

**Clinical and morphological aspects in children affected with  
hydranencephaly: an overview****Aspectos clínicos e morfológicos em crianças afetadas com hidranencefalia:  
uma visão geral**

DOI:10.34115/basrv4n3-056

Recebimento dos originais: 10/04/2020

Aceitação para publicação: 25/05/2020

**Simone Martins dos Santos**

Formação acadêmica mais alta: acadêmica de enfermagem

Instituição: faculdade Mauricio de Nassau - UNINASSAU

Endereço: BR 104, KM 68, n 1215 - Agamenon Magalhães, Caruaru - PE, 55000-000

E-mail: symone-saopaulo@hotmail.com

**Michael Gabriel Agustinho Barbosa**

Formação acadêmica mais alta: acadêmico de biomedicina

Instituição: centro universitário do vale do Ipojuca-UNIFAVIP/WYDEN

Endereço: Av. Adjar da Silva Casé, 800 - Indianópolis, Caruaru - PE, 55024-740

E-mail: michaelgabrielssc1210@hotmail.com

**Joas Francisco da Silva**

Formação acadêmica mais alta: acadêmico de farmácia

Instituição: centro universitário do vale do Ipojuca-UNIFAVIP/WYDEN

Endereço: Av. Adjar da Silva Casé, 800 - Indianópolis, Caruaru - PE, 55024-740

E-mail: joasfrancisco123@hotmail.com

**Emanuela Ingridy da Silva**

Formação acadêmica mais alta: acadêmica de biomedicina

Instituição: Faculdade Mauricio de Nassau- UNINASSAU

Endereço: Antônio Carvalho de Souza, 295- Estação velha, Campina Grande- PB

E-mail: emanuela9530@gmail.com

**Ana Paula Barbosa de Souza**

Formação acadêmica mais alta: acadêmica de enfermagem

Instituição: centro universitário do vale do Ipojuca-UNIFAVIP/WYDEN

Endereço: Av. Adjar da Silva Casé, 800 - Indianópolis, Caruaru - PE, 55024-740

E-mail: memeleticia1234@hotmail.com

**Anderson José da Silva**

Formação acadêmica mais alta: acadêmico de enfermagem

Instituição: centro universitário do vale do Ipojuca-UNIFAVIP/WYDEN

Endereço: Av. Adjar da Silva Casé, 800 - Indianópolis, Caruaru - PE, 55024-740

E-mail: andezil@hotmail.com

**Thays Wany Silva Carvalho**

Formação acadêmica mais alta: acadêmica de biomedicina

Instituição: centro universitário do vale do Ipojuca-UNIFAVIP/WYDEN

Endereço: Av. Adjar da Silva Casé, 800 - Indianópolis, Caruaru - PE, 55024-740  
E-mail: thayswanycarvalho@hotmail.com

**Tatiana Acioli Lins**

Formação acadêmica mais alta: Graduada em Licenciatura Plena Em Educação Física pela Universidade de Pernambuco (2003). Especialização em Avaliação da Performance Humana pela Universidade de Pernambuco. Mestre em Educação Física pelo mestrado Associado da UPE com a UFPB. É pesquisador do Centro Nacional de Excelencia Esportiva(CENESP).

Instituição: centro universitário do vale do Ipojuca-UNIFAVIP/WYDEN  
Endereço: Av. Adjar da Silva Casé, 800 - Indianópolis, Caruaru - PE, 55024-740  
E-mail: tatiana.lins@unifbv.edu.br

