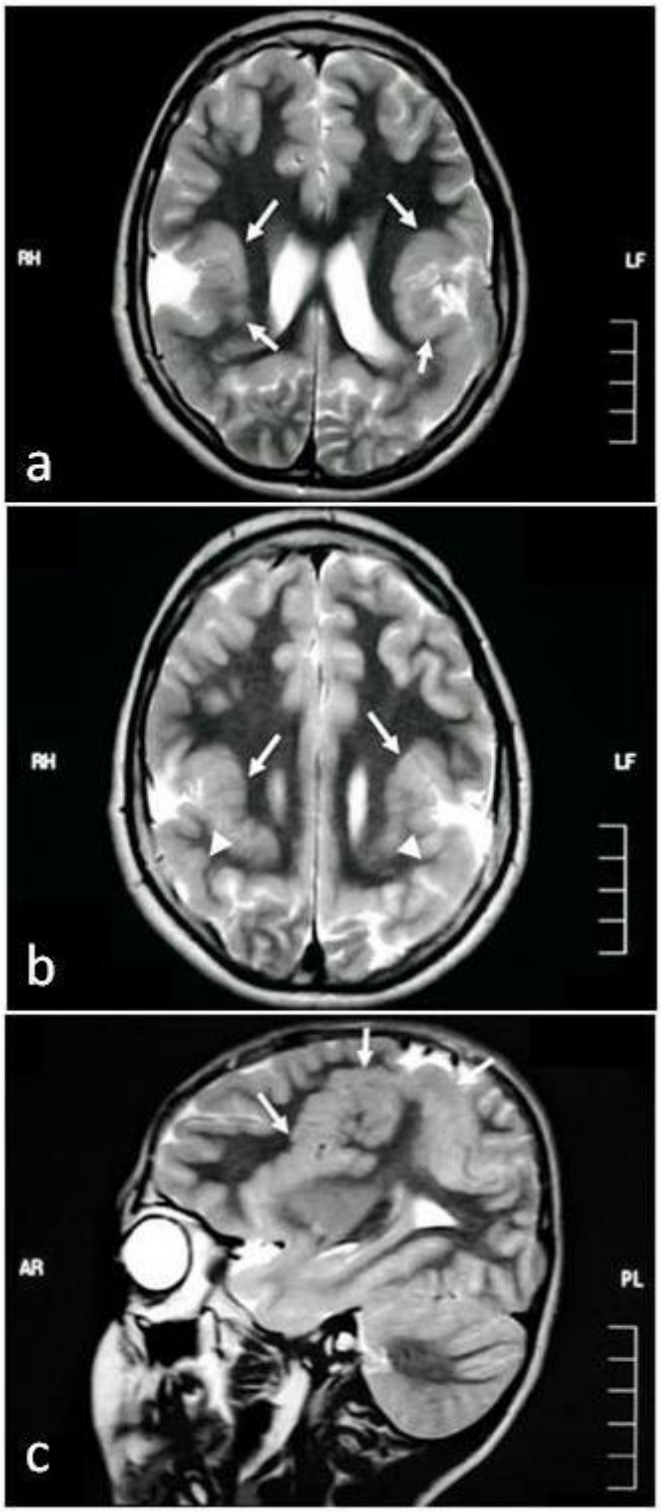

Figure 1. A 11-year-old boy. (a, b) Axial T2-weighted MR scans demonstrate that the sylvian fissures are abnormally formed bilaterally (white arrows) and are lined by polymicrogyric cortex extending posteriorly into the parietal parasagittal area (arrowheads), (c) Sagittal T2-weighted image shows polymicrogyria of the perisylvian cortex, posteriorly extending into the parieto-occipital cortex (white arrows).

The main features of CBPS are pseudobulbar palsy, cognitive deficits, epilepsy, and perisylvian abnormalities on imaging studies [2–4]. The cortical abnormality seen in the perisylvian region is consistent with polymicrogyria and is usually symmetric but varies in extent among patients [4, 5]. Patients have paralysis of the face, throat, tongue and the chewing process, with dysarthria (speech difficulties) and drooling. Most of patients have cognitive deficit and epilepsy. Fixed deformity of the ankle joints (arthrogryposis) has been described in some patients. Seizures usually begin between the ages of 4 and 12 years and are poorly controlled in about 60 per cent of patients. The most frequent seizure types are atypical absences, tonic or atonic drop attacks and tonic-clonic seizures, often occurring as Lennox-Gastaut syndrome. A minority of patients (26 %) have partial seizures [6].

Polymicrogyria may have a focal or regional distribution or involve the whole cortical mantle (covering of the brain). There are consequently a wide spectrum of clinical manifestations which include children with severe encephalopathies (brain impairments) and intractable epilepsy, or normal individuals with selective impairment of cognitive functions (mental processes) in whom the mild cortical abnormality is only detected on pathological brain study [6]. The patient described in this report had seizures, similarly to previously reported cases with CBPS.

