Radiology Case Reports 16 (2021) 1153–1157

Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/radcr

Case Report

Presacral medulloepithelioma with peritoneal carcinomatosis in an 11-year-old boy: An extremely rare association ☆

Gabriel Soares Mendonça, MD[a,*], *Camilla Burla Artiles, MD*[b], *Glícia Campanharo Malheiros, MD*[c], *Viviane Brandão Amorim, MD, PhD*[a,b]

[a] *Department of Diagnostic Radiology, National Cancer Institute José Alencar Gomes da Silva - INCA, Rio De Janeiro, Brazil*

[b] *Department of Diagnostic Radiology, Fleury Group, Rio De Janeiro, Brazil*

[c] *Department of Pathology, National Cancer Institute José Alencar Gomes da Silva - INCA, Rio De Janeiro, Brazil*

ARTICLE INFO

Article history:
Received 20 September 2020
Revised 18 December 2020
Accepted 25 December 2020

Keywords:
Child
Computed tomography
Magnetic resonance imaging
Medulloepithelioma
Presacral medulloepithelioma
Peritoneal carcinomatosis

ABSTRACT

Medulloepithelioma is a rare and highly malignant tumor of infancy and early childhood, and classified as a primitive neuroectodermal tumor. Considering that most cases occur in the central nervous system, development in atypical sites associated with secondary comorbidities, such as peritoneal carcinomatosis, becomes an extremely rare association due to its high morbimortality. This study reports a rare case of peripheral medulloepithelioma with peritoneal carcinomatosis in an 11-year-old boy, with a 4-year history of intestinal constipation alternated with fecal incontinence, taken to the emergency room due to increasing abdominal pain and urinary retention. This report aims to contribute to a better understanding of this rare pathology, as well as assist in the establishment of early diagnosis and treatment.

Introduction

First described by *Bailey and Cushing* in 1926, medulloepithelioma (ME) is a rare and highly malignant tumor of infancy and early childhood, classified as a primitive neuroectodermal tumor [1].

Usually located either in the central nervous system (CNS) or in the intraocular region, the location of ME is a significant survival prognostic factor. Tumors in peripheral locations or in the CNS demonstrate high potential for aggressiveness, with reported average survival rate of less than 1 year after diagnosis [1–3].

Peripheral ME, such as those found in the pelvic cavities, has been rarely reported, and the optimal therapeutic approach as well as treatment are still not well established [4–8].

Considering that most cases occur in the CNS, the patient usually dies from complications related to the growth and spread of the tumor [1–3]. Development of ME in atypical sites associated with secondary comorbidities, such as peritoneal carcinomatosis, is an extremely rare association.

☆ Competing Interests: None.

* Corresponding author.
E-mail address: soaresm.gabriel@gmail.com (G.S. Mendonça).

