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MALFORMAÇÕES CONGÊNITAS DA FOSSA POSTERIOR: UMA REVISÃO DAS CARACTERÍSTICAS CLÍNICAS E RADIOLÓGICAS POR MEIO DE UM GUIA ILUSTRADO

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Congenital Malformations of the Posterior Fossa: A Review of Clinical and Radiological Features Through an Illustrated Guide

Malformações Congênitas da Fossa Posterior: Uma Revisão das Características Clínicas e Radiológicas Por Meio de Um Guia Ilustrado

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ABSTRACT

Congenital malformations of the posterior fossa (PF) encompass a wide spectrum of morphological anomalies that arise during embryonic development. This paper provides a narrative review of these most common disorders from a morphological approach based on the division into cystic (Dandy-Walker malformation, Blake's Pouch cyst, mega cisterna magna, arachnoid cyst, and cerebellar vermis hypoplasia) and non-cystic malformations (Joubert syndrome, rhomboencephalosynapsis, and pontocerebellar hypoplasia). The embryogenesis of PF structures is briefly outlined. While magnetic resonance imaging is the preferred modality for evaluation, computerized tomography and ultrasonography serve complementary roles. In conjunction with clinical aspects, specific radiological features, such as cerebellar vermis morphology, Torcular Herophili position, and the presence/absence of mass effect, are highlighted for accurate diagnosis. We also present some typical radiological findings in non-cystic malformations, such as molar tooth sign, diamond-shaped fourth ventricle, and dragonfly-shaped cerebellum. This comprehensive review aims to assist radiologists, neuropediatricians, and general neurologists in recognizing and describing PF malformations, thereby facilitating appropriate management strategies.

Keywords: Cranial Fossa, Posterior; Neuroimaging; Congenital Abnormalities.

RESUMO

As malformações congênitas da fossa posterior (FP) abrangem um amplo espectro de anomalias morfológicas que surgem durante o desenvolvimento embrionário. Este artigo fornece uma revisão narrativa desses distúrbios mais comuns a partir de uma abordagem morfológica baseada na divisão em malformações císticas (malformação de Dandy-Walker, cisto da bolsa de Blake, mega cisterna magna, cisto aracnoide e hipoplasia do vermis cerebelar) e não císticas (syndrome de Joubert, rombencefalossinapse e hipoplasia pontocerebelar). A embriogênese das estruturas da FP é brevemente descrita. Embora a ressonância magnética seja a modalidade preferida para avaliação, a tomografia computadorizada e a ultrassonografia desempenham funções complementares. Em conjunto com os aspectos clínicos, características radiológicas específicas, como a morfologia do vermis cerebelar, a posição da torcula Herophili e a presença/ausência de efeito de massa, são destacadas para um diagnóstico preciso. Apresentamos também alguns achados radiológicos típicos de malformações não císticas, como sinal do dente molar, quarto ventrículo em forma de diamante e cerebelo em forma de libélula. Esta revisão abrangente visa auxiliar radiologistas, neuropediatras e neurologistas gerais no reconhecimento e na descrição das malformações da FP, facilitando, assim, estratégias de manejo adequadas.

Palavras-chave: Fossa Craniana Posterior; Neuroimagem; Anomalias Congênitas.

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INTRODUCTION

Congenital malformations of the posterior fossa (PF) encompass a large spectrum of morphological anomalies that occur during embryonic development. Detecting these features during the pre- and post-natal periods is possible. Typically, genetic abnormalities are involved, which may be inherited from the parents or arise as de novo mutations that appear solely in the affected child, in addition to being acquired.[1]

For many years, the congenital abnormalities of the PF were neglected by scientific literature. However, in recent decades, advances in the genetic sequencing processes and in neuroimaging techniques have enabled a greater understanding of these entities.[1-4] This directly impacted the management of these conditions alongside the families, since it is now possible to establish diagnoses, prognoses, and management strategies more clearly, regarding genetic counseling and family planning.[1]

Anatomically, the posterior fossa has a floor made up of portions of the sphenoid, temporal, and occipital bones. Its roof is limited by the cerebellar tent, presenting the pons, the medulla oblongata, the fourth ventricle, and the cerebellum as its main components.[5-7] Magnetic resonance imaging (MRI) is the modality of choice for evaluating these structures, and will be the focus of this discussion. The computerized tomography (CT) can be used, but it is usually an inaccurate examination for analyzing the PF.[8] In the prenatal period, the ultrasonography (US) can also be used for evaluation and diagnosis, despite its limitations.[9-11]Although advances have been made in genetic testing techniques, imaging recognition often plays a crucial role in diagnosis and guiding further studies of these disorders. This is because the genetic diagnostic yield remains low for some PF anomalies, such as rhombencephalosynapsis.

The classification of congenital PF abnormalities is challenging. This is due to the variety of terms and criteria used, which often generate controversy among researchers.[7,9] The chosen approach for this study is based on the division into cystic malformations, in which there are abnormal cerebrospinal fluid (CSF) collections in retrocerebellar and/or infracerebellar region, and non-cystic, with absence of this CSF abnormality. The aim of this article was to emphasize the clinical characteristics and typical radiological findings of the most common congenital disorders of the PF through an illustrated guide.

