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Case Report

Atypical Congenital Meduloblastoma: Case Report

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Abstract

Medulloblastomas are WHO grade 4 embryonal neuroepithelial tumors that typically occur in the posterior fossa in the pediatric population. They represent 68% of embryonal tumors. The study of congenital cases is scarce due to the rarity of reported patients. There is also no specific bibliography, however histologically and molecularly they can be classified. On the other hand, due to the young age of these patients, they are difficult to manage and have a higher risk of complications in the treatment. In addition to this, patients are born with alterations in the development of the central nervous system due to chronic intracranial hypertension caused by said tumor and have little functional development. The objective of this study is to teach us to recognize the diagnosis early, the evolution of the disease to lead to better treatment for these patients in the future.

***Keywords:** Medulloblastoma, Congenital brain tumors (CBT), Treatment*

Introduction

Congenital brain tumors (CBT) represent less than 2% of all pediatric brain tumors, however they have a mortality rate of 5 to 20% and have a generally poor prognosis (5). They are detected within the first 6 months of life (9). Teratoma appears to be the most common, followed by astrocytomas (1).

Medulloblastoma (MB) is a neuroepithelial, embryonic tumor that corresponds to the most common malignant pediatric brain tumor, which represents around 25% of all intracranial neoplasms and around half of posterior fossa tumors. It has a maximum incidence peak between 3 and 7 years (1.7).

Congenital medulloblastoma (CMB) is extremely rare. It is usually diagnosed postnatally, being a highly aggressive neoplasm (1). In the cerebellar location it represents only 8.3% of intracranial tumors during the neonatal period (10). There is little information about these congenital tumors.

We present a case of congenital medulloblastoma operated on in a highly complex center specialized in pediatric pathology, which was initially suggested as a probable vascular pathology, due to its atypical radiological presentation. It is described as the first case reported in this hospital. It is important to expose this type of extremely rare neoplasms, so that in the future, along with other reported cases, it can be detected early and a therapeutic scheme can be protocolized.

Case Presentation

Term newborn with prenatal ultrasound checks revealing macrocephaly without apparent cause. After birth by cesarean section, somatometric measurements were taken, highlighting a head circumference (HC): 44cm. On the first day of life, the patient awake, spontaneously ventilates primitive reflexes present. Due to this, a transfontanelar ultrasound was performed, revealing severe hydrocephalus. At 10 days of age, he was referred to the National Children's Health Institute of San Borja for care. Upon admission, a magnetic resonance imaging (MRI) without contrast was performed to expand the study, finding a poorly defined cystic lesion in the pineal region with displacement and dilation of the third ventricle and a diffuse intraparenchymal image in the cerebellar vermis hemispheres and brain stem, which led to a marked obstructive hydrocephalus (Figure 1,2,3).

