A case of congenital bilateral perisylvian syndrome due to bilateral schizencephaly

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ABSTRACT – A case of congenital bilateral perisylvian syndrome (CBPS) associated with bilateral perisylvian schizencephaly in a 24-year-old woman is reported. She presented the classical clinical triad of CBPS, which included congenital facio-masticatory diplegia, epilepsy and only mild mental retardation, despite the presence of bilateral, open-lip clefts in the perisylvian region. We hypothesize that the minimal loss of cortical tissue, along with the possible sparing of vital white matter association fibers and neuronal plasticity might have contributed to the better functional outcome in this patient.

Key words: congenital bilateral perisylvian syndrome, schizencephaly, polymicrogyria

The congenital bilateral perisylvian syndrome (CBPS) was initially described by Kuzniecky et al. (1993) in a group of 31 patients with the syndrome of congenital faciopharyngomasticatory diplegia, epilepsy and mild mental retardation, along with bilateral perisylvian cortical malformation on neuroimaging. MRI showed abnormally thick cortex intermixed with shallow sulci and broad gyri. On inversion recovery sequences, the abnormal cortex showed increased interdigitations between white and gray matter and small, fused gyri suggestive of polymicrogyria. The abnormality was symmetrical in more than 80% of cases. Autopsy studies performed in two patients confirmed the presence of four-layered polymicrogyria in these regions (Kuzniecky and Andermann 1994). Several subsequent reports described variant imaging findings such as marked asymmetry and associated involvement of the septum pellucidum (Sejima et al. 2001), pituitary hypoplasia and ectopic neurohypophysis (Yekeler et al. 2004) and other structural abnormalities of the cortex. Gropman et al. (1997) described a group of 12 children with classical features of CBPS; two of them had unilateral perisylvian schizencephaly with contralateral perisylvian polymicrogyria. There were no differences in the clinical presentation of these patients compared to the rest of the group. The authors hypothesized that this might be due to the presence of polymicrogyria along the walls of the cleft. Here, we report a case of bilateral perisylvian schizencephaly presenting with features of CBPS in a young woman. We were not able to locate any previous reports of such an association.
Figure 1. Inter-ictal EEG-common average referential montage: paroxysm of generalized, irregular, high amplitude theta and delta slow waves can be seen, preceded by small spikes, asymmetrically prominent over the frontotemporal regions.

Figure 2. MRI brain axial images (T1 weighted and FLAIR): the perisylvian clefts are seen extending to the lateral ventricles on both sides, suggesting bilateral open-lip schizencephaly.
Case report

This 24-year-old woman, the third child born to non-consanguineous parents, attended our epilepsy clinic because of seizures of three years duration. She was born at full term, with normal delivery and without any perinatal complications. According to her mother, there were no delays in her motor and mental milestones. However, her expressive speech was slow and effortful from early childhood. Reading aloud was difficult, although comprehension and writing were normal. She also used to take a lot of time chewing and swallowing, with occasional episodes of choking. She displayed drooling of saliva from early childhood that was persistent even at the time of presentation. Her academic achievement was comparable to her siblings.

Her epilepsy had begun at the age of 21 years. The stereotyped seizure started with sudden behavioral arrest without any preceding aura. This was followed by a vacant stare, twitching of the lips, forced opening of the lower jaw and drooling of saliva. Generalized tonic-clonic seizures ensued in all instances. The post-ictal phase was characterized by significant dysthria, along with dysphagia and excessive drooling of saliva lasting for several hours. At the time of her first visit, she was taking zonisamide and carbamazepine with good seizure control. However, she complained of anorexia and excessive weight loss.

Examination revealed dolichocephaly, with a head circumference of 51 cm. There were no other dysmorphic features. Systemic examination showed spastic dysthria with slow movements of the tongue. Lingual and labial sounds were significantly affected. Emotional-voluntary dissociation was noted for facio-oral movements. The jaw-jerk was exaggerated and all other deep tendon reflexes were symmetrically brisk. Mild mental retardation (IQ 54) was documented on formal psychometric assessment. Her EEG showed frequent paroxysms of generalized, irregular, high amplitude theta and delta slow waves preceded by occasional small spikes, asymmetrically prominent over the frontotemporal regions (figure 1). MRI brain showed bilateral, perisylvian open-lip schizencephaly with no other structural malformations (figure 2).

Discussion

Schizencephaly is a malformation of cortical development characterized by a congenital cleft of the cerebral mantle extending from the pial surface to the lateral ventricle, lined by cortical gray matter (Granata et al. 1996). It is thought to be a defect arising in the late stage of cortical organization. Thus, it is closely linked to polymicrogyria, which are also produced by a defect at the same stage of cortical development. Moreover, morphological studies have shown that the clefts of schizencephaly are lined by polymicrogyric cortex (Kuzniecky and Barkovich 2001). In cases of unilateral schizencephaly, polymicrogyric cortex has been shown in corresponding areas in the opposite hemisphere (Hayashi et al. 2002). Accordingly, the recent classification of malformations of cortical development considers both schizencephaly and polymicrogyria as a continuous spectrum of disorders (Barkovich et al. 2005). A significant insult results in open-lip schizencephaly with a major loss of cortical tissue and disruption in white matter connections leading to severe intellectual deficits. Milder insults result in polymicrogyria with minimal intellectual impairment. However, there seems to be no correlation between severity of epilepsy and the extent of cortical damage. Epilepsy is reported to be more severe in unilateral compared to bilateral schizencephaly (Kuzniecky and Barkovich 2001). This may be attributed to the more extensive disruption of the pathways of seizure-spread in bilateral clefts.

The neurological deficits and the electroclinical features of epilepsy in this patient were similar to the reported cases of CBPS due to perisylvian polymicrogyria (Kuzniecky et al. 1994, Tanaka et al. 2000). However, the severity of the functional deficit was significantly less compared to the extent of cortical damage. This may be attributed to the minimal loss of cortical tissue in spite of the presence of bilateral clefts. Moreover, sparing of the vital white matter association fibers along with neuronal plasticity might have contributed to the less severe functional impairment (Barkovich and Kjos 1992).

This case adds further evidence favoring the concept of a polymicrogyria-schizencephaly spectrum of malformations. It can be postulated that the clinical presentation of CBPS may arise from a range of perisylvian cortical malformations occurring at the stage of late neuronal migration and cortical organization, varying from mild to severe forms. Hopefully, further, advanced structural and functional neuroimaging studies may be able to shed more light on this condition.

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References