https://doi.org/10.1016/j.radcr.2020.12.060

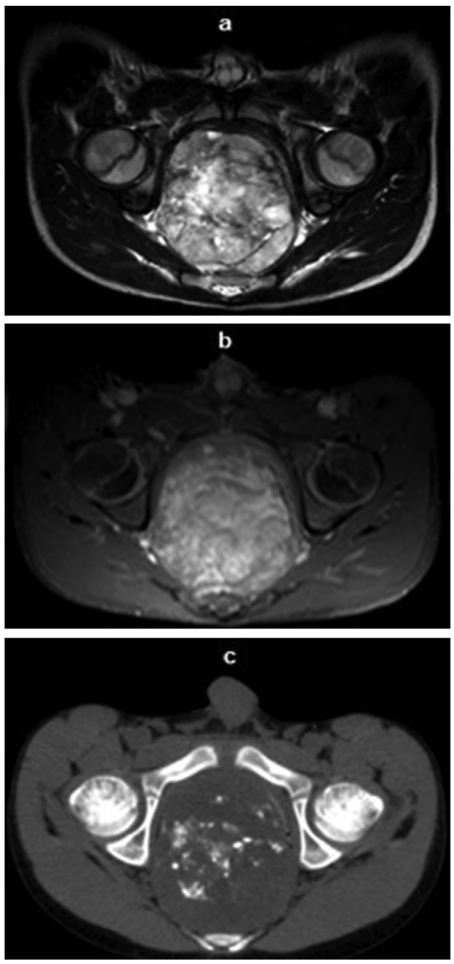

Fig. 1 – Axial contrast-enhanced T2 (a) and T1 (b) weighted imaging, showing the large presacral heterogeneous mass, extending inferiorly to the ischiorectal fossa. In addition, displacement and compression of the rectus with significant bladder compression are also seemed. The lesion presents areas of cystic / necrotic degeneration and contrast-enhanced solid component. The CT scan (c) without endovenous contrast, axial view, shows the calcified pattern of the lesion.

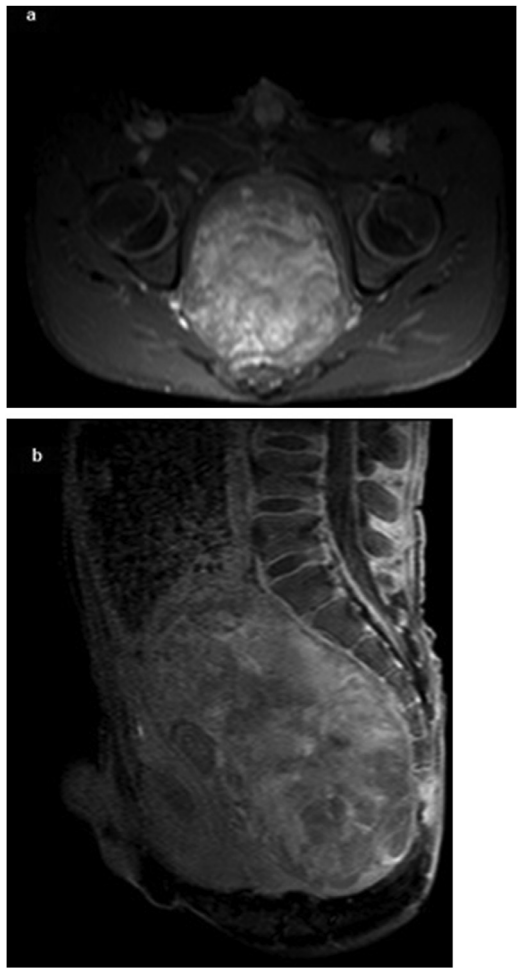

Fig. 2 – Axial T1 (a) and sagittal T1 Fat-Sat (b) weighted imaging, with endovenous contrast use, showing the three largest dimensions of the tumor. The tumor occupies the entire pelvic region, extending inferiorly from L5 / S1, displacing the bladder anteriorly and superimposing the intestinal loops.

Review of the literature reports only 1 case of ME with secondary peritoneal carcinomatosis, but with no illustrations [7]. This study reports a rare case of peripheral ME occurring in the right presacral region with concomitant peritoneal carcinomatosis.

Case Report

The paper describes the case of an 11-year-old boy with a 4-year history of intestinal constipation alternating with fecal incontinence taken to the emergency room due to increasing abdominal pain and urinary retention.

The CT scan showed an expansive calcified mass (15 × 10 × 8 cm), in the right presacral region, extending downwards to the rectal ischium, compressing pelvic structures, as well as displacing and collapsing the rectum (Fig. 1).

The MRI scan showed an expansive, encapsulated, lobulated solid-cystic mass, with predominance of the cystic area, high signal at T1 sequence indicative of hematic or high protein content, and the peripheral solid part with heterogeneous contrast enhancement. This mass had an important local effect, with anterior displacement of the bladder and of left side of the rectum, causing an important accumulation of intestinal residue (Figs. 1 and 2).

An open biopsy of the lesion was performed. The pathology revealed a malignant neoplasm characterized by cell proliferation with papillary, tubular, and trabecular arrangement, mimicking an embryonic neural tube.

