Radiology Case Reports 15 (2020) 2433–2439

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

Case Report

Perinatal atypical teratoid/rhabdoid tumor involving the deep ear structures and complicated by arterial infarction ☆,☆☆,★,★★

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ARTICLE INFO

Article history:
Received 15 July 2020
Revised 25 August 2020
Accepted 28 August 2020

Keywords:
Atypical teratoid rhabdoid tumor
Inner ear
Skull base
Infarction

ABSTRACT

Atypical teratoid/rhabdoid tumor is a malignant pediatric brain tumor. Unusual invasive behavior of the dura and bony involvement of the deep ear structures and rapid progression in size complicated by arterial infarction have not been described before. A newborn girl presented with increased intracranial pressure. Medical imaging revealed a large mass centered in the left cerebellopontine angle and left middle cranial fossa with large supratentorial components associated with destruction of the left petrous bone with involvement of the inner and middle ear structures. Shortly, the tumor rapidly progressed in size and complicated by left middle cerebral artery territory infarction. The patient passed away after a short hospital course. This case report illustrates how rapid and aggressive the natural history of atypical teratoid/thabdoid tumor can be with unusual skull base destruction and deep ear structures involvement.

☆ Informed Consent: The consent was not required from institutional review board in our institute due to the anonymized and retrospective nature of this report.

☆☆ Funding: No funds were received in support of this study.
★ Competing Interests: There are no conflicts of interest.
★★ Acknowledgments: No benefits in any form have been received or will be received from a commercial party related directly or indirectly to the subject of this paper.

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https://doi.org/10.1016/j.radcr.2020.08.060

Introduction

An Atypical teratoid/rhabdoid tumor (AT/RT) is a highly malignant tumor and classified as WHO grade IV [1]. AT/RT is a common pediatric tumor in the age group of fewer than 3 years [2]. Moreover, the prior series considered it as the most common central nervous system (CNS) malignancy below the age of 6 months [3,4]. AT/RT has been described in various locations within the central nervous system. The posterior fossa