**ABSTRACT**

Objective: To demonstrate a general view on the clinical and morphological aspects in children with Hydranencephaly. Methodology: This is a study that uses the systematic review as a method. Data collection took place in September 2019 through the SciELO and EBESCO databases with the following descriptors: "hemihydranencephaly", " Rare diseases ", 'malformation'. For the selection of articles, the following were used: inclusion criteria: articles in Portuguese, English, full articles that portray the theme related to the literature review and articles published and indexed in the referred databases in the period from 2007 to 2019. Results and Discussion: Hydranencephaly is a condition that in Most are lethal with exceptions, this malformation of the central nervous system occurs after the closure of the neural tube, defined as total or partial absence of cerebral hemispheres, the entire cortex that would be irrigated by the carotids is nonexistent. however, the brainstem, diencephalon and part of the occipital cortex, irrigated by the posterior cerebral arteries, are formed and may exhibit electrical activity. In some cases they have severe neurological sequelae, psychomotor disability, visual impairment, deafness and progressive macroencephaly. Conclusion: Patients with this rare condition are subjected to several surgeries and antibiotic therapies for prolonged periods in an attempt to provide the best survival for the patient. irrigated by the posterior cerebral arteries, are formed and may exhibit electrical activity. In some cases they have severe neurological sequelae, psychomotor disability, visual impairment, deafness and progressive macroencephaly. Conclusion: Patients with this rare condition are subjected to several surgeries and antibiotic therapies for prolonged periods in an attempt to provide the best survival for the patient. irrigated by the posterior cerebral arteries, are formed and may exhibit electrical activity. In some cases they have severe neurological sequelae, psychomotor disability, visual impairment, deafness and progressive macroencephaly. Conclusion: Patients with this rare condition are subjected to several surgeries and antibiotic therapies for prolonged periods in an attempt to provide the best survival for the patient.

**Keywords:** hemihydranencephaly, malformation, rare diseases.

**RESUMO**

Objetivo: demonstrar de maneira geral uma visão sobre os aspectos clínicos e morfológicos em crianças realizadas com a Hidranencefalia. Metodologia: é um estudo que utiliza a revisão sistemática como método. Uma fila de dados ocorreu durante o período de setembro de 2019 através das bases de dados SciELO e EBESCO com os seguintes descritores: "hemihidranencefalia", "Doenças raras", "má formação", para seleção dos artigos foram usados os seguintes critérios de inclusão: artigos em português, inglês, artigos completos que retratam o tópico referente à revisão de literatura e artigos publicados e indexados nos referimos a bancos de dados no período de 2007 a 2019. Resultados e Discussão: A hidranencefalia é uma condição

que em Sua maioria é letal havendo exceções, essa malformação do sistema nervoso central ocorre apos ou datação do tubo neural, definido como ausência total ou parcial dos hemisférios cerebrais, todo ou córtex que seria irrigado carotídeo e inexistente. Enquanto isso, ou tronco cerebral, diencéfalo na parte do córtex occipital, são formadas cascas irrigadas das artérias cerebrais posteriores e, portanto, pode haver atividade elétrica. Em alguns casos, existem sequelas neurológicas graves, deficiências psicomotoras, deficiência visual, surdez e macrocefalia progressiva. Conclusão: pacientes com essa condição rara estão sujeitos a várias cirurgias e antibióticos por períodos prolongados, na tentativa de proporcionar sobrevida ao paciente.

**Palavras-chave:** hemi-hidrananencefalia, malformação, doenças raras.

## 1 INTRODUCTION

Hydranencephaly is a rare condition, where patients have an absence of brain hemispheres that are replaced by bags filled with cerebrospinal fluid. This pathology is considered as a serious expression of the brain's nutritional failure before birth, with consequent severe and permanent neurological deficiencies. In hydranencephaly, the brain is reduced compared to normal and has a thin amount of nervous tissue responsible for basic life-sustaining activities such as breathing and heartbeat. Newborns affected by this disease may present with a large head (macrocephaly) or small head (microcephaly). Over the months the tendency is for the head to grow progressively.(GARDEA et al., 2015).

The literature has yet to define which factor is associated with the development of the disease, but it is believed to be due to infectious processes, such as syphilis, toxoplasmosis, infectious hepatitis or influenza; of toxic origin, such as carbon monoxide poisoning; of traumatic origin, in attempts at abortion by mechanical means, all are associated with pathology. Treatment is symptomatic and supportive. Life expectancy for people with hydranencephaly is minimal. These small patients die in the first months of life. However, in rare cases, they can survive for several years. The incidence of registered cases is less than one in 10,000 live births (CASAGRANDA et al., 2016.)

When there is an association in the presence of congenital malformations affecting other organs or systems 57.5% of newborns had more than one congenital defect linked to the congenital malformation of the CNS. The most prevalent site is craniofacial, followed by orthopedic, cardiovascular, genitourinary and gastrointestinal congenital malformations. Approximately 21% of congenital malformations involve the central nervous system (CNS), constituting one of the most common birth defects, which may occur alone or in combination with other malformations of this or another segment (BARROS et al., 2014).

