Chapter

Rare Brain Tumors with Infrequent Clinical Manifestations: Illustrative Cases

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Abstract

This chapter describes the epidemiology, clinical and neuroimaging features, histological characteristics, surgical approach, outcomes, and prognostic factors of different cases of very rare intracranial tumors, associated with complex clinical syndromes. Highlighting the important aspects in the diagnosis and management that were considered relevant through the experience of our center. Here we included an intracranial Rosai-Dorfman disease manifested as an apparent multiple meningiomatosis, a choroid plexus papilloma clinically manifested as a hemifacial spasm originated by a compression of the facial colliculus, and a neuroenteric cyst associated with Klippel-Feil syndrome. This type of tumor presents a challenge to the neurosurgeon, originating various questions about its management. In this chapter, we present the experience we had with these pathologies to establish the most appropriate management decisions.

Keywords: rare intracranial tumors, multiple meningiomas, Rosai-Dorfman disease, Hemifacial spasm, choroid plexus papilloma, Klippel-Feil syndrome, neuroenteric cyst

1. Introduction

Brain tumors according to their location and growth rate can produce very typical clinical manifestations [1], in addition to the classic characteristics of imaging studies that provide the possibility of approaching the diagnosis of the specific type of tumor and guide to establish the treatment modality [2]. However, when the incidence of some of these tumors is very low and they present with very varied clinical manifestations, added to the radiological findings that do not provide too much information to approximate the diagnosis, these cases condition stricter study protocols where the undoubtedly diagnoses alters the treatment modality for each particular case [3]. For this reason, knowledge of the existence of some of these tumors should be the subject of study, to understand the difficulty in diagnosis and treatment, seeking to reduce errors in addressing these cases and improve the result.

2. Rosai-Dorfman disease manifested as an apparent multiple meningiomatosis

The first case presented in this chapter corresponds to an intracranial Rosai-Dorfman disease (RDD), which was manifested as an apparent multiple meningiomatosis that affected the anterior fossa, the sphenoidal plane, and the clivus. RDD is a non-Langerhans histiocytosis described in 1965 by Pierre P. Destombes and then characterized by J. Rosai and R. Dorfman [4, 5]. RDD has a prevalence of 1 in 200,000 and an incidence of 100 cases per year. It can occur at any age but is usually more common in adolescents [6]. The typical clinical manifestations of this disease are the presence of painless bilateral cervical lymphadenopathy added to the presence of fatigue, fever, and weight loss, associated with elevated erythrocyte sedimentation rate, anemia, fever, and hypergammaglobulinemia. Extranodal involvement occurs in less than 43% of cases, mostly in the skin, nasal cavity, and bone [7]. The central nervous system (CNS) is affected in less than 5% of cases, the isolated affection is possible, without systemic manifestations. There are approximately 200 reported cases of intracranial RDD [8, 9]. This case illustrates a patient with multiple intracranial lesions, where the symptoms and characteristics per image simulated the presence of multiple meningiomas, where the RDD finding was made until the moment of the histopathological study.

2.1 Case presentation

A 59-year-old male patient with a history of gradual right hearing loss that later presents the same symptoms in the contralateral ear. His current condition began 8 months ago with a high-intensity holo-cranial headache that predominated in the mornings accompanied by occasional dizziness. Three months before his hospital admission, he reported non-quantified weight loss, asthenia, and adynamia.

Two months before hospital admission, the patient reported decreased visual acuity and compromise of the temporal hemifields added to hyposmia. The reason for hospital admission in our institution was the presence of two generalized tonic–clonic seizures lasting more than one minute (less than five minutes), these seizures were characterized by the absence of aura, with a postictal period of 20 minutes. The second seizure required hospital admission for control. On physical examination, the cognitive functions were preserved, evaluation of the cranial nerves demonstrate hyposmia and bitemporal hemianopia. Fundoscopy showed edema of the papilla in the left eye and an atrophic papilla of the right eye. Regarding the complementary studies, the electroencephalogram showed abnormal bifrontal activity. Computed campimetry confirmed the bitemporal hemianopia, and regarding the neuroimaging studies, the computed tomography (CT)-scan showed three isodense with homogeneous enhancement lesions located in the midline in the floor of the anterior fossa in the cribriform plate, the sphenoidal plane with extension to the tuberculum sellae, and the middle and lower portion of clivus. The magnetic resonance imaging (MRI) revealed isointense lesions with peritumoral edema, with intense and homogeneous gadolinium-enhancement demonstrating a dural attachment (**Figure 1A–F**).

The diagnosis of multiple meningiomas was established, supported by the neuroimaging features and previous experience. The surgical plan was to resect the two main symptomatic lesions (olfactory groove and tuberculum sellae). The surgical approach was made through a bicoronal incision to perform a bifrontal craniotomy and a sub-frontal approach. The surgical approach allowed a complete resection of the lesions, after the olfactory groove lesion resection was possible to access the lesion of the tuberculum sellae. Debulking of the lesions was made