According to the location, polymicrogyria can appear as a diffuse or focal lesion [1, 4]. Focal polymicrogyria can demonstrate either bilateral or unilateral involvement. Bilateral lesions with involvement of the frontal, parietal, occipital, frontoparietal, or temporoparietal areas have been described by Barkovich and Kjos [4]. Bilateral frontoparietal abnormalities were also described by the latter investigators in four patients [7] A more extended classification of gyral abnormalities, based on 90 patients, was published by Borgatti et al. [9] Three forms of bilateral polymicrogyria (perisylvian, parieto-occipital, and frontal) have been differentiated by these investigators. Patients with frontal involvement present with cognitive and motor delay, and have spastic quadriparesis. Patients with involvement of the perisylvian cortex (CBPS) present with phonation problems and speech delay. They have symptoms of dysarthria, oro-facial paresis and typically the inability to move the tongue from side to side. Patients with involvement of the parieto-occipital cortex have a serious cognitive deficit, minor motor dysfunction and seizures. Patients with extensive regions of polymicrogyria all have severe cognitive and motor delay, seizures and cerebellar dysfunction [4–8]. In our case, the bilateral involvement of the insular cortex explains the delayed language development, the difficulties with palatal and lingual movements and speech delay.

The etiology of CBPS is heterogenous. Bilateral perisylvian polymicrogyria occurs sporadically in most patients. The role of enviromental, acquired factors has been sugested by the topographic arrangement of the lesions, the frequency of bilateral symmetry, and historical data. Transient intrauterine perfusion failure and intrauterine infections, such as cytomegalovirus, toxoplasmosis, syphilis and varicella-zoster, can be responsible for the development of polymicrogyria [7]. The role of toxic insults was also suggested and indeed, injection of ibotenate, a glutamaterjic agonist in developing mouse neopallidum produces lesions that mimic microgyrias. Based on these experiments the involvement of excitotoxicity, associated with hypoxia/ischemia, was postulated as a contributing factor to the development of polymicrogyria [8]. Several malformation syndromes featuring bilateral polymicrogyria have been described, including bilateral perisylvian polymicrogyria (the most frequent form), bilateral parasagittal parietooccipital polymicrogyria, bilateral frontal polymicrogyria and unilateral perisylvian or multilobar polymicrogyria [8, 9].

Bilateral perisylvian polymicrogyria has been reported in children born from identical twin pregnancies which were complicated by twin-to-twin transfusion syndrome [6]. Recently, Borgatti et al. [9] described a family in which CBPS was present in 6 individuals of 3 consecutive generations. In our patient, the etiology of the

polymicrogyria was unknown. In the present case, the localization was not consistent with distribution of any cerebral artery, and no evidence of any intrauterine infectious or toxic insult was evident. Some other malformations and syndromes encountered with CBPS, such as septum pellucidum defect, Kabuki syndrome, congenital constriction band syndrome, oesophageal malformations, limb atresia, jejunal atresia, Ehlers-Danlos syndrome, and trisomy-13 syndrome, have been reported in the literature [10, 11].

Yekeler et al. [12] reported in their study a case of CBPS with pituitary hypoplasia and ectopic neurohypophysis, and they claimed that when bilateral polymicrogyria has been identified, the whole brain parenchyma should be evaluated with detailed MRI. Several families with multiple affected members have been reported with possible autosomal recessive, X-linked dominant and X-linked recessive inheritance. Some families have been linked to the Xq27-q28 chromosomal region but the causative gene is not known at present [6].

Polymicrogyria may be difficult to demonstrate with CT but is identifiable on MRI as thickened cortex, poorly developed sulci and an irregular margin at the cortical white matter junction. Abnormalities of cortical venous drainage and difficult venous access are often present. In Bilateral Perisylvian Polymicrogyria, the opercula are dysplastic and incomplete. The sylvian fissures are wide and underdeveloped. Sagittal images may show posterior extension of the sylvian fissure, exposure of the insula and apparent thickening of the cortex. The bodies of the lateral ventricles show inverted appearance, typical of this disorder. Proton MR spectroscopy of the brain allows noninvasive in vivo assessment of metabolites, which may be useful in understanding the biology of malformations of cortical maldevelopment. The neurons of glia in these areas and the metabolites appear to be similar to those of normal adult frontal white matter [6, 8, 9]. In our patient, cranial CT demonstrated symmetrical bilateral perisylvian cortical thickening. In these regions, the cortex was thick and smooth, and the sylvian fissures were slightly enlarged. Cranial MRI confirmed bilateral perisylvian involvement; with multiplanar higher resolution. No nodular heterotopia or other abnormality was seen on MRI. On MRI examination, polymicrogyria can mimic pachygyria, as the cortex is slightly thickened and the sulci are shallow [13]. In polymicrogyria, the cerebral cortex is not as thick as in pachygyria; it is in the 5–7 mm range, normal being 3 mm and a thickness of more than 8 mm is seen in most cases of pachygyria [14].