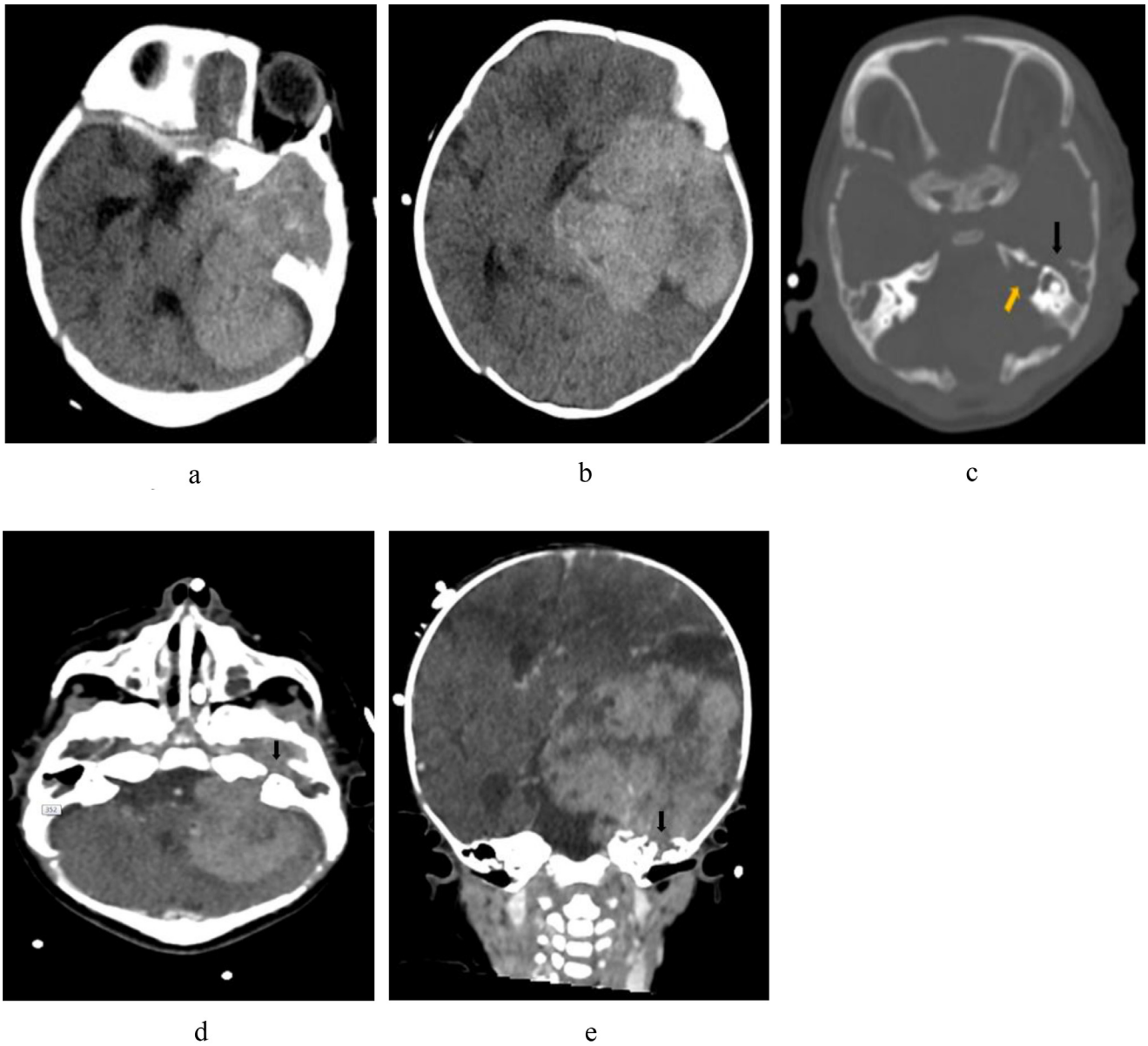

Fig. 1 – Selected cross-sectional axial images without intravenous contrast at presentation, at the level of the pons (a), third ventricle (b) and petrous bones (c). (a, b) There is a sizeable hyperdense mass centered in the left cerebellopontine angle and left middle cranial fossa with a large supra-tentorial component associated with mass effect on the adjacent structures with midline shift and hydrocephalus (c,d, and e). There is an extension into the left internal auditory canal and involvement of both inner ear structures (yellow arrow in c) and middle ear cavity with the destruction of the tegmen (black arrows in c, d, and e)