The neoplastic cells showed a diffuse strong positivity for GFAP (glial fibrillary acidic protein), focal weak positivity for Cytokeratin EMA, and AE1/AE3, negativity for CK7 e PLAP

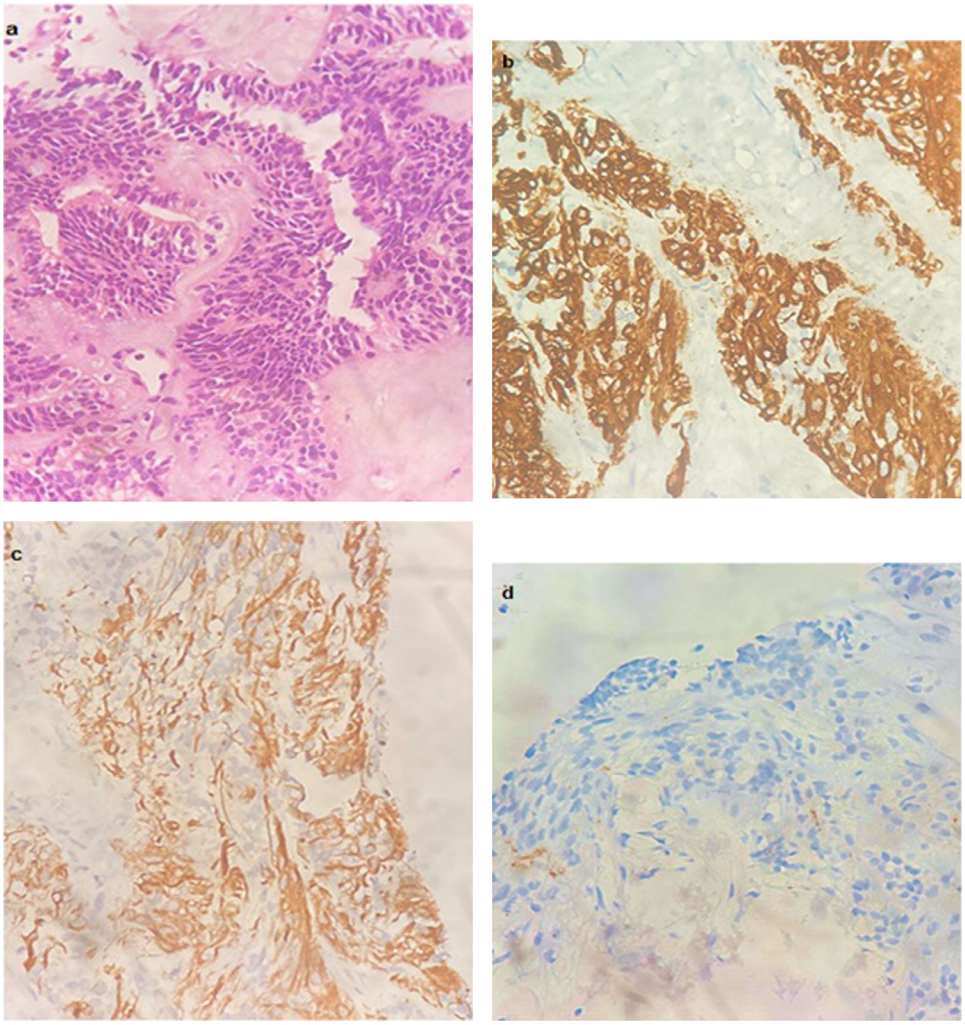

Fig. 3 – Pathology of the tumor at diagnosis. Papillary and tubular patterns, with trabecular arrangement and cell proliferation represent the appearance of medulloepithelioma; HE 10x (a). Diffuse strong positivity for GFAP 40x (b), focal weak positivity for AE1/AE3 40x (c), and Cytokeratin EMA 40x (d).

(placental-like alkaline phosphatase; Fig. 3). The diagnosis was ME.

Initially, debulking surgery of the presacral lesion was performed by exploratory laparotomy, followed by hyperthermic intraperitoneal chemotherapy (HIPEC protocol) with no further complications. After surgery, chemotherapy was initiated using the nonmetastatic GALOP protocol with alternated use of Doxorubicin, Vincristine, Cyclophosphamide and Mesna with Ifosfamide and Etoposide.