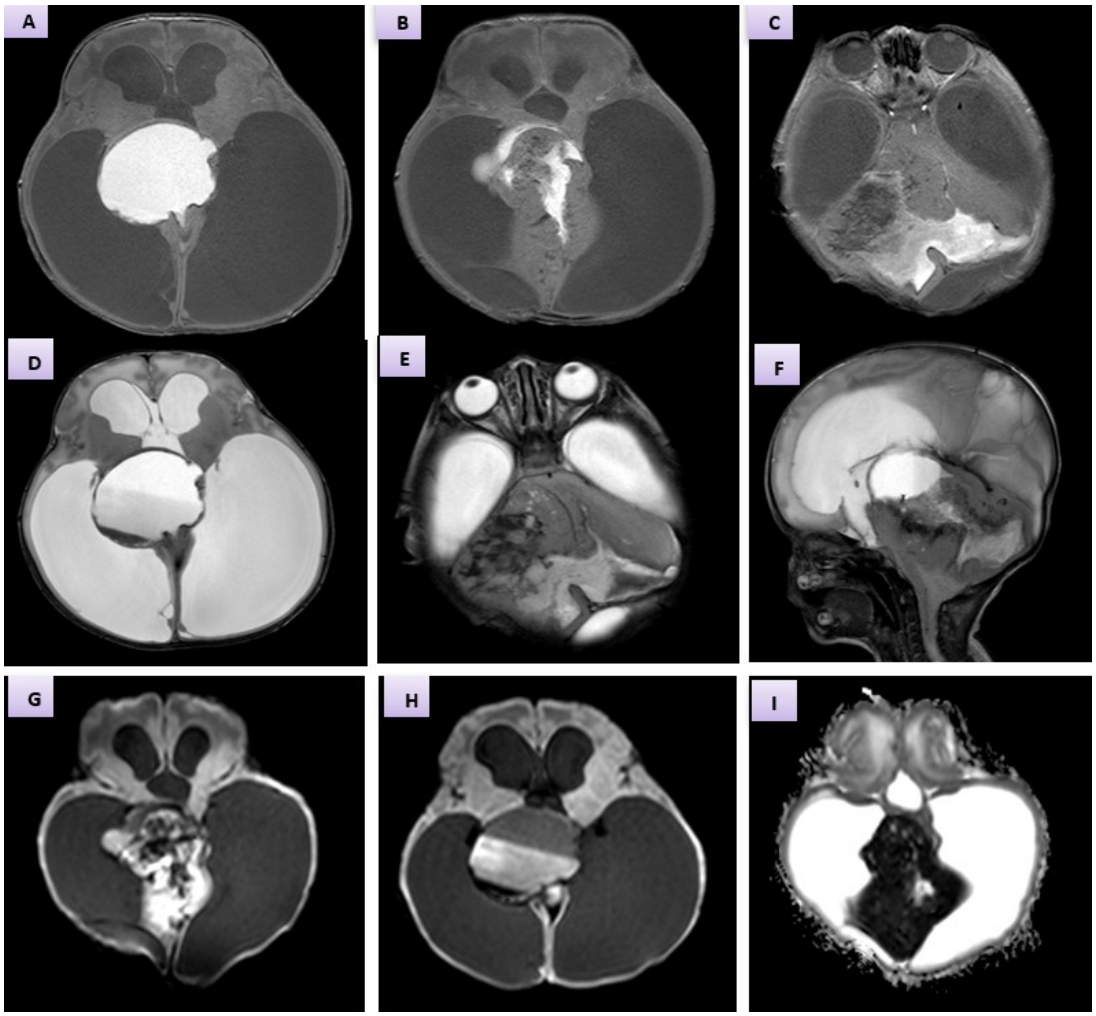

Figure 1: *A, B and C: MRI images T1 sequence without contrast where homogeneous hyperintensity is seen in the pineal region with suggestion of a cyst with blood content and as it reaches the posterior fossa, heterogeneous hyperintensity is observed at the level of the diencephalon with loss of the anatomy, it infiltrates the upper part of the brain stem and cerebellar hemispheres. Likewise, extreme supratentorial hydrocephalus is seen with little cortico-subcortical neural tissue. E, D and F: T2 sequence, cystic content is corroborated with a level inside and absence of the fourth ventricle. G, H and I: Diffusion images and ADC map where it can be seen that solid images of the trunk and cerebellum restrict diffusion. FIGURE 1: A, B and C: MRI images T1 sequence without contrast where homogeneous hyperintensity is seen in the pineal region with suggestion of a cyst with blood content and as it reaches the posterior fossa, heterogeneous hyperintensity is observed at the level of the diencephalon with loss of the anatomy, it infiltrates the upper part of the brain stem and cerebellar hemispheres. Likewise, extreme supratentorial hydrocephalus is seen with little cortico-subcortical neural tissue. E, D and F: T2 sequence, cystic content is corroborated with a level inside and absence of the fourth ventricle. G, H and I: Diffusion images and ADC map where it can be seen that solid images of the trunk and cerebellum restrict diffusion.*