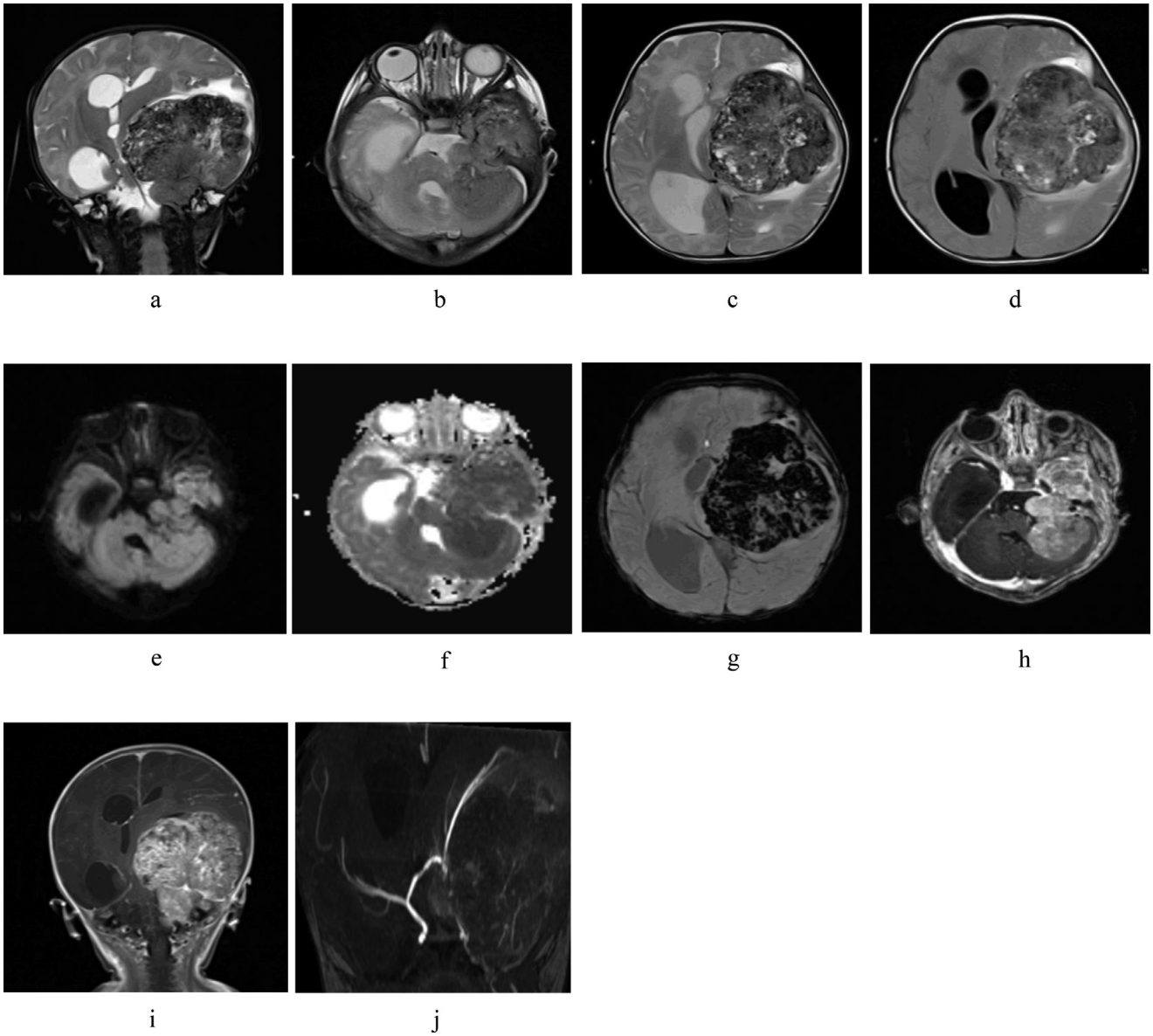

Fig. 2 – Selected multi-planar multi-sequential MRI images of the brain tumor one day after the initial CT scan. Coronal T2 (a), Axial T2 (b), Axial T2 (c), Axial FLAIR (d), DWI (e), ADC (f), SWAN (g), Axial T1 withGd (h), Coronal T1 withGd (i) and Coronal MIP TOF MRA (j). (a-j) There is a large mass centered in the left cerebellopontine angle and left middle cranial fossa with a significant supra-tentorial component. The mass is demonstrating heterogeneous signal intensity on T2 WI with small peripheral cysts. There is an extensive blooming artifact, representing the hemorrhagic components. There is an avid heterogeneous enhancement on postcontrast images with diffusion restriction in DWI/ADC mapping. The tumor invading the terminal segment of left internal carotid artery and causing mild narrowing in A1 segment of the left anterior cerebral artery

is a common location of involvement [5,6]. Bony extension and destruction are rarely described in malignant brain tumors [7–11]. Here we present an unusual case of large AT/RT lesion centered in the left cerebellopontine angle and left middle cranial fossa with large supra-tentorial component associated with destruction of the left petrous bone involving the inner and middle ear structures, and complicated by arterial infarction.