Patients with this pathology are often submitted to ventricular shunt with numerous complications, these complications lead to the need for multiple surgeries for revisions and / or changes in the shunt systems and antibiotic therapies for prolonged periods (AMARAL et al., 2014).

There is no curative treatment for these patients, and life expectancy usually ends up until the age of three. Some patients exceed this average age of expectation and, in all cases, require palliative and therapeutic treatment, as well as physical therapy, support measures and multidisciplinary support (BARRÓN et al., 2016).

The multidisciplinary team plays a fundamental role in the care provided to the patient, as well as in accepting the diagnosis and helping to live with this rare disease. Thus, it develops comprehensive care for patients and families, through active and qualified listening in order to reduce anxiety due to fear of the disease and the future (SANTOS et al., 2018).

One of the methods used to obtain the diagnosis is obstetric ultrasound, which has great sensitivity in the screening of fetal malformations of the CNS, especially with the constant improvement and mastery of specialized methods, for example, Doppler and volumetric ultrasound (3D / 4D), which helps to establish itself as a modality of choice in this routine. Complementary to these methods, magnetic resonance imaging may provide support for better perinatal care (BARROS et al., 2014).

In view of the above, this study aims to demonstrate in a general way a view on the clinical and morphological aspects in children affected with hydranencephaly.

## **2 METHODOLOGY**

This is a study that uses literature review as a method. The search for publications in the Virtual Health Library (VHL) was carried out from 22 and 23 September 2019, using the Scientific electronic library online database (SciELO) and Elton B. Stephens Company (EBESCO). This database was selected because it is understood that it has a vast collection of scientific papers published on the health sciences area with renowned journals and that they can cover closer geographic locations. Health descriptors were used according to the DeCS: “hemihydranencephaly”, “rare diseases” and “malformation”, using the Boolean descriptors “and and or”, being selected as the time period from 2007 to 2019. The inclusion criteria were defined: scientific works that addressed the morphological and pathological aspects of patients with rare diseases and associated comorbidities, with emphasis on those with an emphasis on hydranencephaly, published in Portuguese, English and Spanish, with full, available and free

text. Exclusion criteria were defined: not meeting the inclusion criteria or being the same as another job already found. After reading and initially evaluating the abstracts, publications that met the criteria defined for the research were selected and read in full. After reading the publications in full, the themes were analyzed and organized: What are the main rare diseases cited in the literature that are associated with hydranencephaly? What morphological factors justify the association between rare diseases and the development of hydranencephaly in children?

### 3 RESULTS AND DISCUSSIONS

According to the inclusion and exclusion criteria pre-established in the study, the sample consisted of eight articles, described according to table 1:

**Table 1** - Distribution of references included in the study, according to the authors, year of publication, title and objectives.

| <b>AUTHOR / YEAR</b>      | <b>TITLE</b>  | <b>Methodology</b>   | <b>Results</b>  |
|---------------------------|---|--|---|
| <b>AMARAL et al, 2014</b> | Choroid plexus endoscopy coagulation in the primary treatment of hydranencephaly and external hydrocephalus | Describe the neuroendoscopic coagulation technique of the choroid plexus and publicize it as a primary treatment option for hydranencephaly and external hydrocephalus | Hydranencephaly and extreme hydrocephalus are extremely serious and complex pathologies from a medical and socioeconomic point of view. Patients with these pathologies are frequently submitted to ventricular bypass with numerous complications (CSF fistula, infections of the central nervous system, dysfunction of the bypass system and bedsores on the reservoirs of the ventriculoperitoneal bypass). |
| <b>BARROS et al, 2012</b> | Malformations of the central nervous system and associated malformations diagnosed by obstetric ultrasound  | Identify the prevalence of congenital malformations of the central nervous system (CNS) and associated malformations diagnosed by obstetric ultrasound                 | Obstetric ultrasonography has good sensitivity in the screening of fetal malformations of the CNS, especially with the constant improvement and mastery of specialized methods, such as Doppler and volumetric ultrasonography (3D / 4D), contributing to establish itself as a modality of choice. in this   |