MATERIALS AND METHODS

In order to compose the illustrated guide, self-authored drawings were produced based on real neuroimaging exams, aiming to point out relevant characteristics of each malformation. We also added images from our personal archive, such as computerized tomography and magnetic resonance imaging, of children affected by the congenital malformations of the posterior fossa highlighted in this article. This study was approved by the Ethics in Research Involving Human Subjects Committee at the Federal University of Piauí, protocol number 3.131.058 and the Presentation Certificate to Ethics Assessment, registry number 03106518.3.0000.5214. The written informed consent was obtained from all participants in the study.

To support our discussion, a narrative review of the medical literature was performed using PubMed and Google Scholar. In this search, we used the terms “congenital malformations of the posterior fossa” and “embryology of the posterior fossa”.

EMBRIOLOGY

The formation of the human brain is complex and marked by steps controlled by different cellular and molecular signaling pathways associated to genetic components.[11] A full discussion about the embryogenesis of the PF components is beyond the scope of this article, but a brief review of this process is warranted.

After the closure of the neural tube, around the fourth week of gestation, two constrictions appear in the cephalic region of the central nervous system structure, dividing it into forebrain, midbrain, and hindbrain, which are called primary cerebral vesicles.[4,9,12] The secondary vesicles originate from the forebrain, which is subdivided into the diencephalon and telencephalon. They also originate from the midbrain, which forms a homonymous vesicle, and from the hindbrain, which gives rise to the metencephalon and myelencephalon. The pons and the upper portion of the fourth ventricle derive from the metencephalon, while the medulla oblongata and the lower portion of the fourth ventricle derive from the myelencephalon.[6,7] The alar plates of the cranial portion of the metencephalon give rise to the cerebellar hemispheres, whereas the cerebellar vermis arises from the alar plates of the caudal third of the midbrain.[10]

As the embryo's brain grows, the cephalic and caudal flexures are formed. Around 6 weeks of gestation, the pontine flexure appears between them. The *plica choroidalis*, a precursor of the choroid plexus, divides the roof of the fourth ventricle into anterior and posterior membranous areas.[8] The latter is responsible for the formation of the cisterna magna, a space filled with CSF located infracerebellar and retrobulbar. It is worth mentioning that Blake's pouch usually disappears between 16 and 18 weeks of embryonic development, marked by the fenestration of the fourth ventricle and emergence of Magendie foramen.[6,10]

CYSTIC MALFORMATIONS

Dandy-Walker Malformation

Dandy-Walker Malformation (DWM) is a sporadic entity with a low recurrence rate (1-5%) whose estimated prevalence is 1 per 30,000 live births.[1,13] According to the most accepted pathophysiological theory, DWM is characterized by an interruption in the development of the hindbrain and absence of fusion of the cerebellum in the midline, along with the persistence of the anterior membranous area. As a result, this area expands and herniates posteriorly, becoming interposed between the choroid plexus and the hypoplastic vermis.[11]

Typical signs and symptoms are associated with increased intracranial pressure and macrocephaly, which are present in most patients with DWM before 1 year of age.[1,14] DWM can occur alone or associated with chromosomal anomalies and several disorders, for example, craniocerebellar-cardiac syndrome, Meckel-Gruber syndrome, and Smith-Lemli-Opitz syndrome.[15-17] Therefore, the degree of cognitive impairment varies. Other already documented manifestations are ocular (chorioretinal coloboma, strabismus and nystagmus) and auditory problems, seizures, vomiting, syndactyly, polydactyly, cleft palate, delay in psychomotor development, ataxia, and spasticity.[17,18]

The radiological findings of classic DWM include hypoplasia of the cerebellar vermis, which is usually elevated and rotated counterclockwise, cystic dilatation of the fourth ventricle, and an enlargement of the PF with elevation of adjacent structures, such as the tentorium and torcula (figure 1, A-C).[1,5,9] These are classic and widespread criteria for recognizing DWM. Nonetheless, the modern phenotype of this disorder, according to new studies, is best defined by inferior predominant vermian hypoplasia, an enlarged tegmentovermian angle, inferolateral displacement of the choroid plexus, and an obtuse fastigial recess.[19,20] Additionally, supratentorial hydrocephalus, absence of the cerebellum falx, variable degrees of hypoplasia and displacement of the cerebellar hemispheres, callosal dysgenesis or agenesis, polymicrogyria, heterotopias, occipital encephalocele, syringomyelia, and schizencephaly can be found.[13,16] MRI is the technique of choice for evaluating these findings. The differential diagnosis of DWM includes the other cystic malformations of the PF as suggested by the flowchart we created (figure 2).