After the conclusion of the initial chemotherapy cycle 3 months after the first surgery, a second surgery was performed via exploratory laparotomy to remove the residual presacral tumor.

Maintaining the initial protocol, chemotherapy treatment continued with the addition of radiotherapy (45 Gy + 10.8 Gy) for 2 more months, in which period the patient presented signs of peritoneal irritation, and new imaging showed extensive peritoneal carcinomatosis (Fig. 4).

The patient started chemotherapy rescue protocol, using a second-line chemotherapy regimen with Carboplatin, Etoposide and Topotecan for 4 months.

A third surgery was performed after the completion of second-line chemotherapy, aiming at reducing peritoneal recurrence. Splenectomy and surgical removal of most of the peritoneal lesions were performed, with compromised surgical margins.

Once the peritoneal recurrence was characterized as non-surgical, the patient started third-line chemotherapy, but he died at home 6 months later, due to progression of peritoneal carcinomatosis.

Discussion

ME is a rare neoplasm, highly malignant, derived from the primitive medullary plate and neural tube. Classically occurring in the intraocular region or in the CNS [1–3], it is essentially a childhood tumor with no specific gender predilection. Cases of extracranial ME are scarce, and few reports of pelvic tumor have been documented [4–7].

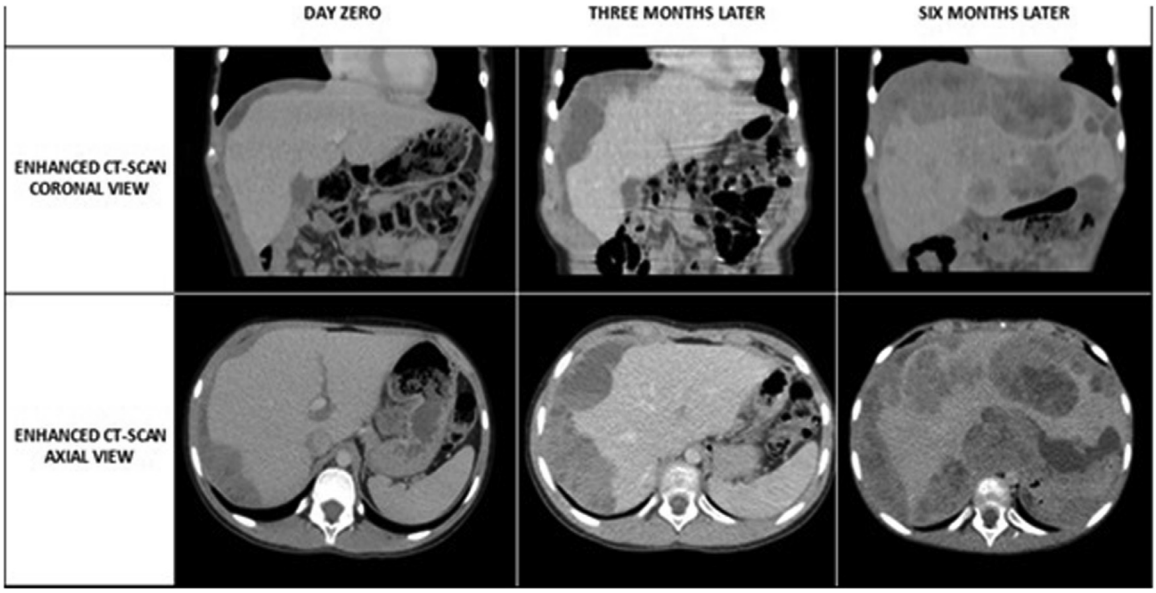

Fig. 4 – Radiological aspects of the peritoneal carcinomatosis progression on enhanced CT-scan since its diagnosis. The image shows the expressive increase in the dimensions of expansive peritoneal tissue, with soft-tissue density and heterogeneous enhancement, where the largest components are next to the hepatic surface, causing compression on the adjacent parenchyma, characterizing peritoneal carcinomatosis, secondary to medulloepithelioma.