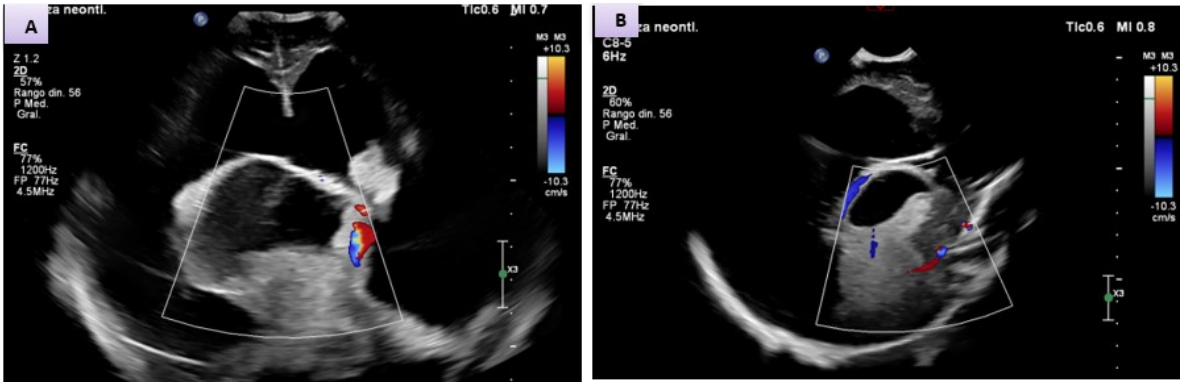

Figure 2: *Doppler scan of tumor mass; A: cystic lesion in which no blood flow is observed inside, B: solid lesion without flow, images not compatible with vascular lesion.*

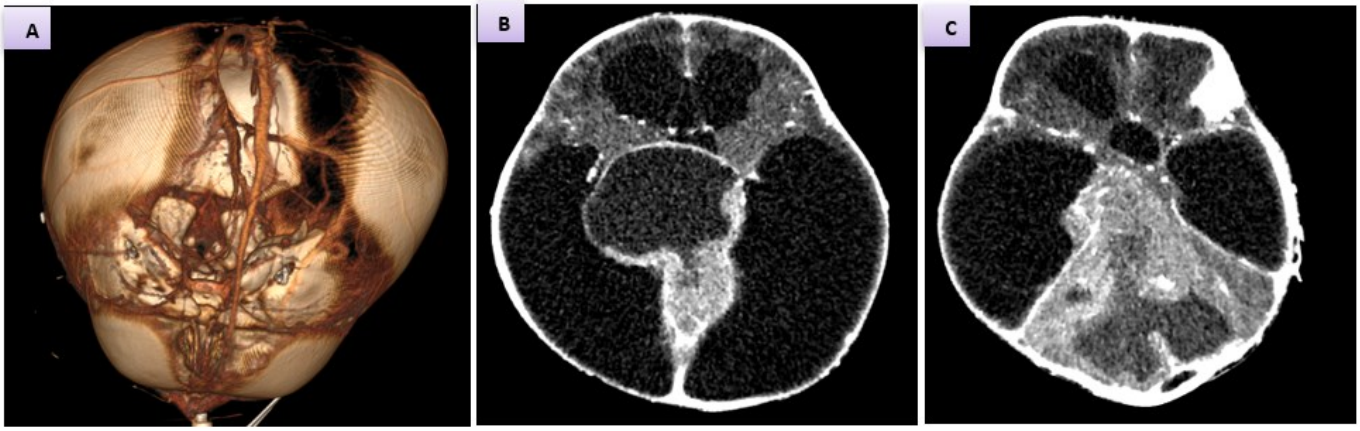

Figure 3: *A: Brain angiotomography, without visualization of congenital vascular lesion. B and C: Brain tomography with contrast, ring enhancement is observed in the cystic lesion and heterogeneous uptake in the solid tumor region in the posterior fossa.*

The presence of a vascular malformation was ruled out and a tumor of congenital embryonal origin was suspected. The hydrocephalus progressively increased, so a surgical intervention had to be performed: placement of a ventriculoperitoneal shunt + medial suboccipital craniectomy for cytoreduction, decompression and sampling of the lesion. Surgical intervention was decided on the 21st day of life. Intraoperative findings were observed: tumor of indurated consistency, mixed gray, violaceous and whitish placement, highly vascularized without extensive infiltrative cleavage plane, normal parenchymal tissue was observed intertwined with tumor, the cyst opened, which had chocolate blood content (Figure 4). As much of the tumor as possible was removed and sent to pathology for study. The anatomopathological result of the surgical specimen revealed Classic medulloblastoma (Corroborated in 2 highly complex centers: INSNSB and INEN), immunohistochemistry revealed Ki67: 30-40%, P53 Diffuse Positive, B-Catenin: Positive.