Dandy-Walker Variant

Not all patients exhibit the classic DWM neuroimaging phenotype identified by MRI. For these cases, the terminology Dandy-Walker variant (DWV) was introduced, in which the basic difference is the degree of extension of fourth ventricle enlargement. Although large, it is not enough to cause PF enlargement.[9]

Clinical features include enlargement of the head due to hydrocephalus and neurodevelopmental delays. Cognitive deficits are more pronounced when the condition is associated with supratentorial alterations.[15]

Neuroimaging shows the fourth ventricle less dilated than in DWM, without a significant widening of the PF. Partial hypoplasia and counterclockwise rotation of the cerebellar vermis are often present, but to a lesser degree when compared to DWM. Supratentorial hydrocephalus may or may not be present, thus this feature cannot be used to differentiate DWM and DWV. Furthermore, the

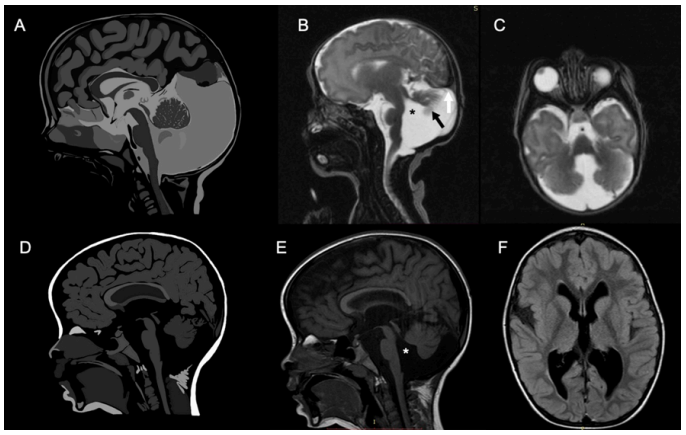

Figure 1. Dandy-Walker malformation and Blake's pouch cyst. Schematic representations and MRI findings of Dandy-Walker malformation (A-C) and Blake's pouch cyst (D-F). Schematic representation (A) and sagittal T2-weighted MRI (B) showing cystic dilatation of the fourth ventricle (asterisk), vermian hypoplasia with anterosuperior rotation (black arrow), and elevated tentorium and torcula (white arrow). (C) Axial T2-weighted MRI highlighting a dilated fourth ventricle that communicates with the posterior fossa and a lateral displacement of the cerebellar hemispheres. Schematic representation (D) and sagittal T1-weighted MRI (E) showing the dilated fourth ventricle (asterisk). The vermis is intact and in a normal position. Note the thinning of the corpus callosum. (F) Axial FLAIR brain MRI highlighting cerebral atrophy and mild colpocephaly.

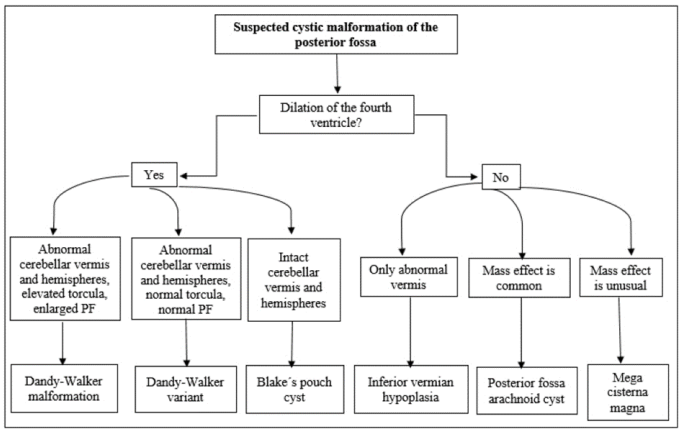

Figure 2. Flowchart for the differential diagnosis of cystic malformations of the posterior fossa.

cerebellar falx is usually identified, as well as agenesis of the corpus callosum, cerebral heterotopias, and holoprosencephaly.[10]

Some researchers use the terminology Dandy-Walker Complex (DWC) or Dandy-Walker spectrum to group a continuum of congenital PF anomalies in which vermian hypoplasia and incomplete formation of the fourth ventricle, that commonly communicates with an important CSF space in the FP, can be found.[11] DWM, DWV, cystic persistence of Blake's pouch, and mega cisterna magna are part of this spectrum.[10,11] These terminologies, as well as the entities that are actually part of the DWC, however, are controversial, and several authors claim that they are dubious, obsolete and unspecific.[1,7]

Blake's Pouch Cyst

Blake's pouch cyst (BPC), also known as cystic persistence of Blake's pouch, is a posterior fossa abnormality characterized by the failure of regression of Blake's pouch. This failure appears to be related to the non-fenestration of the median aperture (foramen of Magendie).[21,22] Once such perforation does not occur, there is no longer a connection between the subarachnoid space and the fourth ventricle, resulting in subsequent expansion of the inferior medullary velum into the cisterna magna.[23] It is a condition that occurs sporadically and without reported cases of recurrence.[1]

Regarding clinical aspects, they are diverse. In the neonatal period, macrocephaly and hydrocephalus are common findings.[1] There are patients with intracranial hypertension and complications within the cyst, for instance, infections and hemorrhages.[11,24] Others, however, may be asymptomatic or have mild cognitive impairment, and the diagnosis is often made incidentally.[25] Seizures, cerebellar ataxia, dizziness, and mental retardation,[17,18] if present, may manifest themselves in the first years of life. In the long term, if there are no associated supratentorial anomalies, the prognosis is usually favorable.[8,14]

MRI is the preferred modality for evaluating this condition. Typical radiological characteristics involve the presence of a cyst in the infra- or retrocerebellar region caused by dilatation of the fourth ventricle and cerebellar vermis with normal morphology and without rotation (figure 1, D-F). Torcula is in a normal position. Moreover, contrast-enhanced MRI usually shows displacement of the choroid plexus inferior to the vermis.[18] PF is usually normal but may be enlarged. A slight mass effect can be noticed on the lower vermis and the cerebellar hemispheres, although this does not alter their morphology, in addition to anterior displacement of the brainstem against the clivus.[9,14] Except for hydrocephalus, supratentorial alterations are uncommon.[18]

