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CLASSIC FINDING OF DANDY-WALKER MALFORMATION- CASE REPORT

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ABSTRACT

Summary: The frequency and importance of posterior fossa evaluation has increased significantly in the last 20 years due to advances in neuroimaging. Today, conventional and advanced neuroimaging techniques allow a detailed assessment of the complex anatomical structures within the posterior fossa. A wide spectrum of congenital anomalies (anomalies due to an alteration of the primary developmental program caused by a genetic defect) and due to the breakdown of a structure that had a potential for normal development has been demonstrated.

Knowledge of the spectrum of congenital anomalies of the posterior fossa and its well-defined diagnostic criteria is essential for optimal therapy, an accurate prognosis, and correct genetic counseling.

Objective: Analyze the importance of the precise diagnosis of congenital anomalies of the posterior fossa, emphasizing Dandy Walker syndrome, through a clinical case.

Design: Prospective, observational in a single center.

Methodology: This is a systematic review, Dandy Walker syndrome, detailing its clinical characteristics and short-term complications. The information and images obtained belong to the medical personnel in charge of the case, whose reinforcements are provided by the Excel, Word and JPG statistical package.

KEYWORDS : syndrome, Dandy Walker, malformation.

INTRODUCTION

Over the past two decades, significant advances in pre- and postnatal neuroimaging techniques, the development of next-generation genetic sequencing, and the increase in animal model research have resulted in a more precise definition and classification of abnormalities. congenital disorders of the posterior fossa and a better understanding of its pathogenesis.

Congenital anomalies of the posterior fossa can be the result of inherited (genetic) or acquired (disruptive) causes

A malformation is defined as a congenital morphological abnormality of a single organ or part of the body, due to a change in the primary developmental program caused by a genetic defect.

Genetic mutations that because malformations can be de novo (that is, new to the affected child, rather than being present or passed on by the parents).

Accurate diagnoses of these complex anomalies are of utmost importance for three main reasons: -Inheritance pattern and risk of recurrence. - Compromise of other systems. -Prognostic implications for the disease, the child and his family.

In this article we will talk about Dandy Walker disease: it is a congenital anomaly that usually occurs in childhood and is characterized mainly by the presence of hydrocephalus, changes in the development of the cerebellum and dilation of the fourth ventricle that leads to an enlargement of the fossa later.

The etiology of this syndrome is very heterogeneous, in some cases it is described as the cause of a recessive gene and in others as the cause of exposure to measles, cytomegalovirus, toxoplasmosis, alcohol, and isotrethionoin in the first trimester of pregnancy.

Dandy Walker syndrome is more common in women, with a ratio of 3: 1 and its incidence is estimated between 1 in 25,000 and 1 in 30,000. This syndrome is included within the cases of hydrocephalus. Neuroimaging plays a key role in the diagnosis of posterior fossa abnormalities, and the challenge for the neuroradiologist is to provide the physician with accurate and up-to-date diagnostic information.

Case Presentation

The case is presented with the consent of the couple for its description and information on the results of the tests carried out on the newborn.

It is a 42-year-old pregnant woman, with no significant pathological or surgical history.

no toxic habits, married, obstetric gynecological history. Gestures: 4, Births: 4, Living children: 4, Abortions none. In her current pregnancy, no prenatal controls were performed.

At week 31, she suddenly presented contraction-type abdominal pain, so she came to our health home, where a pregnant patient was evaluated, with vital signs within normal parameters, vaginal examination: dilated 9 erased 80%, proceeding from emergency cesarean section, and a live male newborn is received. One minute after the birth of the newborn with Apgar 2, they proceed to resuscitation, which improves their Apgar to 6, however; Due to poor ventilatory mechanics, intubation was carried out, achieving saturations of 90%.

Upon physical examination, blood pressure: 130 / 70mmHg, Temperature of 36 °C, radiant cradle, HR: 115 bpm, FR: 29 rpm; Eutrophic, not very reactive, regular general appearance, lungs: abolished vesicular murmur, heart

arrhythmic heart sounds, which is why chest and abdomen radiography is requested (Photo 1). Evidence of left pneumothorax, pneumomediastinum and pneumop eritoneum.