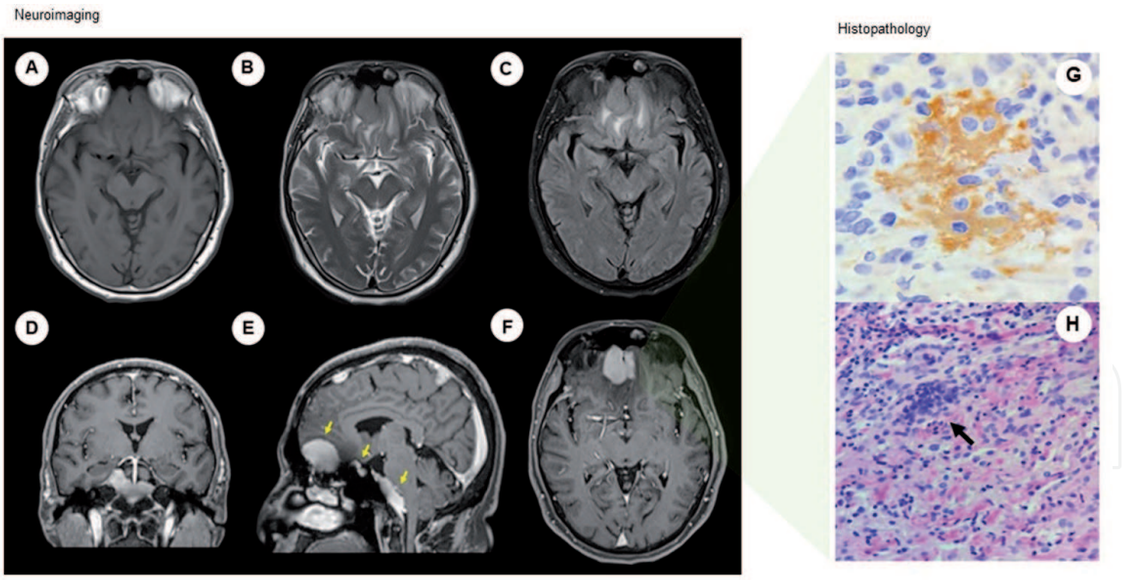

Figure 1.
Pre-operative brain magnetic resonance imaging (MRI) studies. Non-contrasted brain MRI shows an isointense olfactory groove lesion with perilesional edema in the T1-weighted (A), T2-weighted (B), and FLAIR (C). Contrasted brain MRI reveals three homogeneous enhancement lesions with dural attachment (yellow arrows in the sagittal section) in the floor of the anterior fossa in the crista Galli and cribriform plate, the sphenoidal plane with extension to the tuberculum sellae, and the middle and lower portion of clivus, observed in the coronal (D), sagittal (E), and axial sections (F). Histopathological analysis. G. Positive immunohistochemical profile for S100 protein. Furthermore, other immunochemical profiles show positive expression of CD68 (macrophages), CD20 (B lymphocytes), and CD2 (T lymphocytes), in which lymphagocytosis was observed. IgG and IgG4 positivity were also identified. A negative expression for CD30 and CD15 (reed Stenberg cells), and CD1A (Langerhans cells) was observed. H. H&E Stain: Mixed inflammatory infiltrate with plasma cells, lymphocytes, and macrophages, no evidence of meningothelial cells, emperipolesis was observed (black arrow).

with an ultrasonic aspirator, and according to meningioma surgery principles, it was decided to perform anterior fossa drilling to reduce the recurrence probability. The tumor lesions showed low vascularity and close contact with the optic chiasm (**Figure 2**). Immediately post-operative the patient remained without complications and was discharged five days after surgery, the CT scan performed 5 days after surgery showed complete resection of the lesions (olfactory groove and tuberculum sellae), with residual lesion of the middle and lower portion of clivus (**Figure 2F**). Due to the residual lesion, the patient was observed for the clinical oncology service to decide adjuvant management.

Histopathological findings established the diagnosis of RDD (**Figure 1G** and **H**). In the subsequent follow-up, extension studies were carried out, which were ruled out other infiltrates with a thoracic and abdomen-pelvic CT. The patient received treatment with prednisone and remain asymptomatic without clivus lesion growth at 8 months follow-up.

2.2 Case discussion

For the diagnostic process of RDD, it is important to consider the observations by imaging studies that usually mimic the characteristics of a meningioma, either as one or multiple extra-axial lesions with homogeneous contrast enhancement surrounded by vasogenic perilesional edema, they can arrive to present a dural tail and its location is very diverse [1]. Proton MRI spectroscopy improves the specificity of preoperative diagnoses in some patients; for example, in meningiomas, alanine is usually elevated in spectroscopy, with a peak at 1.48 ppm, in a patient with RDD disease, spectroscopy revealed an increased choline level [10]. In a review of 10 cases, a dural tail was found in all cases. Therefore, in data suggestive