Mega cisterna magna

Mega cisterna magna (MCM) consists of a congenital malformation in which the PF is increased due to an enlargement of the cisterna magna, usually above 10mm, although this value is still discussed.[26] It is believed that this occurs due to a late permeabilization of Blake's pouch, causing the pouch to increase and expand towards the PF before its fenestration takes place.[9,22] This condition has no reported recurrence rates and is very frequent, representing more than 50% of all cases of congenital anomalies in the PF.[7,8]

Many patients are asymptomatic, and more than 90% of children with the isolated condition develop normally, with most cases discovered incidentally.[22] Despite this, developmental delays, personality disorders, and somatization have been reported. The most symptomatic picture appears in cases in which there is association with supratentorial alterations.[8]

On neuroimaging, in addition to the aforementioned increase in the cisterna magna and PF (figure 3, A-C), there is an intact vermis, differentiating it from DWM, absence of fourth ventricle dilatation, unlike what occurs in BPC, and absence of mass effect in the cerebellum, distinguishing it from an arachnoid cyst.[27] The fourth ventricle is normal in size, with free communication between the cisterna magna and the adjacent subarachnoid space.[8] If there is a very pronounced increase in the cisterna magna, occipital thinning may occur.[22,28] Supratentorial anomalies – agenesis of the corpus callosum, holoprosencephaly, diencephalic cyst – and posterior meningocele have already been found, despite more rarely.[1]

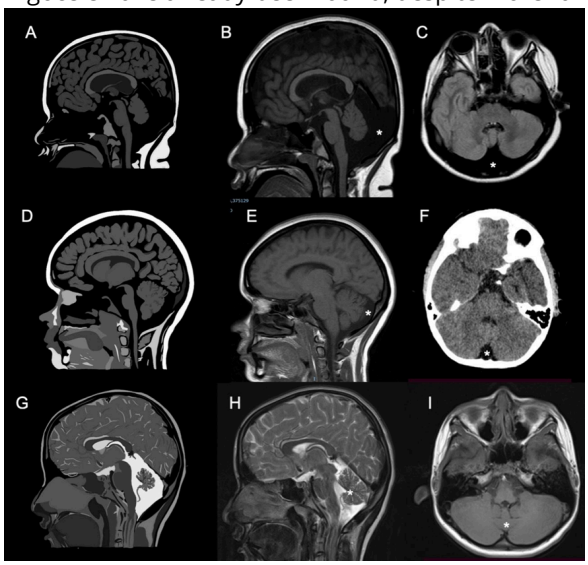

Figure 3. Mega cisterna magna, posterior fossa arachnoid cyst, and inferior vermian hypoplasia. Schematic representations and MRI/CT findings of mega cisterna magna (A-C), posterior fossa arachnoid cyst (D-F), and inferior vermian hypoplasia (G-I). Schematic representation (A) and sagittal T1-weighted MRI (B) showing enlargement of the CSF space on the posteroinferior aspect of the midline of the posterior fossa (asterisk), thinning of the occipital bone, without enlargement of the fourth ventricle. (C) Axial FLAIR brain MRI highlighting cyst with a content isointense to CSF (asterisk). The cerebellar architecture is preserved. Schematic representation (D) and sagittal T1-weighted MRI (E) evidencing a cyst with a content isointense to CSF in the posterior-inferior region of the cerebellum (asterisk). The size and position of the vermis and fourth ventricle are normal. (F) Axial CT section showing retrocerebellar arachnoid cyst (asterisk) with mild mass effect. Schematic representation (G), sagittal T2-weighted MRI (H), and axial T1-weighted MRI (I) showing hypoplasia of the cerebellar vermis (asterisks).

Posterior Fossa Arachnoid Cyst

Arachnoid cysts (AC) are benign cysts formed from the separation or duplication of the arachnoid membrane, leading to filling of the arachnoid membrane by CSF.[29] They can occur in different locations, being more common in the middle cranial fossa.[7,8] Less commonly found in the PF (approximately 10% of cases), AC are typically located in the retrocerebellar, supravermian regions, or anterior regions of the brainstem.[9] It is a sporadic condition that has no documented recurrence and rarely associated with supratentorial anomalies.[1]

Clinical manifestations depend on the patient's age, on the size of the cyst, and on the presence of complications (infections and hemorrhage).[9] AC are usually asymptomatic. In children, for instance, macrocephaly, developmental delays, and signs of increased intracranial pressure can occur, caused by a mass effect in adjacent structures.[9,14] Ataxia is frequent, but headache and dysdiadochokinesia are rarer presentations.[29] In newborns, rapid expansion of the arachnoid cyst may occur due to hemorrhage secondary to birth trauma, worsening the symptoms.[9,29]

On imaging, both on MRI and CT, AC usually appear as unilocular, ovoid, or crescent-shaped masses filled with fluid. They appear as structures with well-defined margins, with the cystic content being isointense in relation to the CSF (figure 3, D-F).[8,9,29] It is required absence of cystic communication with the ventricular system in this anomaly. The fourth ventricle, vermis, cerebellar hemispheres, and brainstem are usually normal, unless mass effect occurs, which is common only when the cyst is very large. When present, the mass effect can lead to asymmetry in the calvaria, secondary obstruction in the ventricular system, hydrocephalus, and thinning of the adjacent occipital bone.[9]