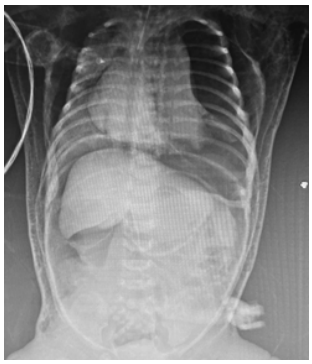

Photo 1: X-ray of the chest and abdomen showing a candle sign (thymus elevation), continuous diaphragm sign: caused by air located posterior to the pericardium. (left pneumothorax, pneumomediastinum, and pneumop eritoneum).

In extension tests: leukocytes 13.00 Neutrophils: 86%, Hemoglobin 15.30 g / dl, procalcitonin 2 ng / ml, creatinine 1.2 ng / dl.

A chest tube was placed to improve his respiratory distress, however, the patient remained stationary, so a transfontanelar ultrasound was requested (Photo 2).

Where the dilation of the fourth ventricle is observed.

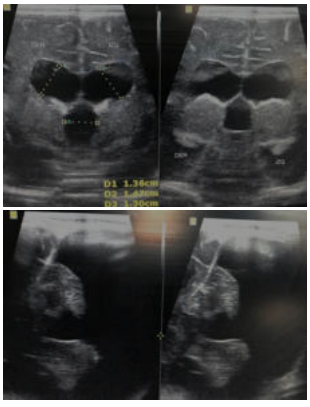

Photo 2-3: (Dandy Walker syndrome): Transfontanellar ultrasound showing dilation of the fourth cystic ventricle

Once diagnosed with pneumothorax (resolved), pneumoperitoneum, and Dandy Walker Syndrome, the newborn remained with torpid evolution, and died after 12 days of hospitalization.

DISCUSSION

Dandy-Walker Malformation: Most common posterior fossa malformation and typically occurs only sporadically, with a low overall risk of recurrence (1% -5%). It can be isolated or it can occur as part of chromosomal abnormalities.

Most patients with this entity present before 1 year with signs and symptoms of increased intracranial pressure. Macrocephaly is the most common manifestation, affecting 90-100% of children during the first months of life. Today, the diagnosis of this pathology is carried out prenatally in an increasing number of patients.

Limited evidence shows that normal lobulation of the cerebellar vermis and the absence of associated brain abnormalities (particularly callosal dysgenesis) may be prognostic for normal cognitive function.

The key neuroimaging features are hypoplasia or occasionally agenesis of the cerebellar vermis, cystic-appearing dilation of the fourth ventricle, which can consequently fill the entire posterior fossa. Both findings are constantly present and are necessary for diagnosis. The posterior fossa is usually enlarged and the tentorium, as well as the torcular and transverse sinuses, are elevated.

In 30% -50% of individuals there may be additional malformations, such as dysgenesis or agenesis of the corpus callosum, occipital encephalocele, and heterotopia. Hydrocephalus is associated in approximately 90% of patients. Cerebrospinal fluid (CSF) flow studies can be added to better understand complex hydrodynamic changes. If aqueductal stenosis or occlusion is shown on CSF flow studies, placement of a cystoperitoneal shunt is not sufficient and placement of a ventriculoperitoneal shunt is required.

Our case presented was a moderate premature newborn, as the only prenatal history of an elderly mother, without being the cause of her pathology. The newborn had respiratory distress due to the presence of pneumothorax, pneumome diastinum and pneumoperitoneum (without being able to find a probable cause of intestinal perforation), likewise due to its stationary evolution, a significant dilation of the 4th ventricle with cystic appearance was evidenced in the transfontanellar ultrasound, a compatible finding with the Dandy Walker malformation. The ideal is to be able to diagnose it in the gestational stage from week 18; however, our patient did not have any control of her pregnancy.

Due to the rarity of the case presented, it is presented to him; without forgetting the importance of early diagnosis by prenatal ultrasonography.

CONCLUSION

Congenital anomalies of the posterior fossa represent a wide variety of disorders that include both malformations and abnormalities.

Dandy Walker syndrome is one of them, its cause is still unknown, however, ultrasound can reliably diagnose it, where dilation of the fourth ventricle can be seen as cysts, and hypoplasia of the cerebellar vernix.

Our case was associated with several catastrophic pathologies, such as pneumothorax, pneumoperitoneum that, together with the finding of Dandy Walker Syndrome, led to his death.

We intend to emphasize the importance of prenatal controls in order to facilitate its long-term management.

CONFLICT OF INTERESTS

The authors declare that they have no conflict of interest.

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