American Medical Journal 3 (2): 228-230, 2012
ISSN 1949-0070

Schizencephaly: Case Report

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Abstract: Problem statement: Whereas schizencephaly is a malformation of cortical development which exact cause remains unknown, is almost entirely characterized by clefts extending from the pial surface to ependymal surface and could be unilateral or bilateral. Oftentimes, it also occurs in association with other abnormalities. Over the years, two types were described in literature, lips closed and open. Therefore, our intention is describe a case of schizencephaly monitored by the Neurology Service of Nova Iguacu University Hospital. **Approach:** A 29-year old single male student, presenting clinical history of epilepsy since he was 23 years old (partial seizures with secondary generalization) showed a global delay in neuropsychomotor development. **Results:** The patient has been admitted in the emergency department and his seizures were controlled with IV diazepan, followed by phenobarbital and phenotoyn. CT scan has revealed a cortical maldevelopment described as bilateral schizencephaly. After these procedures, he was released six days later, seizure-free, using carbamazepine and phenotyn. **Conclusion:** Is indeed that prognosis for individuals with schizencephaly varies depending on the size of the clefts and the neurological deficit degree. In general, treatment for individuals with schizencephaly usually consists of physical therapy, occupational therapy, treatment for seizures and, in a few cases complicated by hydrocephalus, a shunt is required.

Key words: Epilepsy, schizencephaly, brain malformation, computed tomography

INTRODUCTION

Schizencephaly is a developmental malformation of cerebral cortex, a congenital disorder whose exact cause still remains unknown. Nevertheless, the most accepted theory that have been accepted until nowadays, is the defective proliferation and/or neuronal migration and cortical layer organization (Marinelli *et al*., 2011; Rodrigues *et al*., 2006; Santra and Sen, 2008). Recent studies suggest the involvement of EMX2 gene in this disorder physiopathogenesis (Rodrigues *et al*., 2006; Spalice *et al*., 2009; Squier and Jansen, 2010).

Schizencephaly was first described by Yakovlev and Wadsworth in 1946. It consists of gaps extending from the cortex reaching into the ventricle, from the pial membrane level up to the ependyma layer, with its margins being lined by gray cortical matter, primarily affecting the perissylvian areas (Marinelli *et al*., 2011; Spalice *et al*., 2009; Amaral *et al*., 2001). It also has been associated with other abnormalities such as polimicrogyria, pquigyria, heterotopias, agenesis of the septum pellucidum, optic nerve hypoplasia as well as agenesis of corpus callosum (Rodrigues *et al*., 2006; Amaral *et al*., 2001). Two types of schizencephaly are commonly described: type 1 or “closed lip” showing

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both cortical borders juxtaposed and type 2 or "open lip" whose hiatus or gap (or cavity) is filled with cerebrospinal fluid, this last one is the most common of both (Marinelli *et al*., 2011; Santra and Sen, 2008; Amaral *et al*., 2001; Kalia and Saggar, 2009).