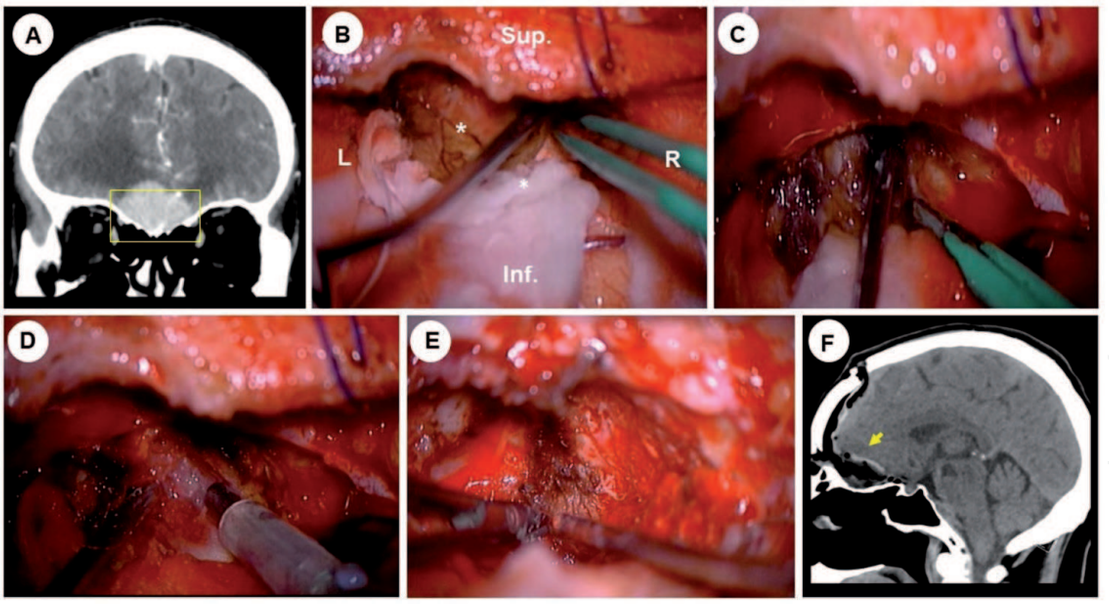

Figure 2.
(A) Preoperative enhanced coronal head CT-scan with an olfactory groove lesion (square). Surgical procedure: Approach to the anterior fossa lesion. Perimeter dissection of the anterior cranial fossa lesion (asterisk) is shown (B-C). Debulking with ultrasonic aspiration (D) and complete resection of olfactory groove (E) is demonstrated. (F) Sagittal section of postoperative CT with complete resection (arrow).

of meningioma obtained by neuroimaging studies, RDD is a differential diagnosis [11]. Zhu et al. [11] concluded that, unlike meningiomas, a typical hypointensity non-related to calcification on T2-weighted or fluid attenuation inversion recovery (FLAIR) images could suggest the RDD diagnosis.

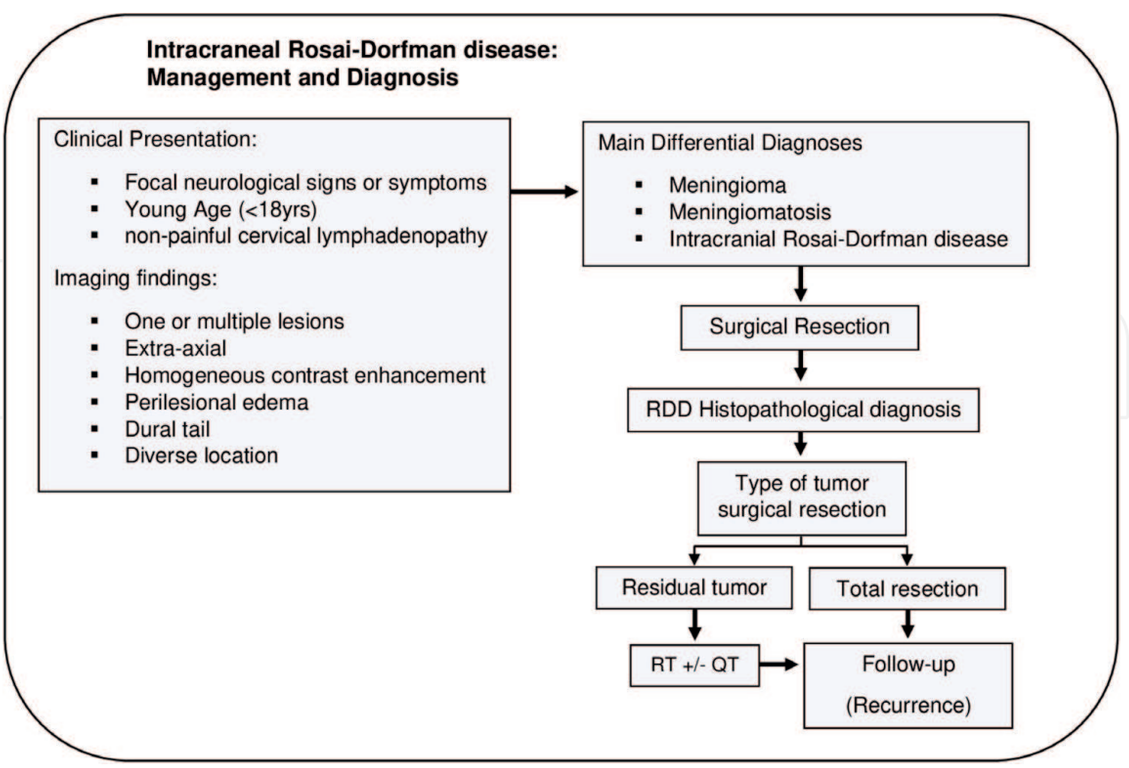

Figure 3.
Algorithm for management and diagnosis (clinical suspect) for intracranial Rosai-Dorfman disease. Clinical suspicion is obtained by clinical evaluation and imaging studies. The differential diagnosis is made with meningioma, corroborating it by the histopathological and immunohistochemical study. Relative to management, the presence of residual tumor after surgery suggests performing radiotherapy with or without chemotherapy. Strict monitoring after surgery is recommended due to the risk of recurrence.

Histopathological characteristics correspond to a large lymphohistiocytic infiltrate, with a large or vesicular nucleus, well-defined nuclear membranes and a single and prominent nucleolus. The main characteristic is the intracytoplasmic presence of lymphocytes and, to a lesser extent, intact erythrocytes, plasma cells, and neutrophils ("emperipolesis"), however, it may be absent in 30% of leptomeningeal lesions [7, 9]. Associated with histiocyte proliferation, a perivascular plasmacytic infiltrate can be observed. From the immunohistochemical point of view, they are characterized by presenting protein S100 +, CD68 +, CD11c +, MAC387 +, lysozyme +/−, being negative for CD1a, a positive marker in Langerhans cell histiocytosis [10, 12].