Case report

A newborn baby girl, a product of uneventful non-instrumented spontaneous vaginal delivery, was born in an outside healthcare facility with left-sided facial weakness manifested immediately after birth as left-sided dropping mouth and inability to close her left eyelid. Although an

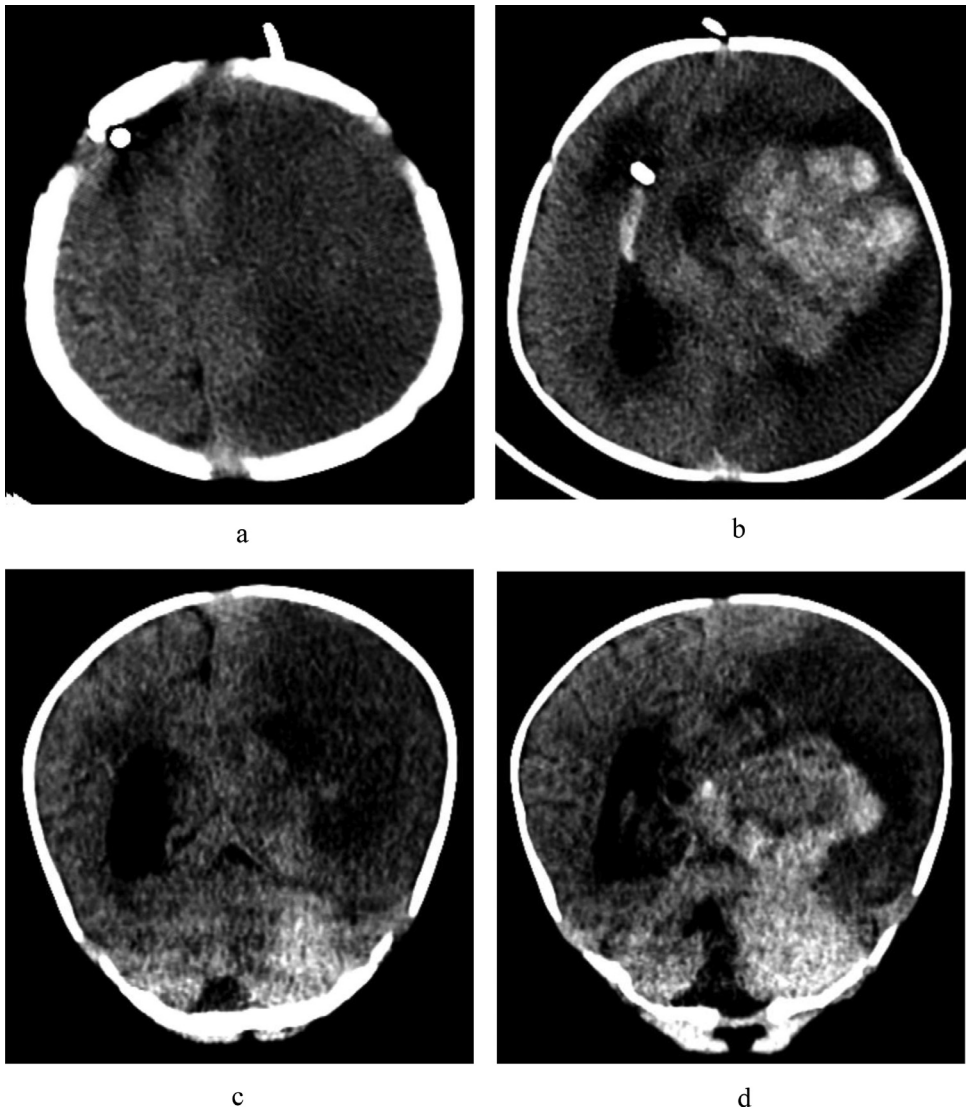

Fig. 3 – Selected cross-sectional axial and coronal images without contrast, 4 days after the previous MRI showed interval development of large hypo-density in left middle cerebral artery territory representing left acute infarction, likely due vascular invasion/compression by the tumor. There is also an interval increase in the size of the tumor and mild worsening of the hydrocephalus

outpatient follow-up appointment had been arranged to evaluate the left facial weakness noted perinatally, the patient was admitted to the hospital at 1-month of age with respiratory distress. During this admission, she was starting to have decreased level of consciousness, unequal pupils, and signs of increased intracranial pressure. She was immediately admitted to the Pediatric Intensive Care Unit. An urgent brain computed tomography (CT) was done revealing large left-sided hemorrhagic brain tumor exerting midline shift and acute hydrocephalus (Fig. 1). The patient underwent external ventricular drain insertion.