Cerebellar Vermis Hypoplasia

Cerebellar vermis hypoplasia (CVH) is a congenital anomaly characterized by partial hypoplasia of the vermis, more commonly affecting its lower portion. When the term isolated inferior vermian hypoplasia (IIVH) is used, there are no other evident alterations. This is justified by the cerebellar development, which occurs in the craniocaudal direction. CVH can occur in isolation or related to other syndromes.[30]

Most patients with IIVH have a favorable prognosis, with a milder clinical picture. Some cases may present with mild functional deficits in fine motor activities and receptive language, in addition to discreet cognitive impairments.[28]

In IIVH, neuroimaging shows partial hypoplasia of the inferior vermis, while the rest of the vermis, as well as the cerebellar hemispheres, the fourth ventricle, and the PF are normal in size, findings well highlighted in the sagittal plane by MRI (figure 3, G-I). The fourth ventricle may present some degree of enlargement, but without major repercussions. The diagnosis of this condition is challenging and, even with the MRI, false-negative rates can reach 32%.[1]

Although we have approached CVH as a separate condition, some authors consider it an alternative denomination to DWV, placing it in the same category, that is, CVH as a constituent of DWC. In DWV, according to the line of reasoning we adopted, the fourth ventricle, despite not being able to widen the PF, is dilated. In CVH, however, this same structure is normal or slightly enlarged in size, but it is still not enlarged compared to DWV. For this reason, we consider that this condition does not fully meet the DWV criteria, which justifies our choice of approach. The clinical and radiological correlation of cystic PF malformations is summarized in the Table 1.

Table 1. Key clinical and neuroimaging features of the cystic congenital posterior fossa malformations.

Malformation	Clinical features	Neuroimaging features						
		Vermis	Cerebellar Hemispheres	Fourth ventricle	Posterior fossa	Torcula	Hydrocephaly	Mass effect
Dandy-Walker malformation	Macrocephaly, variable degree of cognitive and neurodevelopmental impairments, craniocerebellar-cardiac syndrome, Meckel-Gruber syndrome, and Smith-Lemli-Opitz syndrome	Hypoplastic	Hypoplastic	Enlarged	Enlarged	Elevated	Yes	No
Dandy-Walker variant	Macrocephaly and variable degree of cognitive impairment (less pronounced)	Hypoplastic	Hypoplastic	Normal or slightly dilated	Normal	Normal	Rarely	No
Blake´s Pouch Cyst	Miscellaneous: asymptomatic or symptomatic (macrocephaly, hydrocephalus, mild cognitive impairment, ataxia, seizures, dizziness)	Intact	Intact	Enlarged	Normal	Normal	Yes	Slightly
Mega cisterna magna	Usually asymptomatic. Rare reports: neurodevelopmental delay, personality disorders and somatization	Intact	Intact	Normal	Normal or enlarged	Normal	No	No
Aracnoid Cyst of the Posterior Fossa	Macrocephaly, neurodevelopmental delay, signs of intracranial hypertension, ataxia.	Intact	Intact	Normal	Normal	Normal	Possible	Yes
Vermian Hypoplasia	Mild deficits in fine motor activities and receptive language	Hypoplastic (inferior portion)	Normal	Normal	Normal	Normal	No	No

NON-CYSTIC MALFORMATIONS

Joubert Syndrome

Joubert syndrome encompasses a group of rare autosomal recessive disorders, also known as ciliopathies. Men tend to be more affected than women, with an approximate ratio of 2:1.[8,10] The pathophysiology involves malformations in several structures' secondary to defects in the immobile primary cilium organelle, which is important in the formation and functioning of several cells, such as neurons, retinal photoreceptors, and bile ducts.[31]

Clinical manifestations include ataxia, hypotonia, abnormal eye movements, abnormal neonatal breathing (alternating apnea with tachypnea), facial dysmorphism, and cognitive impairment of varying intensity.[9,28,31] The most common abnormal eye movements are nystagmus and congenital ocular motor apraxia (COMA).[32,33] Retinal dystrophy and hepatic fibrosis can also be found.[9,28]

Typical radiological aspects are varying degrees of hypoplasia or dysplasia of the vermis and the classic "molar tooth sign", which is characterized by a deep

interpeduncular fossa and long superior cerebellar peduncles, that are thick, horizontally arranged, and do not decussate (figure 4, A-C).[31,34] These findings are seen on axial MRI sections. In about 25% of cases, there may be an association with cystic changes in the PF, as dilation of the fourth ventricle with upward displacement of the fastigium, which makes the diagnosis more challenging.[17] Other associated disorders include PF enlargement, hydrocephalus, polymicrogyria, corpus callosum dysgenesis, pituitary absence and occipital encephalocele.[7,9]