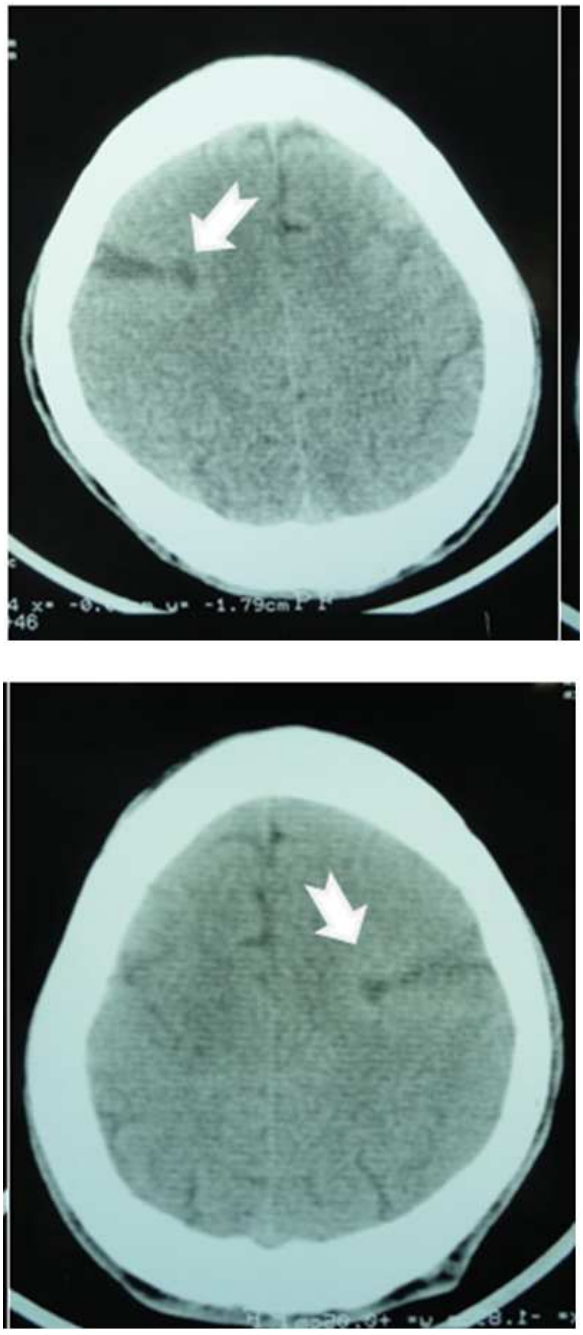

Fig. 1: Imaging studies showed parietal gaps of closed lips, small and on both sides, taken together confirms the schizencephaly diagnosis

The clinical picture presents itself according to the severity of symptoms, depending upon the exact extent of the anatomical defect and also on the location of the brain defect (Amaral *et al*., 2001). The most common clinical findings include: motor deficit, epilepsy/seizures and mental retardation associated to delayed neuropsychomotor development (Rodrigues *et al*., 2006; Amaral *et al*., 2001).

The diagnosis establishment comes through neuroimaging: CT or MRI, the latter being much better at detecting the gaps and spaces, abnormalities coexistence (Amaral *et al*., 2001; Kalia and Saggar, 2009). Nowadays, prenatal diagnosis is also possible through ultrassound and MRI, especially in type 2 or "open lip" schizencephaly (Witters *et al*., 2007; Mancini *et al*., 2001).

Case report: A 29-year old single male student with a history of epilepsy since 23 years old (partial seizures with secondary generalization) presenting a global delay in neuropsychomotor development. Eventual seizures persist, particularly after medication on an irregular basis (100 mg of phenobarbital 12/12 h). The patient has been accepted in the emergency department of Nova Iguaçu General Hospital in July/2011 due to seizures. His seizures were controlled using IV diazepan, followed by phenobarbital and phenotoyn. CT scan has revealed a cortical maldevelopment described as bilateral schizencephaly (Fig. 1). The patient was released six days later, seizure-free, in use of carbamazepine (200 mg 8/8h) and phenotyn(100 mg 12/12 h). He is currently in follow up at this hospital´s neurology unit.

DISCUSSION

In mammals, the conformation of cerebral cortex is a process featured of proliferation, migration, differentiation and cell death (Meneses *et al*., 2006; Silva and Cavalheiro, 2004). The cortical development disorders results in changes in cytoarchitecture of the cerebral cortical layers (Meneses *et al*., 2006; Sarkis *et al*., 2010). Innumerable classification of these disorders ranging from anatomy, histopathology, embryology until genetics (Meneses *et al*., 2006; Barkovich *et al*., 2005). As such, schizencephaly is a developmental cortical desorder, integrating group of malformations in cortical organization. It is believed that an initial genetic or enviromental injury occurs just after neuronal migration, under this circunstances, the cytoarchitecture of the cortex when continuity begins to develop between itself and the telecephalon (Meneses *et al*., 2006; Palmini, 2000).

Several cortical malformations represents a multifaceted group of etiologies/causes related to epilepsy and also difficult the clinical control (Meneses *et al*., 2006). Our patient demonstrated delay in neuropsychomotor development of milestones (at first, he walked at six years old and learned to read at ten), Alongside the presence of a 4/d spastic tertraparesis, hyperrreflexia and cognitive deficits were associated to a clinical epilepsy history, in his third decade of life. Features on neuroimaging studies revealed parietal gaps of closed lips, small and located on both sides, ratifying the schizencephaly diagnosis, even though craneal MRI, considered the gold standard for diagnosis was not performed. Although less meticulous, CT scan is cheaper and more accessible to the general population (Amaral *et al*., 2001). The patient currently undergoes physical therapy as well as occupational therapy to handle his motor sequelae.

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