Illustrative case	Pearls and pitfalls
Intracranial Rosai-Dorfman Disease (RDD)	• The most affected population group are young people (<18 yrs), where the main clinical manifestation of is massive cervical lymphadenopathy. • RDD: Suggestive data of meningioma by neuroimaging studies • Brain MRI: hypointensity non-related to calcification on T2-weighted or FLAIR images could suggest the RDD diagnosis. • Total resection is the most recommended management. • Radiation therapy is indicated in case of residual disease. • Chemotherapy can be helpful in extensive (disseminated) disease. However, there is insufficient evidence on its efficacy in isolated disease. • A periodic follow-up (3–6 mos) with imaging studies should be carried out, in search of local recurrences or extensive disease.
Choroid plexus papilloma (Tumoral compression of the facial colliculus)	• Differentiation between primary and secondary HFS is elementary, imaging studies are fundamental, supported by electrodiagnostic studies. • In HFS not related to vascular compression, we recommend to intentionally search tumoral compression at the facial colliculus level at the floor of the fourth ventricle through an MRI scan with contrast. • Surgical approach: Telovelar approach to the fourth ventricle. • Intraoperative EMG register is an elemental tool to determine the impact of surgical treatment and resolution of symptoms.
Neuroenteric cyst (NEC) on posterior fossa	• Cystic lesion located in posterior fossa (90%). Differential diagnoses are mainly cystic lesions; arachnoid cysts, epidermoid cyst, dermoid cyst, neurocysticercosis, or metastases, cholesteatoma, ependymoma, schwannoma, hemangioblastoma, and pilocytic astrocytoma. • Diagnosis can be suspected in recurrent meningitis due to a fistula to the aerodigestive tract. • Slight diffusion restriction in Diffusion-weighted imaging (DWI) due to xanthogranulomatous changes or presence of melanin, hemosiderin, proteins, mucopolysaccharides and cholesterol. • Surgical treatment recommended for NEC is complete resection. If resection is partial, remnants adhered to neurovascular structures should be electrocoagulated to avoid reaccumulation. • Cystoperitoneal and ventriculoperitoneal shunts are second-line procedures recommended in recurrence with high difficulty for a new excision. • Minimum follow-up is recommended for 10 years, every 6 months at the first 2 years (complemented with CA 19–9 measurement on CSF).

RDD, Rosai-Dorfman disease; MRI, Magnetic resonance imaging; FLAIR, Fluid attenuation inversion recovery; HFS, Hemifacial spasm; EMG, Electromyography; NEC, Neuroenteric cyst; DWI, Diffusion-weighted imaging; CA 19-9, carbohydrate antigen 19-9; CSF, Cerebrospinal fluid.

Table 1.
Rare tumors: Pearls and pitfalls in diagnosis, surgical management, and prognosis.

Related to the management of this disease is primarily with surgery, seeking to eliminate the mass effect and the associated neurological sequelae. Total resection is the main objective, although a partial resection is allowed in case the lesions are in complex regions. The optimal management of residual disease remains unclear due to the rarity of the disease [13]. The use of radiotherapy has shown some efficacy of residual or recurrent disease, postoperative doses of 20 Gy in 10 fractions in 2 weeks after subtotal resection has shown a good effect in reducing symptoms and reducing the size of the lesions [14]. The use of chemotherapeutic agents such as alkaloids and anthracyclines, alkylating agents, and methotrexate have shown variable efficacy [15]. Rivera et al. [16] reported the use of the modified CHOP regimen in two cases of intracranial RDD observing a long-term remission [16].

Adeleye et al. [17] reported a series of 111 cases of RDD involving the CNS [17]. Of the population studied, 77% presented intracranial disease, which received various treatments with surgery, radiotherapy and chemotherapy. 37% of the study population had a long-term follow-up beyond one year (41% of these patients had no recurrence of the disease), where a relapse or growth of the most residual tumor was determined in 12%. Therefore, active monitoring after the surgery is prudent; however, it is not well established how often to follow up with imaging studies and when it would be prudent to classify the disease as remitted. Rivera et al. [18] reported the longest reported follow-up time (7 years) [18], of two patients with intracranial RDD with surgical resection and chemotherapeutic management with the modified CHOP scheme consisting of 8 cycles of cyclophosphamide (1 g), Vincristine (2 mg), doxorubicin (50 mg), and prednisone (50 mg) for 5 days every 3 weeks. Where it was observed that during follow-up there were no recurrences. Due to the low incidence of the disease, it is difficult to standardize diagnostic, therapeutic and prognosis. For this reason, we propose a simple algorithm for the diagnosis and management of intracranial RDD (**Figure 3**). **Table 1** describes the fundamental aspects in the management of this pathology.

3. Hemifacial spasm associated with compression of the facial colliculus by a choroid plexus papilloma

As a second case, we have an extremely rare cause of hemifacial spasm (HFS). HFS is an involuntary neuromuscular disorder in which it affects the facial musculature, which usually has as its primary cause a mechanical compression over the cisternal portion of the facial nerve in the root entry zone by an aberrant or ecstatic vessel in the 60–70% of the cases [19]. Etiology's different than vascular compression are called secondary causes, which correspond commonly to those pathologies that occupy the space in the cerebellopontine angle such as: aneurysms, arteriovenous malformations or tumor growths. Compression of the facial colliculus due to the presence of a tumor is an extremely rare cause, representing less than 0.6% of HFS cases [20]. What makes this case even more exceptional is that the tumor that was conditioning the compression of the facial colliculus was a choroid plexus papilloma (CPP), an uncommon benign intraventricular neuroepithelial tumor [21, 22].

