

Pallister-Hall Syndrome: A case Report

L de La Ossa, L Moscote-Salazar, K Kafury-Benedetti

Citation

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Abstract

Pallister-Hall Syndrome (PHS) is characterized by hypothalamic hamartoma, polydactyly, and other malformations. This disorder is inherited in an autosomal dominant pattern. We report a case of child of 4 months managed in our neurosurgical service.

INTRODUCTION

In 1980, Hall and colleagues first described a syndrome characterized by congenital hypothalamic "hamartoblastoma," hypopituitarism, imperforate anus, postaxial polydactyly, and various visceral anomalies. The importance of recognizing Pallister-Hall syndrome should be stressed not only for clinical management but also for future genetic counseling. (1)

CASE REPORT

A 4-month-old male patient with sickle cell trait, who was being evaluated for managed by convulsions. Physical examination was found to limb and genital abnormalities. An MR image of the brain was obtained that revealed a large sellar and suprasellar mass neurosurgical resection of the mass was scheduled. Partial resection was performed now in satisfactory postoperative ambulatory control.

DISCUSSION

The development of hamartomas are non-neoplastic lesions, which are essentially normal in structure, but with ectopic localization. Hamartoma is the Greek 'hamartia' which means "Error" and the term was first coined by Albrecht in 1904 and later refined by Willis. Its incidence is one in every 50,000 to 100,000 tumors of the nervous system. These lesions have an appearance reminiscent of the normal gray matter, and contains varying proportions of white and gliar and nerve bundles. It is considered a congenital lesion that originates from the hypothalamic embryonic plate between 35 and 45 days of gestation. (2)

Arita et al described two subtypes:

1. Hamartoma hypothalamus associated precocious puberty
2. Hamartoma intrahypothalamico associated with gelásticas crisis or crisis of laughter.

CLINICAL

The clinical features are seizures or precocious puberty. According to different authors seizures are about 65%, and equally precocious puberty. In relation to convulsive table can be given from the neonatal period through adolescence, with a range that goes from the first day of life up to 27 years. The type of crisis are the most frequently reported gelásticas by 92%, consisting of brief attacks of laughter in that stereotypical way the patient does not produce happiness, but on the contrary, anxiety or fear, only a crisis are considered gelásticas neocortical origin. Seizures accompanied by other components such as semiological: autonomic phenomena, automatisms, left - vu leaves - lived. Epigastric auras, crying, motor symptoms. (3) .Some patients do progress to symptomatic generalized tables of crisis type absence, atonic, tonic, tonic clonic seizures. One potential route of spread of the crisis is mamilotalamico tract, which connects with the anterior nucleus of the thalamus, and this is projected toward the turn of the previous cingulate. The hamartomas that make contact with the mamillars body are associated with also manifestations of the temporal lobe, and those that are located medial mind are manifestations of frontal lobe .

Precocious puberty occurs apparently because these lesions have cells that produce Gn-RH, which work independently of neurophysiological normal. Described cases of hamartomas are reported to secrete corticotropin releasing hormone with high serum levels of CRH and ACTH. The hypothalamic hamartomas can be accompanied by

anomalies such as agenesis of the corpus callosum, polymicrogyria, hemispheric heterotopia, mental retardation, visual disturbances, and psychological disorders associated malformations such as Pallister syndrome - Hall⁶⁴. This syndrome was first described in 1980 as a pleiotropic disorder of human development, characterized by the presence of hypothalamic hamartoma, polydactyly, imperforate anus and genitals pequeños. Presents an autosomal dominant and has been mapped to chromosome 7p13. This syndrome has been described as lethal, due to hypopituitarism. Has been reported that appear to be minor variants of this syndrome. (4,5)

RADIOLOGY

Are lesions in the brain are manifested Isodense TAC and not reinforced with intravenous contrast in MRI brain isointense are in T1 and T2 and not reinforce the contrast.

TREATMENT

Medical treatment is used for endocrine drugs using the GnRH analogues in order to reduce its secretion. Some authors have shown that medical treatment not only improves the picture but endocrinological seizures. (6)

Surgical treatment is disappointing, because they are lesions that have the same aspect of normal brain, and not easily distinguished from this and therefore are almost always incomplete resections or biopsies performed, several types of approaches have been used as its front, peritumoral, transcallosal, transcortical. Another surgical treatment is the Gamma-knife radiosurgery generates fewer side effects and improve the crisis 50% ⁶⁶.

Radiotherapy and chemotherapy housed poor results.

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Author Information

Leonardo Dominguez de La Ossa, MD

neurosurgeon Chief of Neurosurgery, Cartagena University

Luis Rafael Moscote-Salazar, MD

Resident of Neurosurgery, Cartagena University

Kalil Kafury-Benedetti, MD

Neurosurgeon, Cartagena University