The patient was transferred to our institute pediatric intensive care unit for further management. The day after the initial CT, MRI of the brain showed a large mass lesion centered in the left cerebellopontine angle and left middle cranial fossa with large supra-tentorial components associated with destruction of the left petrous bone involving the inner and middle ear structures (Figs. 1c and 2). Four days after MRI, follow-up brain CT scan showed progression in tumor size with interval development of left middle cerebral arterial territorial infarction (Fig. 3).

During the hospital course, she underwent left frontotemporal mini craniotomy for a biopsy and mass debulking, which revealed high-grade undifferentiated neoplasm, consistent with Atypical Teratoid/Rhabdoid Tumor (Fig. 4). The surgical note described the tumor to be invading the brain parenchyma. On immune-histochemical examination, the lesion was positive SMA, EMA, and P53. There is loss of INI1/BAF47 immunostaining due to inactivation of the SMARCB1 gene. CK (AE1/AE3) and GFAP were negative. Synaptophysin was focal. Ki-67 was 60%.

A few days later, a follow-up CT brain revealed further progression in tumor size (Fig. 5). The tumor board meeting was held, concluding no role of surgery, chemotherapy, or

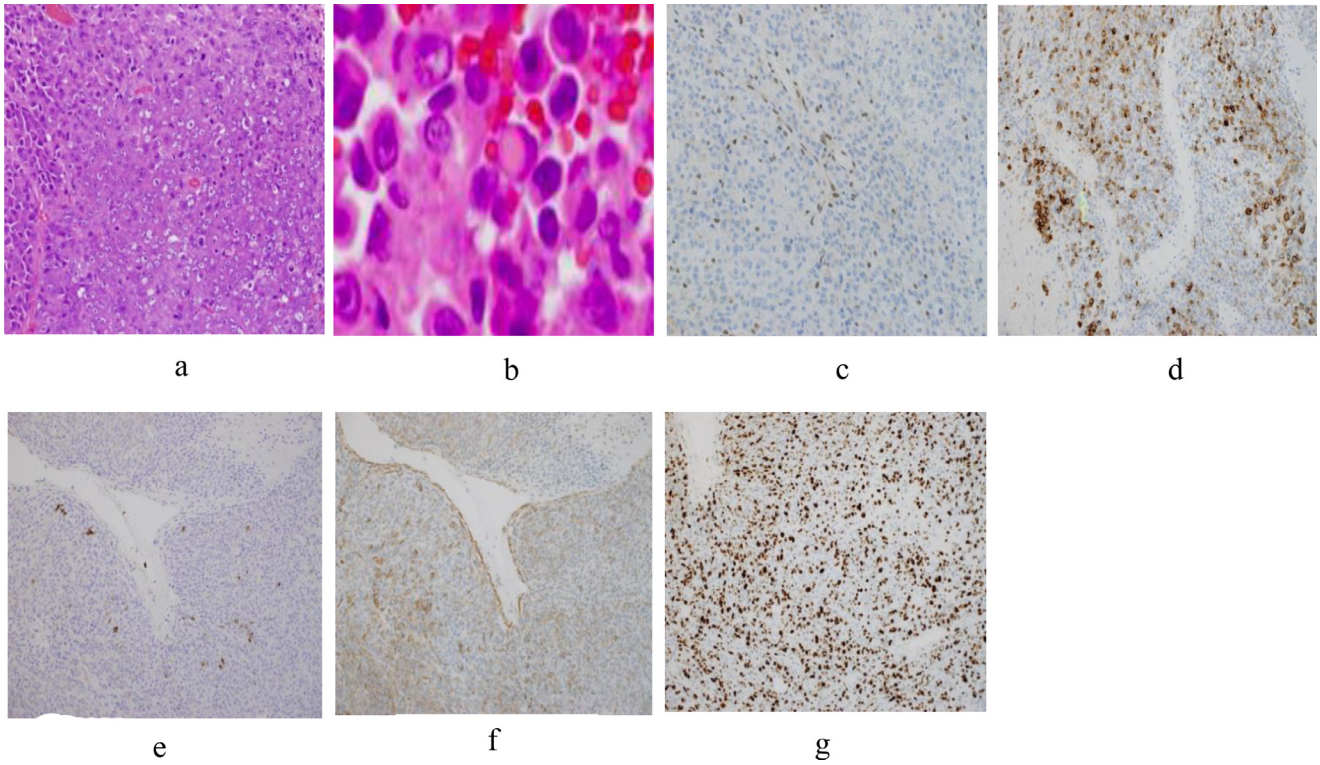

Fig. 4 – Under-microscopy Hematoxylin and Eosin stain (a,b) and Immunohistochemical stain (c-g) shows (a) Rhabdoid cells with abundant eosinophilic cytoplasm, vesicular chromatin, and prominent nucleoli. Abundant mitotic figures and apoptosis are noted in this figure. (b) Eosinophilic globular cytoplasmic inclusion (arrow). (c) Loss of expression of INI-1 in nuclei of tumor cells with retained expression in the blood vessels and scattered inflammatory cells. (d) Strong focal expression of EMA (indicates epithelial differentiation). (e) Scattered cells are expressing synaptophysin (indicates neuroectodermal differentiation). (f) Strong focal expression of SMA (indicates mesenchymal differentiation). (g) Ki-67 proliferation index is approximately 60%

radiotherapy. Seventeen days after the admission, the patient passed away at the age of 2 months.

Discussion