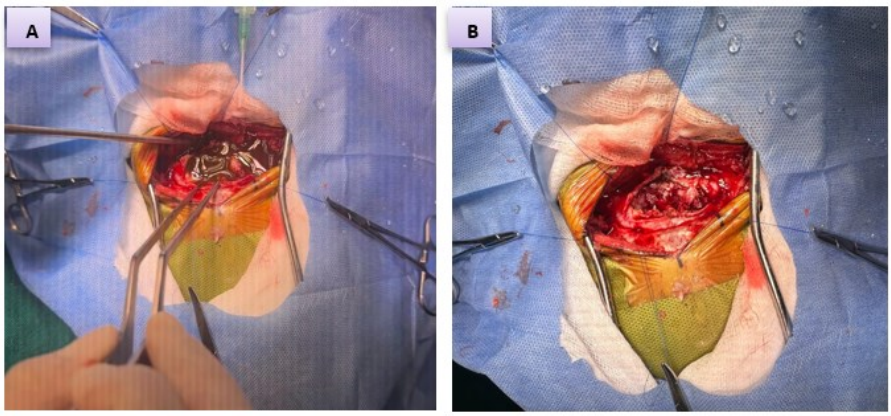

Figure 4: *Intraoepratorial images after dural opening in an infratentorial supracerebellar approach, A: chocolate cyst content is observed from the pineal region. B: mixed heterogeneous indurated bleeding tumor that appears immediately after durotomy.*

The patient was discharged from the neonatal ICU after 8 days. During hospitalization, he presented respiratory failure due to hospital-acquired pneumonia, which ceased after completing his antibiotic therapy regimen and was finally discharged after 7 days (52 days). age), his status was; awake, no crying, ventilating spontaneously, quadriparesis, hyperreflexia, tremor with nasogastric tube feeding.

During follow-up, MRI showed tumor recurrence with spinal metastasis. A multidisciplinary medical meeting was held with the services of medical oncology, radiotherapy and palliative care, where the decision was made that due to poor prognosis and age (<3 years) it is not amenable to surgical or adjuvant treatment. (Figure 5).