In 1926, *Bailey and Cushing* [1] originally classified ME as the most primitive neoplasm of the CNS. Later on, *Rorke* [3] et al. classified the tumor into 2 subtypes: (1) ME not otherwise specified; (2) ME with differentiation into astrocytes, ependymal cells, neuronal cells, mixed cellular elements, oligodendrocytes, and others groups, including melanin and mesenchymal cell [5].

The ME originating from primitive undifferentiated cells usually involves frontal, occipital, parietal, temporal lobes or the posterior fossa [5,7]. Other reported sites include the cerebral hemispheres, brain stem, cerebellum, and peripheral sites. The sacral and presacral locations seem to be originated from undifferentiated embryonic cells, forming the presacral remnants of the neurenteric canal [4–7].

Microscopically, ME shows pseudo-stratified primitive epithelium with multilayered rosettes and laminated structures. It presents polygonal and large cells, with hyper chromatic nuclei and scant cytoplasm. Mitotic cells are also common [5].

The stratified epithelium is stained by periodic acid-Schiff stain (PAS), due to the presence of a coating around the basal membrane [5].

The astroglial and ependymal differentiation lines are positive for glial fibrillary acidic protein (GFAP) staining, and neuronal differentiation lines are positive for synaptophysin or neuronal specific enolase (NSE) [5].

Radiological appearance of ME varies according to the tumor location. In noncontrast CT scans, it can appear as a well-circumscribed, isodense, or hypodense tumor, with variable heterogeneity and calcification [8]. Typical MR imaging presents isointense or hypointense signals in T1-weighted imaging, hyperintense signal in T2-weighted imaging and usually not enhanced with gadolinium contrast. Recent studies have associated post contrast enhancement, particularly in solid parts, with more aggressive nature and higher recurrence possibilities [8].

Since the tumor presents high mortality rates, most children have an average survival rate of 1 year after being diagnosed [5–8]. Considering that most cases occur in the CNS, development in atypical sites associated with secondary comorbidities, such as peritoneal carcinomatosis, becomes an extremely rare association, due to its high morbimortality and the usual outcome of the disease [5–8].

The optimal management of ME is unknown. Complete resection followed by radiation therapy or chemotherapy are available options [5–7].

Aggressive chemotherapy and autologous bone marrow transplant seem to be a promising strategy but need further research [5–7].

The evaluation of radiation therapy must be cautious and careful, considering that most cases of ME occur in the CNS of children under 2 years of age and that radiation can cause developmental and cognitive impairment [6].

REFERENCES

[1] Bailey P, Cushing H. A classification of the tumour of the glioma group on a histogenic basis with correlated study of prognosis. Philadelphia, London and Montreal: Lippincott Williams & Wilkins; 1926.

[2] Louis DN, Perry A, Reifenberger G, et al. The 2016 World Health Organization Classification of tumors of the central nervous system: a summary. Acta Neuropathol 2016;131:803–20.

[3] Rorke LB, Gilles FH, Davis RI, et al. Revision of the World Health Organization classification of brain tumors for child hood brain tumors. Cancer 1985;56:1869–86.

[4] Figarella-Branger D, Gambarelli D, Perez-Castillo M, et al. Ectopic intrapelvic medulloepithelioma: case report. Neuropathol Appl Neurobiol 1992;18:408–14.
[5] Donner LR, Teshima I. Peripheral medulloepithelioma: an immunohistochemical, ultrastructural, and cytogenetic study of a rare, chemotherapy-sensitive, pediatric tumor. Am J Surg Pathol 2003;27:1008 1012.
[6] Norris LS, Snodgrass S, Miller DC, et al. Recurrent central nervous system medulloepithelioma: response and outcome following marrow ablative chemotherapy with stem cell rescue. J Pediatr Hematol Oncol 2005;27:264.
[7] De Pasquale MD, De Ioris MA, Gallo A, et al. Peripheral medulloepithelioma: a rare tumor with a potential target therapy. J Transl Med 2014;12:49.
[8] Molloy PT, Yachnis AT, Rorke LB, et al. Central nervous system medulloepithelioma: a series of eight cases including two arising in the pons. J Neurosurg 1996;84:430–6.