3.1 Case presentation

A 43-year-old female who presents left severe HFS, associated with headaches, symptoms started 6 months before presentation at our service, characterized by the onset of periocular and expression muscles, increasing in intensity and frequency. The patient had the antecedent of one episode of left facial palsy 6 years ago with full

recuperation 2 months later. The first management was with botulin toxin, showing a low response to initial treatment. Clinical examination at our functional neurosurgery service, found left HFS with labial commissure deviation, palpebral occlusion, and extension of the spasm to the neck. The patient did not refer pain and has no evidence of facial palsy. HSF presented every 2 min with a 15 seconds duration. Other cranial nerves did not show any alterations, and hearing was not affected. Clinical assessment was complemented with Brain MRI with gadolinium, showing a tumoral growth in the floor of the fourth ventricle that homogeneously captured gadolinium without infiltrating the floor of the fourth ventricle (**Figure 4A** and **B**). Preoperative and intraoperative electromyography (EMG) recordings were considered for the management. The preoperative register showed normal auditory and motor-evoked potentials. EMG was free from synchronic neuromyotonic discharges in muscles innervated by the left facial nerve, corresponding to the HFS clinically founded.

It was decided to perform surgery to remove the tumor growth of the superior colliculus to improve the clinical status of the HFS. A telovelar approach with intraoperative neurophysiology recordings of the facial nerve was performed. The surgical procedure was performed with the patient in the prone position and head fixation. An incision of 1 cm was made above the inion up to the C2 spinous process, the C1 posterior arch was recognized, and the tectorial membrane was dissected. Conventional suboccipital craniectomy was conducted. Dural opening in Y was realized before transverse sinus identification. Under the microscopic vision, the tela choridea was opened, and the tumor was identified. The tumor had a pearly appearance. After dissection complete resection was made. At the extraction of the tumor, there was a nervous hyperexcitability correction in intraoperative EMG recording

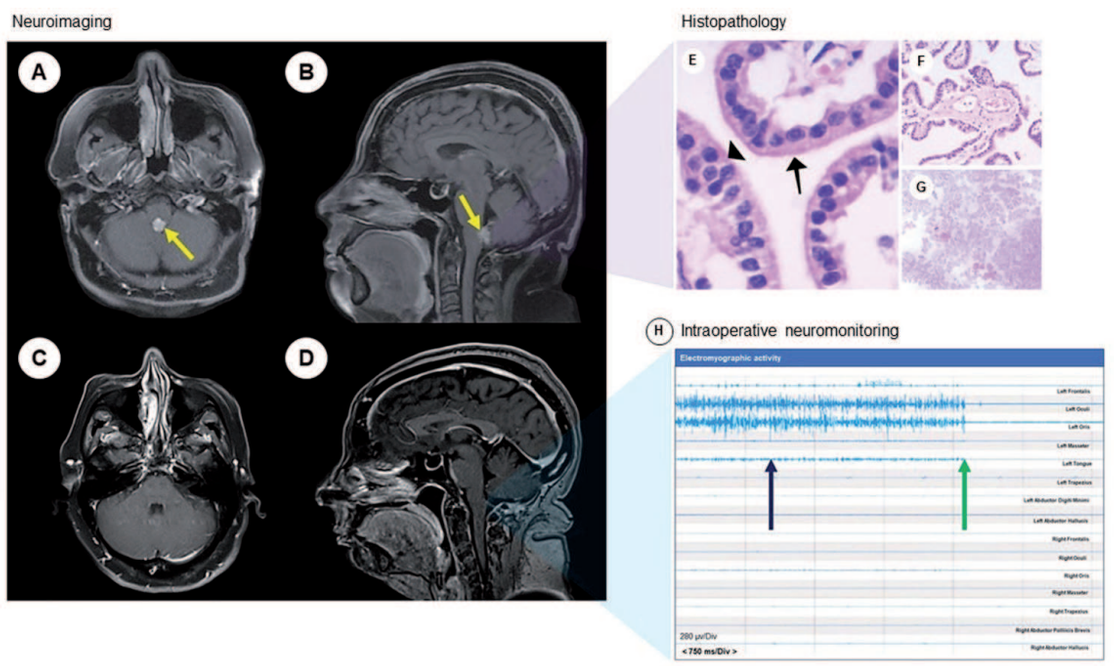

Figure 4.
Neuroimaging: Preoperative gadolinium-enhanced T1-weighted MRI showed that the right side of the fourth ventricle was occupied by a hyperintense tumor (arrows). (A) Axial view. (B) Sagittal view. T1-weighted sequences with gadolinium showing complete resection of the tumor. (C) Axial view. (D) Sagittal view. Histopathology: Choroid plexus papilloma. (E) HE x40: Cylindrical coating epithelium with flat apical domain and multiple microvilli (arrow). Cells have round-to-oval nuclei, moderate amount of acidophilous cytoplasm, and some focally pseudostratified (arrowhead) and oriented to the basal domain. (F) HE ×10: Lesion mimics the papillary architecture of a normal choroid plexus with thin fibrovascular stems and coated by a simple cylindrical epithelium. (G) HE ×4: Epithelial neoplasia. (H) Intraoperative neuromonitoring: Electromyographic activity shows synchronic neuromyotonic discharges in muscles innervated by the left facial nerve (blue arrow). Cessation of irritative activity over the left orbicularis oculi and orbicularis oris after en-bloc removal of the tumor (green arrow).

of the facial musculature (**Figure 4**). Motor-evoked potentials did not show alterations during the surgical intervention. Postoperative histopathology examination demonstrates CPP in the fourth ventricle (**Figure 4E–G**). The clinical outcome of the patient in the immediate postoperative period was a diminution in the intensity and frequency of spasms. At 12 months of follow-up, complete symptom resolution was observed without associated neurological deficits. Postoperative MRI at 1 year of follow-up showed complete resection of the tumor (**Figure 4C** and **D**).