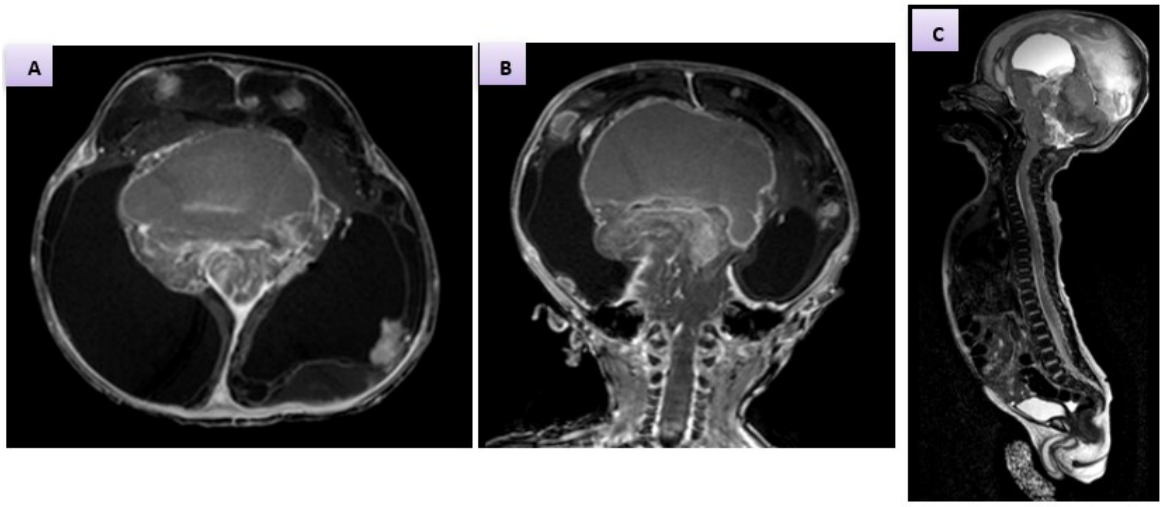

Figure 5: *A& B: Tumor regression is observed with greater supratentorial growth and spinal invasion (C).*

Discussion

Congenital tumors of the central nervous system (CNS) are classified as “absolutely congenital” (detected at birth) and “probably congenital” (detected within 6 months of age) (9).

The incidence of congenital central nervous system (CNS) tumors has increased over time due to improved prenatal care and routine ultrasounds that allow for greater intrauterine detection. In the past, the prognosis associated with these tumors has been poor, with an overall survival of only 28%. The outcome is related to multiple factors, including size, location, and WHO grade (6).

The widespread use of prenatal ultrasound combined with the Doppler technique has significantly increased the diagnosis of brain tumors in utero. The majority of congenital brain tumors (60%) are supratentorial in terms of location and originate in the pineal gland, the suprasellar area and the cerebral hemispheres (5).

Medulloblastomas are WHO grade 4 embryonal neuroepithelial tumors that typically occur in the posterior fossa (cerebellum or dorsal brainstem) in the pediatric population. In the cerebellum, they usually arise in the vermis near the apex of the roof of the fourth ventricle (fastigium). They account for 68% of embryonal tumors (12).

The overall 5-year survival for medulloblastoma is approximately 75%, however, long-term treatment-related morbidity remains a major concern (1).

The clinical history is usually brief (6-12 weeks). The predilection for the posterior fossa predisposes to early obstructive hydrocephalus. Usual presentation of symptoms: Infants with hydrocephalus may present with irritability, lethargy, or progressive macrocephaly. Spinal drops may cause back pain, urinary retention, or leg weakness. Common signs: papilledema, truncal and appendicular ataxia, nystagmus.

Classification of medulloblastomas based on a combination of molecular studies combined with histopathological findings is recommended and provides optimal prognostic information. Molecularly defined MDBs are largely associated with specific morphological patterns. (12)

Brainstem invasion generally limits complete staging of surgical excision: all patients should be evaluated for verification of spinal metastasis (imaging of the entire neuroaxis with contrast - brain + spine, preoperatively if possible), and postoperatively with LP. for CSF cytology. (12)

If a preoperative MRI of the spine was not performed, it is recommended that an MRI be performed more than 2 weeks after surgery to avoid false-positive results. (12)

To confirm cerebrospinal fluid (CSF) dissemination at the time of diagnosis, CSF cytology tests are usually performed 2 to 4 weeks after surgery. The differential diagnosis of BML is atypical teratoid/rhabdoid tumor, ependyma, pilocytic astrocytoma, and choroid plexus papilloma. (12)

Current treatment for medulloblastoma consists of maximal safe resection, chemotherapy, and CSI. The surgical standard is the maximum safe resection which was established based on several studies supporting the relationship between the extent of resection and survival rate. Aggressive resection is not recommended where high neurological morbidity is expected, such as when dealing with the brainstem (2).

This case leads to great clinical interest, due to its rarity. We currently have few references of this type of tumor in the neonatal period. For all of the above, it is necessary to rule out the presence of tumors that could worsen the patient's prognosis and survival in the study of congenital hydrocephalus. Our patient returned to his place of origin with his parents, under palliative management.

Conclusions

It is necessary that, in our environment, despite the scarcity of resources, and not having in utero surgeries, it is necessary after birth to study the cause of congenital hydrocephalus, and if the origin was tumor, establish a timely treatment scheme. treatment, both surgical and adjuvant, that improves the survival of these patients, and thus is included with future cases for a larger study.

Conflict of Interest

None.

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