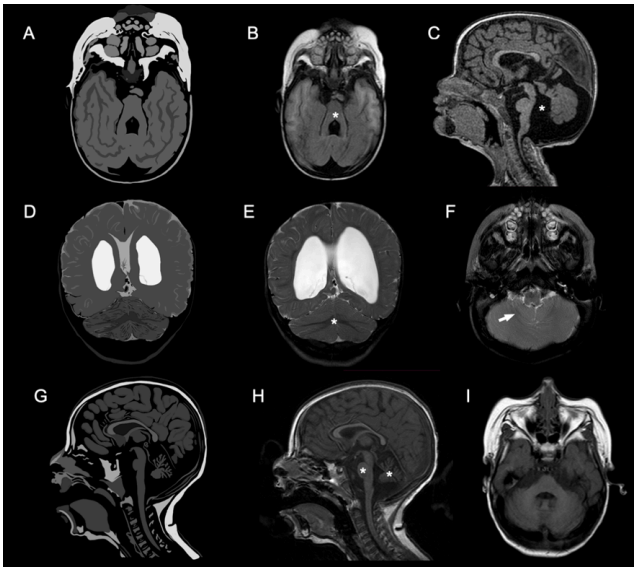

Figure 4. Joubert syndrome, rhomboencephalosynapsis, and pontocerebellar hypoplasia. Schematic representations and MRI findings of Joubert syndrome (A-C), rhomboencephalosynapsis (D-F), and pontocerebellar hypoplasia (G-I). Schematic representation (A) and axial FLAIR brain MRI (B) highlighting the molar tooth sign (asterisk) with a deepened interpeduncular fossa and elongated, thickened, and horizontally oriented superior cerebellar peduncles. (C) Sagittal T1-weighted MRI showing hypoplastic cerebellar vermis and dilated fourth ventricle (asterisk). Schematic representation (D) and coronal T2-weighted MRI (E) showing the fusion of the cerebellar hemispheres (asterisk) and an abnormally oriented cerebellar folia. (F) Axial T2-weighted MRI highlighting continuity of the dentate nuclei (arrow), cerebellar peduncles, and cerebellar hemispheres. Schematic representation (G), axial T1-weighted MRI (H), and Sagittal T1-weighted MRI (I) showing marked cerebellar atrophy (involving vermis and cerebral hemispheres) and pons hypoplasia with involvement of the cerebellopontine angle (asterisks).

Rhomboencephalosynapsis

Rhomboencephalosynapsis (RES) is a condition of PF in which the vermis is totally or partially absent and the cerebellar hemispheres, cerebellar peduncles, and dentate nuclei fuse at the midline level.[35] It is a rare disorder whose exact prevalence and pathophysiology are still unknown. This malformation rarely occurs alone, being frequently associated with other intra- and extracranial disorders.[36]

Clinical characteristics are directly related to the degree of severity. Most patients are asymptomatic, especially when RES appears in isolation. However, it commonly occurs with Gomez–Lopez–Hernandez syndrome (parietal alopecia, trigeminal anesthesia, and signs of craniofacial dysmorphia) and VACTERL (vertebral anomalies, anal atresia, cardiovascular anomalies, tracheoesophageal fistula, renal anomalies, and limb defects) syndromes, situations in which the clinical presentation is more prominent.[4,35,36] When symptomatic, ataxia, hyperactivity or impulsivity, delayed motor development, and abnormal eye movements are reported.[36] Although sporadic, hydrocephalus due to aqueduct stenosis points to stereotypical head movements and may be a clinical diagnostic clue.[37]

Typical MRI features are varying degrees of agenesis of the vermis and fusion of the dentate nuclei, cerebellar peduncles, and cerebellar hemispheres, which forms a horseshoe-like arch at the midline level due to abnormally oriented folia, giving the fourth ventricle a keyhole or a diamond shape (figure 4, D-F).[35] RES may also be related to other abnormalities of the central nervous system, such as hydrocephalus, absence of the septum pellucidum and olfactory bulbs, as well as dysgenesis of the corpus callosum. Hypoplasia of the pons and fusion of the anterior pillars of the thalamus, although uncommon, have already been reported.[35,36]

Pontocerebellar hypoplasia

Pontocerebellar hypoplasia (PCH) represents a heterogeneous group of autosomal recessive disorders with prenatal onset marked mainly, but not exclusively, by pons and cerebellar hypoplasia.[38] More than 10 PCH subtypes have been detected with different phenotypes and pathophysiological mechanisms. In some occurrences, depending on the type involved, the involvement of the cerebellar hemispheres is more pronounced. Although it begins during pregnancy, this condition is rarely diagnosed in utero.[38,39] Specific analysis of PHC subtypes is beyond the scope of this discussion.

PCH involves a myriad of clinical features. Broadly, we may find nystagmus, postnatal microcephaly, ataxia, varying degrees of cognitive impairment, seizures, retinopathies, respiratory failure, hypotonia, muscle weakness, optic nerve atrophy, facial dysmorphisms, dystonias, chorea, generalized clonus, and problems with sexual development.[1,38,39]

MRI findings include cerebellar hypoplasia with superimposed atrophy, absence or significant reduction of pontine prominence, and normal or slightly reduced PF size (figure 4, G-I). Moreover, diverse brain involvement may occur, such as atrophies and delays in myelination, abnormal size of the corpus callosum, and microcephaly.[40] Patients who have mutations in the CASK, RELN, VLDRL genes present with a more severe degree of pontine and cerebellar hypoplasia. In these situations, especially in coronal images, this greater involvement presents a dragonfly appearance, characterized by flattened and reduced cerebellar hemispheres with a relatively preserved vermis. It is important to mention that the pattern of neuroimaging features commonly associated with PCH is not restricted to this anomaly; it is also found in other conditions, for example, extreme prematurity and neurometabolic diseases.[41] The clinical and radiological features of non-cystic PF malformations are summarized in the Table 2.

