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SCHIZENCEFALIA - A Bibliographic Review of Clinical Aspects

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ABSTRACT

The main objective of this review is to analyze the clinical findings of patients diagnosed with schizencephaly, elucidating their main characteristics and symptoms and main therapeutic approaches. This is a bibliographic review based on the specialized literature through consultation of scientific articles selected through search of the scielo and peDRO database, from Medline and Lilacs sources. The studies found schizencephaly as a disorder in neural migration characterized by clefts in the cerebral cortex, which lead to cognitive, motor and neuropsychomotor development since it is a congenital pathology. The treatment presented is variable and dependent on the clinical picture of the patient and the histological characteristic of the lesion and its impact on the central nervous system.

Keywords

Central nervous system, Schizencephaly, Cerebral Cortex, Congenital Abnormalities.

Introduction

Schizencephaly is a disorder of neuronal migration characterized by fissures of the cerebral cortex, which extend from the surface of the pial to the lateral ventricle. At the edges of the slits there is a cortical layer with polymicrogyria (a malformation caused by defects in the development of the cerebral cortex), the lamination of which consists of four layers of cells. Schizencephaly is a rare congenital disease characterized by clefts that extend from the thickness of the cerebral hemispheres to the ventricular and subarachnoid spaces, that is, caused by an error in the migration of the neurons that will form the cerebral cortex. It is affected in the mother's belly between the second and fifth month of gestation. It is believed to be due to poor blood circulation of the mother or genetic inheritance [1-3].

But so far there are no specific conclusions that can state such diagnoses. There are two types of schizencephaly, type I schizencephaly has a gray mass of tissue of matter without any cleft or fluid with ventricular or cortical lips that close at one end of a fluid through the abnormal slit of the hemisphere. Type II schizencephaly shows a full cerebrospinal fluid and a slit of variable size and shape that extends through the hemisphere of the eppendimus to peripherally [4-6].

Epidemiology

Because it is considered a rare disease worldwide, the prevalence of schizencephaly among the group of malformations of neuronal migration is between 3-7%. With 1.54 per 100,000 live births in a study based on the US population. There is no evidence to show the existence of racial predisposition or by sex [6].

Etiology

Because it is a low incidence pathology, the main risk factors that trigger this cascade of neural migration disorders are still unknown. However, the most reliable hypothesis concerns an increase in the relative risk of almost four times higher in the offspring of young parents and more than twice in monozygotic twins and also by a point mutation in Chromosome 10q26.1 where the gene is located EMX2 development homeobox, which is responsible for the configuration of the central nervous system and the urogenital tract.

The prognosis is variable, as it depends on the size of the slits and the degree of neurological deficit.

This genetic alteration is not present in all cases and that the same genetic commitment manifests itself with variable expression, suggests that the loss of function in the EMX2 gene may be the first step of a more complex pathogenic mechanism in which other factors can influence the severity and extent of lesions. The careful collection and analysis of clinical, genetic, epidemiological and radiological data can finally identify the specific risk factors involved in the onset of schizencephaly.

Clinical Presentation

Symptomatology of patients with schizencephaly is quite varied and the degree of complexity/severity of some symptoms is related to the extent of the anatomical defect and its location, but recent findings state that epilepsy, motor deficit, and the mental retardation.

The schizencephaly are classified in radiological studies according to the presentation in closed lips or open lips.

The diagnostic evaluation of the esquinzecefalias is performed by means of computed tomography (CT) and magnetic resonance imaging (MRI). Through them unilateral or bilateral slits can be seen, ranging from very narrow or very wide. Four findings allow us to diagnose developmental anomalies and differentiate them from destructive processes in imaging tests such as the presence of clefts surrounded by abnormal cortex; the ventricle has a triangular configuration adjacent the crevice without dilatation; an anomalous vessel is visualized between the cortical surfaces of the slit; the absence of pellucid septum. After diagnosis through CT or MRI, anamnesis and clinical picture of the patients may be based on possible specific and appropriate treatments. Magnetic resonance imaging allows the specific characteristics of schizencephaly to be more accurately identified, including small or associated malformations, which are not observed with other techniques. Delays in neuropsychomotor development, changes in muscle tone, postural changes and the adoption of pathological patterns are also evidenced in these patients [5].

Radiological Findings

Magnetic resonance imaging effectively identifies the specific characteristics of schizencephaly, they are small or associated malformations, which are not observed with other techniques. It is not possible to identify a particular zone of the location of the slits that can vary from one patient to another but are usually found mainly in the parietal lobe, it is valid to classify the image with the correlation of the clinical presentations [2,4].

Treatment

The fundamental treatment in patients with disorders of neuronal migration is not of the etiological type, but symptomatic. The therapeutic orientation should agree with the manifestations presented by the patient that are generally proportional to the type, extent and severity of the lesions.

The conventional and specific treatment for this type of disease is physiotherapy. Because the problem is exactly in the cerebral cortex, it is not possible to be based on surgeries.

The treatment is based on promoting the neuropsychomotor development of the child, minimizing the pathological patterns present, promoting stretching, motor activities, sensorial stimulation with the aim of adjusting tone and with alternation in the posture for the mechanical stimulus that provided even more adequate tone, protection reactions. The stimulation and encouragement of the movements and reactions typical of the patient's age range will provide the integration in their activities and ensure an improvement in their physical, social and emotional state.

Each patient develops according to the maturation of their central nervous system, together with the action of the environment in which they live, it should be considered a sequence to be followed so that in the first phase the early stimulation with the physiotherapy to develop the best possible.

Frequent seizures in these patients with schizencephaly are treated with conventional anticonvulsants. Epileptic seizures may become resistant to drugs and often new antiepileptic drugs are required according to multidisciplinary medical guidance [5].

Final Considerations

It is expected that data from this study may contribute to the performance of health professionals in the area of congenital malformations. And that may encourage new studies on this subject, to deepen the knowledge about congenital malformations especially on the subject addressed with the objective of identifying the causes and risk factors for the implementation of measures aimed at the prevention and care of patients affected and their family.

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