The differential diagnosis of CBPS includes acquired bilateral perisylvian defects (usually vascular in origin, often asymmetric, not associated with polymicrogyria) and bilateral cortical infoldings secondary to shunted hydrocephalus [15]. In CBPS, the main differential diagnosis is the Worster-Drought syndrome (WDS). There are similarities between WDS and CBPS, and it may be that there is a continuum between the two entities. It is possible that patients with the WDS are a subset of patients with bilateral perisylvian syndrome [16]. Similar acquired symptoms have been described in adults as the Foix-Chavany-Marie syndrome due to bilateral anterior opercular infarctions [17].

In conclusion, the CBPS is a rare clinicoradiologic entity. The underlying abnormality is polymicrogyria. The advanced in neuroimaging, particularly, high-resolution MR imaging, have enabled the diagnosis of more subtle forms of cortical abnormalities around the sylvian fissures.

References

1. Kuzniecky R, Andermann F, Tampieri D, Melanson D, Olivier A, Leppik I. Bilateral central macrogyria: epilepsy, pseudobulbar palsy and mental retardation-a recognizable neuronal migration disorder. Ann Neurol 1989; 25: 547–54.
2. Kuzniecky R, Andermann F, Guerrini R. Congenital bilateral perisylvian syndrome: study of 31 patients. Lancet 1993; 341: 608–12.
3. Gropman AL, Barkovich AJ, Vezina LG, Conry JA, Dubovsky EC, Packer RJ. Pediatric Congenital bilateral perisylvian syndrome: Clinical and MRI features in 12 Patients.

Neuropediatrics 1997; 28: 198–203.

4. Barkovich AJ, Kjos BO. Nonlissencephalic cortical dysplasias: correlation of imaging findings with clinical deficits. Am J Neuroradiol. 1992; 13: 95-103.
5. Guerrini R. Polymicrogyria and epilepsy. In: Spreafico R, Avanzini G, Andermann F, eds. Abnormal cortical development and epilepsy. London: John Libbey, 1999; p.191–201.
6. Hattori H, Higuchi Y, Maihara T, Jung EY, Furusho K, Asato R. Congenital bilateral perisylvian syndrome: first report in a Japanese patient. Jpn J Hum Genet 1996; 41: 1189–92.
7. Harding B, Copp AJ. Malformations. In: Graham DI, Lantos PL (eds). Greenfield's Neuropathology 6th edn. London, Sidney, Auckland: Arnold, 1997: p.397–533.
8. Sztriha L, Nork M. Bilateral symmetrical frontoparietal polymicrogyria. Eur J Pediatr Neurol 2002; 6: 229–32.
9. Borgatti R, Triulzi F, Zucca C, Piccinelli P, Balottin U, Carrozzo R, Guerrini R. Bilateral perisylvian polymicrogyria in three generations. Neurol 1999; 52: 1910–3.
10. Humbertclaude V, Pedespan JM, Azais M, Fontan D, Echenne B. Schizencephaly and upper limb malformation. Arch Pediatr 1996; 3: 357–9.
11. Kuker W, Friese S, Riethmuller J, Krageloh-Mann I. Congenital bilateral perisylvian syndrome (CBPS): Do concomitant oesophageal malformations indicate a poor prognosis? Neuropediatrics 2000; 31: 310–3.
12. Yekeler E, Ozmen M, Genchellac H, Dursun M, Acunas G. Congenital bilateral perisylvian syndrome with pituitary hypoplasia and ectopic neurohypophysis. Pediatr Radiol 2004; 34: 908–11.
13. Titelbaum DS, Hayward JC, Zimmerman RA. Pachygyric-like changes: topographic appearance at MR imaging and CT and correlation with neurologic status. Radiology
14. Barkovich AJ, Koch T, Carrol C. The spectrum of lissencephaly; report of ten cases analyzed by magnetic resonance imaging. Ann Neurol 1991; 30: 139–46.
15. Fischbein N, Dillon WP, Barkovich J. Teaching atlas of brain imaging (1st ed). New York: Thieme, 2000.
16. Worster-Drought C. Suprabulbar paresis. Congenital suprabulbar paresis and its differential diagnosis, with special reference to acquired suprabulbar paresis. Dev Med Clin Neurol 1974; 30: 1–30.
17. Mariani C, Spinnler H, Sterzi R, Vallar G. Bilateral perisylvian softenings: bilateral anterior opercular syndrome (Foix-Chavany-Marie syndrome). J Neurol 1980; 223: 269–84.