3.2 Case discussion

HFS's most common pathophysiological mechanism corresponds in 60–70% of the cases to mechanical compression over the cisternal portion of the facial nerve in the root entry zone by an aberrant or ecstatic vessel in the 60–70% of the cases. Conversely, secondary common etiologies are pontocerebellar angle tumors, traumas, demyelination conditions, and infections. Therefore, the tumors in adults related to HFS are rare (0.3–2.5%), and the tumoral compression at the facial colliculus level, at the floor of the fourth ventricle is considered an exceptional etiology, being gliomas, subependymomas, ependymomas the neoplasia's reported [19]. The pathological mechanism of HFS is unclear. However, different theories suggest that the direct compression of the facial nerve in its cisternal portion by a vascular structure is the most related mechanism of injury, which leads to local demyelination. On the other hand, another hypothesis suggests a central/nuclear origin, that states change in the reorganization of functional connections within the facial nerve nucleus, generating irritative activity that produces abnormal discharges, precipitating HFS. A hypothesis that would be more related to the mechanism of HFS production in the presence of a tumor mass growing in the superior colliculus. This case supports the central theory of direct facial nucleus irritability, generating a hyperexcitability state that precipitates the discharges [23]. Microvascular decompression is the most frequent surgical treatment used for HFS. It is usually indicated when a vascular contact is found by MRI, which is usually effective management in more than 80% of cases [24]. However, because in this case a vascular contact was not found, the surgical management is different, focused on the complete resection of the tumor. Therefore, due to the uncertainty that may exist in the clinical outcome as it is an infrequent presentation, intraoperative monitoring is a very useful tool that helps to define the effectiveness that surgical intervention could have in symptomatic improvement, observing changes in symptoms, and synchronic neuromyotonic discharges during resection [25].

In conclusion, secondary HFS are infrequent conditions. Direct compression by tumors at the facial colliculus level associated with HFS is an exceptional case. Clinical findings do not allow differentiation between primary and secondary HFS, for this reason we recommend an adequate evaluation of the brain MRI, supported by electrodiagnostic studies. Tumoral compression at the facial colliculus level at the floor of the fourth ventricle is an exceptional etiology, and there is very few information in the medical literature about management and diagnosis. Therefore, about our experience, in **Table 1** we describe the fundamental aspects in the management of this pathology.

4. Neuroenteric cyst on posterior fossa associated with Klippel-Feil syndrome

Neuroenteric cysts (NEC) are rare benign (the malignant transformation is extremely rare) lesions of the spinal axis composed of heterotopic endodermal

tissue generally located in the intradural extramedullary space in the lower cervical and upper thoracic spine, comprising 0.03% of intracranial lesions and 16% of cystic lesions of the CNS, they can found rarely intracranially, examples in the posterior fossa are ventral to the brainstem or in the cerebellopontine angle, and they present with hydrocephalus, headache, and cranial nerve deficits [26, 27]. The origin of these lesions is not completely understood, histologically are composed of cuboidal epithelium that resembles the gastrointestinal or respiratory tract. Therefore, are thought to arise from rostrally located vestigial remnants of the neuroenteric canal [28]. The consensus for treatment is complete surgical resection, when possible, partial resection should be avoided because of the high risk of recurrence. However, due to their rarity, there is a lack of information about management [29]. On the other hand, Klippel-Feil syndrome (KFS) is defined as the fusion of two or more cervical vertebrae, with a classic triad of limitation of cervical movements, short neck (brevicollis), and low hair implantation in 52% of patients [30]. Association between NEC and KFS has not been described.

4.1 Case presentation

A 21-year-old male patient began his current disease with a severe occipital headache of one month of evolution, the headache was aggravated by Valsalva maneuvers, occasionally associated with nausea and emesis. Physical examination showed brevicollis with restricted range of motion, low hair implantation (KFS). Moreover, examination shows papilledema, dysdiadochokinesia, and right dysmetria. Head CT exhibited a heterogeneous cystic lesion located on the posterior fossa, conditioning an obstructive hydrocephalus, for which an urgent ventriculoperitoneal shunt was placed (demonstrating clear cerebrospinal fluid). Sagittal section of a spine CT (**Figure 5A**) that demonstrates C1-C2 and C3-C4 anterior and posterior elements fusion, without thoracic, lumbar, or sacral alterations. Anteroposterior and lateral static and dynamic cervical spine radiographs and spine CT exposed lordosis rectification, flexion, and extension limitation. Brain MRI showed (**Figure 5B** and **C**) an infratentorial lesion, dorsal to the right cerebellar hemisphere, ovoid shape, with regular and defined borders, composed of a nodular portion in contact with pia mater, and multiple punctate flow absences, isointense on T1, heterogeneously hyperintense on T2 and FLAIR sequences, with diffusion restriction on its central portion and contrast enhancement, whose measurements were 17 × 14 × 15 mm in major axes, and spectroscopy displayed increased N-Acetyl-Aspartate and choline peaks (**Figure 5D**); another remaining cystic portion was hyperintense on T1, hypointense on T2, FLAIR, and apparent diffusion coefficient, without diffusion restriction or contrast enhancement, whose measurements were 46 × 49 × 46 mm. Due to data compatible with KFS, simple contrasted thoracoabdominal CT, echocardiography, and renal function tests were obtained; otorhinolaryngology assessment cursed without hearing alterations, and medical genetics confirmed the syndromic diagnosis.