|  |  |  |  |
|--|--|--|--|
|  |  |  | routine. Complementary to the method, magnetic resonance imaging may provide subsidies for even better perinatal care.   |
| <b>ELVIR et al., 2018</b>                | Hydranencephaly in a newborn due to congenital toxoplasmosis                               | To present the case of a newborn with hydranencephaly due to congenital toxoplasmosis  | Hydranencephaly, affected by congenital toxoplasmosis with prenatal diagnosis, acquires an early infestation during pregnancy, it is worth mentioning the diagnostic elements, such as positive maternal serology for active infection and that of newborns with high specific IgM                     |
| <b>GARDEA-LOERA &amp; VELAZCO-CAMPOS</b> | Clinical aspects of neuroimaging and electrophysiological behavior of hydranencephaly      | To report the cases of 4 children with hydranencephaly associated with bilateral occlusion of the internal carotid arteries, clinical aspects of neuroimaging. | hydranencephaly is defined as absence congenital cerebral hemispheres, is a way Severe presentation of rare porenencephaly, whose etiopathogenesis is not well defined   |
| <b>MUÑOZ et al., 2016</b>                | Congenital hydranencephaly: report of a teenager in northern Mexico                        | To present the case of a male adolescent with congenital hydranencephaly   | Hydranencephaly (HE) is a congenital condition that in most of cases is generally lethal, however, the cases that manage to survive have complications neurological due to impairment of the central nervous system. Although it has been documented HE as an important part of some genetic syndromes |
| <b>WALLS &amp; LÓPEZ, 2014</b>           | Hydranencephaly as the most severe presentation of fetal cerebral aplasia: regarding cases | Present two cases diagnosed in prenatal hydranencephaly  | Ultrasound studies show the absence cerebral supratentorial parenchyma, which is replaced by hypoechoic fluid; O Choroid plexuses may or may not be present brain stem is not affected and the lobes   |

|                                      |  |   |   |
|--------------------------------------|--|---|---|
|                                      |  |   | The occipitals can be partially preserved (70% of cases)  |
| <b>WERLANG &amp; FERNANDES, 2007</b> | Transillumination of the head: an important complementary aspect in the neurological assessment of the neonate | Rescue a technique useful in neonatologist evaluation | The transillumination method is fast, simple and low cost, being able to track and alert to possible diagnostic errors in antenatal procedures. The neurological assessment of the newborn is more accurate using this procedure, as it can easily reveal brain abnormalities, leading to a more precise and safe conduct in urgent and emergency care. |

Source: authors themselves (2019)

As a result, through thematic analysis, it was possible to analyze, interpret and categorize similar data. The following themes emerged from this categorization: What are the main rare diseases cited in the literature that are associated with hydranencephaly? What morphological factors justify the association between rare diseases and the development of hydranencephaly in children?

### 3 MAIN DISEASES CITED BY LITERATURE THAT ARE ASSOCIATED WITH HYDRANENCEPHALY

| DISEASES ASSOCIATED WITH HYDRANENCEPHALY |   |
|--|---|
| <b>HYDROCEPHALY</b>                      | It is characterized by an accumulation of fluid in the internal cavities of the brain. The extra fluid puts pressure on the brain and can cause brain damage. It can compromise vision, cognitive difficulties, loss of motor coordination and incontinence (OSBORN, 2018). |
| <b>MIELOMENINGOCELE</b>                  | Congenital defect in which a developing baby's spinal cord does not develop properly. Myelomeningocele originates from the development or closure of a baby's spinal cord in the womb, and does not happen properly (GOMES, Et al., 2019).                                  |
| <b>AGENESIA OF THE BODY CALLUS</b>       | If you have a congenital malformation that is characterized by the absence (agenesis) of the corpus callosum, with a significant increase in the occipital horns (DUARTE, et al., 2019).  |
| <b>ENCEPHALOCELE</b>                     | Encephalocele consists of a neural tube defect, characterized by herniation of the brain and meninges through openings in the skull (MOZZER, et al., 2019).   |

Source: Authors themselves.

There is evidence to suggest that neurological impairment in congenital toxoplasmosis is associated in some cases with hydranencephaly due to a rare condition and there are few records in the international literature (SANTOS et al., 2018).