Table 2. Key clinical and neuroimaging features of the non-cystic congenital posterior. fossa malformations.

Malformation	Clinical features	Neuroimaging features
Joubert syndrome	Ataxia, hypotonia, abnormal eye movements, apnea alternating with tachypnea, facial dysmorphism, retinal dystrophy, hepatic fibrosis, variable cognitive impairments	Common: hypolasia of the vermis, classic molar tooth sign. Rare: posterior fossa enlargement, polymicrogyria, corpus callosum agenesis
Rhombencephalosynapsis	Gomez–Lopez–Hernandez and VACTERL syndromes features, ataxia, hyperactivity or impulsivity, delays in motor development, and abnormal eye movements. Usually asymptomatic	Variable degree of agenesis of the cerebellar vermis with continuity of the hemispheres across the midline (diamond-shaped fourth ventricle), absence of septum pellucidum and olfactory bulbs
Pontocerebellar hypoplasia	Moderate to severe neurodevelopmental impairment, microcephaly, seizures, early postnatal mortality, muscle weakness, hypotonia, ataxia, impaired sexual development, nystagmus, variable degree of cognitive impairment	Moderate to severe cerebellar hypoplasia, delays in myelination, pons hypoplasia, dragonfly-shaped cerebellum

CONCLUSION

The classification of congenital PF malformations is controversial. More important than adhering strictly to precise terminology or attempting to establish a standardized approach is understanding how to adequately correlate the clinical characteristics and radiological aspects of these anomalies. This approach, combined with knowledge about the normal embryonic development of the PF, may enable radiologists and child neurologists, for instance, to make proper diagnoses.