A total complete surgical resection was decided. Subsequently, a midline suboccipital craniectomy was performed. The surgical procedure involves resection of the C1 posterior arch and tumor excision, obtaining a cystic lesion with a mural nodule at the inferolateral right torcular level, with leakage of greenish fluid (**Figure 5E**). Complete resection of the capsule was achieved, with a histopathological study that reported smooth, opaque, light gray color walls, with tortuous vessels, and peripheral solid, anfractuous, gray-green areas of firm consistency, clear brown content, and soft consistency compatible with NEC, positive to alcian blue and negative to periodic acid-Schiff stains (**Figure 5F**). The patient had a favorable clinical evolution, receiving medical discharge to home

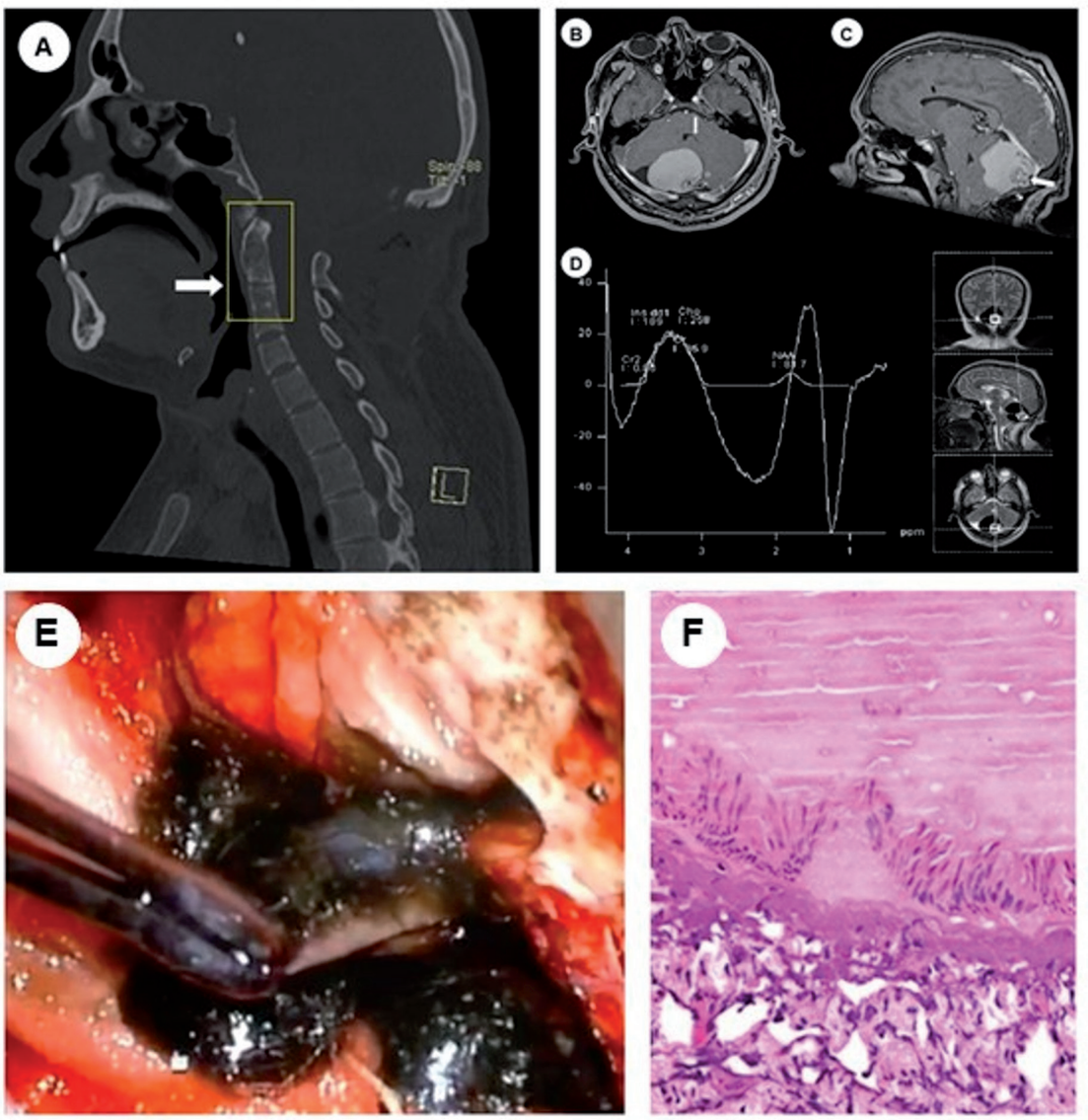

Figure 5.
(A) Cervical spine computed tomography (CT) with evidence of C1-C2 and C3-C4 anterior elements fusion (white arrow). Presurgical brain contrast-enhanced T1-weighted magnetic resonance imaging (MRI). (B) Axial section, there is evidence of an extra-axial lesion in the posterior fossa, which is contrast enhanced and displaces the cerebellum and brainstem ventrally, collapsing the fourth ventricle (white arrow). (C) MRI sagittal section. (D) Spectroscopy without choline (Cho) elevation according to the indicated voxel on T2-sequence hypointense lesion. Intraoperative images: (E) The tumor capsule with a good cleavage plane presents liquid content of oily material inside the capsule. (F) Histopathology image: HE (400X), demonstrate a cyst wall and proteinaceous content with some spaces for cholesterol crystals.

after 3 weeks, with adequate follow-up 9 months after surgery, identifying by MRI a residual nodular image adhered to the straight sinus.