#### **4 MORPHOLOGICAL FACTORS THAT JUSTIFY THE ASSOCIATION BETWEEN RARE DISEASES AND THE DEVELOPMENT OF HYDRANENCEPHALY IN CHILDREN**

Hydranencephaly can be a consequence of gestational intrauterine infections; or this condition may be due to a defect in vascular ontogenesis or by vascular occlusion of both carotids and their main branches (GARDEA-LOERA & VELAZCO-CAMPOS, 2014). What causes the infarction of the internal carotid arteries is still under discussion. It is proposed as alternatives to fetal hypoperfusion, inflammatory phenomena with arthritis related to maternal autoimmune states to fetal infections (toxoplasmosis, cytomegalic virus, herpes virus, rubella, monocytogenic listeria or *Treponema pallidum*) and acute vasoconstriction due to exposure to toxic infections (GAETE et al., 2011).

It was believed that the numbers of new cases are higher due to the outbreak of the ZIKA virus that afflicts the Americas (SANTOS et al., 2018).

Hydranencephaly, transillumination is related to the thinning of the brain lining, which when it is less than 1 cm thick, occurs completely. The absence of a telencephalon in the region irrigated by the carotid arteries results in the formation of a cystic cavity in the meningeal region, being pathognomonic for hydranencephaly (LACUNZA et al., 2014).

#### **5 IMPORTANCE OF FOLLOW-UP OF THE MULTIDISCIPLINARY TEAM IN THE ACCOMMODATION OF CHILDREN AFFECTED WITH HYDRANENCEPHALY AND THEIR FAMILIES.**

Second Hamad (2019), during the gestational period, women prepare for the birth of their child, experiencing moments of personal fulfillment, satisfaction, happiness, expectations, but also worry, anxiety, doubts, anguish, insecurities and fear. Pregnancy is a very significant event for women and their families. With the arrival of a baby with a congenital malformation, feelings arise that break with the idealization of a perfect child, widening the gap between what was imagined and / or desired and what is presented in reality, bringing to light the transformation of imagined reality, in a very particular way for mothers - and those who were involved with it, generating emotional distress, with changes in plans and routines, and in life.



Society establishes and values standards of strength, effectiveness, beauty and perfection, with emphasis on the child's efficiency and health. In general, the child is not expected to become ill, however, when there is no favorable prognosis, this situation can be considered hopeless, especially when there is the possibility that the child will die, or live in a situation of illness (HAMAD., 2019).

It was observed a great rejection, by the families, of children with some disability, in view of this, the authors concluded that when a family member suffers prejudices for having a disability, the whole family is affected. However, several reports of mothers abandoned by their partners, after the birth of a son / daughter with hydranencephaly (TORRES et al., 2017).

The nursing team plays an important role in the humanization of nursing care that aims to incorporate love in work relationships, empathy, respect and dignity on the part of nursing professionals are a differential in child care. Living with the diagnosis of the disease is a shock for the whole family, often destroying dreams about the idealized child, generating disappointments, insecurity, fear and uncertainty about the child's future, as it is a pathology that disrupts the routine of the people involved (PRANDO ., 2018).

When the pregnant woman receives the diagnosis that her child has some abnormality, in view of this situation, it is recommended by the health ministry that this pregnant woman should receive psychological care and guidance in order to reduce the impact of the news. Many times, parents have to abandon themselves when they receive the news, because most of the time they are not prepared to live with the situation (SANTOS., 2016).

## **6 CONCLUSION**

In hydranencephaly cases, about 90% of cases are diagnosed in the intrauterine phase, after birth, these patients usually develop until the first weeks of life, when the symptoms start to develop, the level of mortality increases, due to the treatments they are subjected to. On the other hand, with the application of modern medical methods, it was possible to increase the lives of some patients, which allows them to live for a few more years. Even with the low incidence of hydranencephaly cases, the number of patients with survival above the first year of age has increased considerably in recent decades.

It is worth mentioning that there is still no treatment that can reverse the condition and these patients are submitted to ventricular shunt with several severe complications. These complications lead to the need for surgery to adapt the derivation systems and antibiotic therapies for prolonged periods, resulting in high costs for the health system.

The literature is deficient regarding this rare condition in relation to its carriers, the quality of life, prognosis and survival, as it is restricted to only case studies and experiments. Most case reports are written in the fetal phase, there are few studies in the post-neonatal period.

It is worth mentioning the importance and the need to report and develop new research on hydranencephaly, so that we can better understand how prevention actions take, ways of diagnosis, prognosis, treatment options, pathophysiology and survival rates; generating knowledge bringing with it more positive perspectives for its carriers.

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