REFERENCES

1. Bosemani T, Orman G, Boltshauser E, Tekes A, Huisman TAGM, Poretti A. Congenital abnormalities of the posterior fossa. RadioGraphics. 2015;35(1):200–220.
2. D'Antonio F, Khalil A, Garel C, Pilu G, Rizzo G, Lerman-Sagie T, et al. Systematic review and meta-analysis of isolated posterior fossa malformations on prenatal ultrasound imaging (part 1): nomenclature, diagnostic accuracy and associated anomalies. Ultrasound Obstet Gynecol. 2016;47(6):690–697.
3. Barkovich AJ. Developmental disorders of the midbrain and hindbrain. Front Neuroanat. 2012;6:7.
4. Doherty D, Millen KJ, Barkovich AJ. Midbrain and hindbrain malformations: advances in clinical diagnosis, imaging, and genetics. Lancet Neurol. 2013;12(4):381–393.
5. D'Antonio F, Khalil A, Garel C, Pilu G, Rizzo G, Lerman-Sagie T, et al. Systematic review and meta-analysis of isolated posterior fossa malformations on prenatal imaging (part 2): neurodevelopmental outcome. Ultrasound Obstet Gynecol. 2016;48(1):28–37.
6. Milani HJF, Barreto EQDS, Ximenes RLDS, Baldo CAR, Araujo Júnior E, Moron AF. Fetal posterior fossa malformations: review of the current knowledge. Radiol Bras. 2019;52(6):380–386.
7. Struffert T. Anatomie und Fehlbildungen der hinteren Schädelgrube. Radiologe. 2016;56(11):960–966.
8. Schlatterer SD, Sanapo L, du Plessis AJ, Whitehead MT, Mulkey SB. The role of fetal MRI for suspected anomalies of the posterior fossa. Pediatr Neurol. 2021;117:10–18.
9. Kollias SS, Ball WS, Prenger EC. Cystic malformations of the posterior fossa: differential diagnosis clarified through embryologic analysis. RadioGraphics. 1993;13(6):1211–1231.
10. Shekdar K. Posterior fossa malformations. Semin Ultrasound CT MRI. 2011;32(3):228–241.
11. Cotes C, Bonfante E, Lazor J, Jadhav S, Caldas M, Swischuk L, et al. Congenital basis of posterior fossa anomalies. Neuroradiol J. 2015;28(3):238–253.
12. Niesen CE. Malformations of the posterior fossa: Current perspectives. Semin Pediatr Neurol. 2002;9(4):320–334.
13. Reith W, Haussmann A. Dandy-Walker-Malformation. Radiologe. 2018;58(7):629–635.
14. Vedolin L. Congenital malformation of the posterior fossa: new insights. Top Magn Reson Imaging. 2011;22(6):259.
15. Monteagudo A. Dandy-Walker malformation. Am J Obstet Gynecol. 2020;223(6):B38–41.
16. Jurc MC, Kozma K, Petche AD, Bembea M, Pop OL, Mu G, et al. Anatomic variants in Dandy–Walker complex. Rom J Morphol Embryol. 2017;58(3):1051-1055.
17. Bertholdo D, De Carvalho Neto A, Castillo M. Posterior fossa malformation associated with cerebral anomalies: genetic and imaging features. Top Magn Reson Imaging. 2011;22(6):295–302.
18. Altman NR, Naidich TP, Braffman BH. Posterior fossa malformations. Am J Neuroradiol. 1992;13(2):691-724.
19. Whitehead MT, Barkovich MJ, Sidpra J, Alves CA, Mirsky DM, Öztekin Ö, et al. Refining the neuroimaging definition of the Dandy-Walker phenotype. Am J Neuroradiol. 2022;43(10):1488–1493.
20. Haldipur P, Bernardo S, Aldinger KA, Sivakumar T, Millman J, Sjoboen AH, et al. Evidence of disrupted rhombic lip development in the pathogenesis of Dandy-Walker malformation. Acta Neuropathol (Berl). 2021;142(4):761–776.
21. Oliveira G da P, Moura CGF, Fontenele JL, Mendes MBR, Oliveira JC, Soares NLR. Clinical and neuroradiological features of Blake's pouch cyst. Rev Bras Neurol. 2019;55(4):25–26.
22. Azab W, Shohoud S, Elmansoury T, Salaheddin W, Nasim K, Parwez A. Blake′s pouch cyst. Surg Neurol Int. 2014;5(1):112.
23. Calabrò F, Arcuri T, Jinkins JR. Blake's pouch cyst: an entity within the Dandy-Walker continuum. Neuroradiology. 2000;42(4):290–295.
24. Post A, Norton ME, Monteagudo A. Blake's pouch cyst. Am J Obstet Gynecol. 2020;223(6):B47–50.
25. Chapman T, Mahalingam S, Ishak GE, Nixon JN, Siebert J, Dighe MK. Diagnostic imaging of posterior fossa anomalies in the fetus and neonate: Part 1, normal anatomy and classification of anomalies. Clin Imaging. 2015;39(1):1–8.
26. Zimmer EZ, Lowenstein L, Bronshtein M, Goldsher D, Aharon-Peretz J. Clinical significance of isolated mega cisterna magna. Arch Gynecol Obstet. 2007;276(5):487-490.
27. Kau T, Marterer R, Kottke R, Birnbacher R, Gellen J, Nagy E, et al. Blake's pouch cysts and differential diagnoses in prenatal and postnatal MRI : a pictorial review. Clin Neuroradiol. 2020;30(3):435–445.
28. Chapman T, Mahalingam S, Ishak GE, Nixon JN, Siebert J, Dighe MK. Diagnostic imaging of posterior fossa anomalies in the fetus and neonate: part 2, posterior fossa disorders. Clin Imaging. 2015;39(2):167–175.
29. Pradilla G, Jallo G. Arachnoid cysts: case series and review of the literature. Neurosurg Focus. 2007;22(2):1–4.
30. Robinson AJ. Inferior vermian hypoplasia – preconception, misconception. Ultrasound Obstet Gynecol. 2014;43(2):123–136.
31. Crawford D, Dearmun A. Joubert syndrome. Nurs Child Young People. 2017;29(5):15.
32. Schröder S, Li Y, Yigit G, Altmüller J, Bader I, Bevot A, et al. Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. Genet Med. 2021;23(2):341–351.
33. Serpieri V, D'Abrusco F, Dempsey JC, Cheng YHH, Arrigoni F, Baker J, et al. SUFU haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. J Med Genet. 2022;59(9):888–894.
34. Valente EM, Dallapiccola B, Bertini E. Joubert syndrome and related disorders. Handb Clin Neurol. 2013;113:1879–1888.

35. Aldinger KA, Dempsey JC, Tully HM, Grout ME, Mehaffey MG, Dobyns WB, et al. Rhombencephalosynapsis: fused cerebellum, confused geneticists. Am J Med Genet C Semin Med Genet. 2018;178(4):432–439.
36. Fouda MA, Kim TY, Cohen AR. Rhomboencephalosynapsis: review of the literature. World Neurosurg. 2022;159:48–53.
37. Tully HM, Dempsey JC, Ishak GE, Adam MP, Mink JW, Dobyns WB, et al. Persistent figure-eight and side-to-side head shaking is a marker for rhombencephalosynapsis. Mov Disord. 2013;28(14):2019–2023.
38. Maricich SM, Aqeeb KA, Moayedi Y, Mathes EL, Patel MS, Chitayat D, et al. Pontocerebellar hypoplasia: review of classification and genetics, and exclusion of several genes known to be important for cerebellar development. J Child Neurol. 2011;26(3):288–294.
39. Rudnik-Schöneborn S, Barth PG, Zerres K. Pontocerebellar hypoplasia. Am J Med Genet C Semin Med Genet. 2014;166C(2):173–183.
40. Zakaria RBM, Malta M, Pelletier F, Addour-Boudrahem N, Pinchefsky E, Martin CS, et al. Classic "PCH" genes are a rare cause of radiologic pontocerebellar hypoplasia. Cerebellum. 2023;23(2):418-430.
41. Riquet A, Quesque F, Charkaluk ML, Desnoulez L, Neut D, Joriot S, et al. Differentiating genetic forms of pontocerebellar hypoplasia from acquired lesions resembling pontocerebellar hypoplasia: clinical, neurodevelopmental, and imaging insight from 19 extremely premature patients. J Child Neurol. 2023;38(10–12):622–630.