4.2 Case discussion

NEC was described for the first time in 1928 and the first intracranial NEC was reported in 1962 [26], with more than one hundred cases reported since then [31]. Relative to the epidemiological characteristics of the patients, frequency is higher in men, and the age of presentation ranges from the neonatal period to 70 years. Regarding intracranial location, the initial findings are at the second or third decades of life. The predominant localization is on the posterior fossa (90%), specifically at prepontine and prebulbar cisterns, cisterna magna, cerebellopontine angle, fourth ventricle, and dorsal to the cerebellum. The etiopathogenesis is due to

abnormal endodermal-ectodermal adhesion during gastrulation at embryological development, with the persistence of endodermal elements near the notochord in the neuroaxis, which would explain the association with spinal disorders (spina bifida, diastematomyelia, and vertebral body alterations). Supratentorial localizations are exceptional. Infratentorial lesions usually present headache, nausea, and cranial nerve alterations such as vertigo, hearing loss, tinnitus, hypoesthesia, or trigeminal neuralgia. Diagnosis can be suspected in recurrent meningitis due to a fistula to the aerodigestive tract that causes slow growth because of active secretion from epithelial cells. Accompanying disorders are intestinal malformations and cutaneous abnormalities. Clinical manifestations can be acute or insidious, with a course ranging from 4 months to 40 years [30, 32].

Regarding the characteristics observable by neuroimaging studies in the diagnosis of NEC, in head CT is hypodense lesions without contrast enhancement. However, density depends on protein concentration. MRI shows heterogeneous lesions (well-defined, extra-axial, rounded or lobulated cysts), hyperintense on T1, T2 and FLAIR, without contrast enhancement, with slight diffusion restriction in Diffusion-weighted imaging due to xanthogranulomatous changes or presence of melanin, hemosiderin, proteins, mucopolysaccharides and cholesterol. Differential diagnoses on the posterior fossa are mainly cystic lesions; arachnoid cysts, epidermoid cyst, dermoid cyst, neurocysticercosis, or metastases, cholesteatoma, ependymoma, schwannoma, hemangioblastoma, and pilocytic astrocytoma [33, 34]. During surgery macroscopically visualization corresponds to yellow, milky white, gray, or red cysts, with thin walls similar to arachnoid, and transparent, mucoid or xanthochromic liquid content, unusually blood, pus, calcifications, or keratinized debris adhering to the adjacent pia mater. Histopathological studies reveal benign lesions with simple, pseudostratified, columnar epithelium and collagenous fibrous connective tissue lined with gastrointestinal epithelium, with the presence of goblet cells [26]. In immunohistochemistry, they are positive for cytokeratin, epithelial membrane antigen, and carcinoembryonic antigen. Degeneration to adenocarcinoma is extremely unusual and only occurs in intracranial locations (9 patients reported) [27]. In these cases, carbohydrate antigen 19–9 (CA 19–9) is positive [26], elevated MIB-1 labeling index suggests malignancy [27]. No correlation between imaging findings and pathology has been found [35].

The surgical treatment recommended for NEC is complete resection. Wang et al. [35] described a technique that shows an improvement in prognosis and limits recurrence [35]. If resection is partial, remnants adhered to neurovascular structures should be electro-coagulated to avoid reaccumulation. Surgical approaches depend on the location and the optimal visualization of the lesion and adjacent structures to minimize the risk of neurological deficits [36]. Cystoperitoneal and ventriculoperitoneal shunts are second-line procedures recommended in recurrence with high difficulty for a new excision [28]. Postoperative complications are aseptic meningitis, abducens nerve palsy, pseudomeningocele, and cerebrospinal fluid fistula [36]. Although the prognosis is mostly favorable, one-third of patients experience a symptomatic recurrence in a period of 2 months to 32 years [36]. Minimum follow-up is recommended for 10 years, every 6 months at the first 2 years [35] and can be complemented with CA 19–9 measurement on cerebrospinal fluid to determine recurrence [37].

In this case report, we did not find a specific genetic alteration that explains the relationship between KFS and NEC. The commonly associated disorders in KFS are mostly spinal disorders how congenital scoliosis and spina bifida occulta, in some cases this disease is related to hearing alterations, genitourinary defects, cardiovascular anomalies, and other skeletal abnormalities [38]. The association between KFS and intracranial tumors is mainly related to teratomas, and dermoid cysts [39].

The diagnosis of KFS is usually incidental, the cervical spine X-rays show scoliosis, vertebral fusion, and instability, spinal CT with three-dimensional reconstruction is useful in surgical planning, and the spinal MRI is useful to detect neurologically (spinal compression, stenosis, and syringomyelia). Surgical treatment is based on the detection and management of associated systemic alterations, only 43% of patients will require decompression and spinal stabilization depending on risk patterns determined by Samartzis classification. Our patient did not require surgical management of this malformation due to the lack of clinical repercussion [40].

In conclusion, NEC prognosis is generally favorable, but a significant proportion of individuals undergoing partial resection experience recurrence. The association between KFS and NEC can be related to the persistence of embryological structures. The correct diagnostic approach must be carried out to choose the optimal surgical approach. Therefore, about our experience, in the points of **Table 1**, we describe the fundamental aspects in the management of this pathology.

5. Conclusion

Because these intracranial tumors are uncommon, studies that compare the benefits of various management strategies about outcomes and prognosis factors are lacking. Therefore, the level of evidence of management recommendations is low. However, we consider the knowledge of these entities important, so we determine the important characteristics in the diagnosis, management, and prognosis to establish a comprehensive review of these neoplasms.

Acronyms and abbreviations

CA 19-9	Carbohydrate antigen 19-9
CNS	Central nervous system
CPP	Choroid plexus papilloma
CT	Computed tomography
EMG	Electromyography
FLAIR	Fluid attenuation inversion recovery
HFS	Hemifacial spasm
KFS	Klippel-Feil syndrome
MRI	Magnetic resonance imaging
NEC	Neuroenteric cysts
RDD	Rosai-Dorfman disease

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