AT/RT is a rare malignant central nervous system that was first described in 1987 [12]. It is classified as World Health Organization grade IV in 2000 [1]. It is predominately affecting the pediatric age group, especially below the age of 3 years [2]. It represents around 2% and 7% of the primary central nervous system primary tumors in the childhood age group and below the age of 3 years, respectively [13].

It has been described in various intra-axial and extra-axial locations within the central nervous. The posterior fossa is a common site of involvement, especially the cerebellopontine angle [5,6]. This tumor occurs sporadically. However, it can be associated with other rhabdoid tumor-related syndromes [4]. There is no specific clinical presentation. Signs of increased intracranial hypertension and involvement of the cranial nerves had been described [14]. There are no specific imaging characteristics. Calcifications, hemorrhage, necrosis, cysts formation, heterogeneous enhancement postcontrast administration have all been described in cross-sectional images [15]. Variable T1 and T2 signal intensities, diffusion restriction on diffusion weighted imaging (DWI)/apparent diffusion coefficient (ADC) mapping, blooming artifact due to hemorrhage, necrotic areas, peripheral cysts formation with enhancing wall and heterogeneous avid enhancement postcontrast administration have been described in magnetic resonance imaging [15]. Brain parenchymal, dural, and bone invasions are recognized as aggressive features of the tumor [16]. Even highly aggressive primary brain tumors rarely demonstrate dural or bone invasion because the dura is considered a protective barrier for tumor spread and involvement of the calvarium [17]. The percentage of bone involvement had been estimated in a prior study from 91 patients diagnosed with CNS AT/RT; 5 patients (6.6%) had calvarial involvement with temporal bone involvement was seen in 2 of them [18].

Perinatal RT is rare and perinatal CNS AT/RT is even rarer. A systemic review from the literature was conducted for a 40-year interval, which revealed 72 RT cases. They have classified the cases based on the location of the tumor into 3 groups: extra-renal non-CNS, renal, and CNS. The majority of the cases were extra-renal non-CNS RT (33/72) and renal RT (27/72). CNS RT was the minority group (12/72) [19]. This case report is a case of perinatal/congenital AT/RT due to the presence of

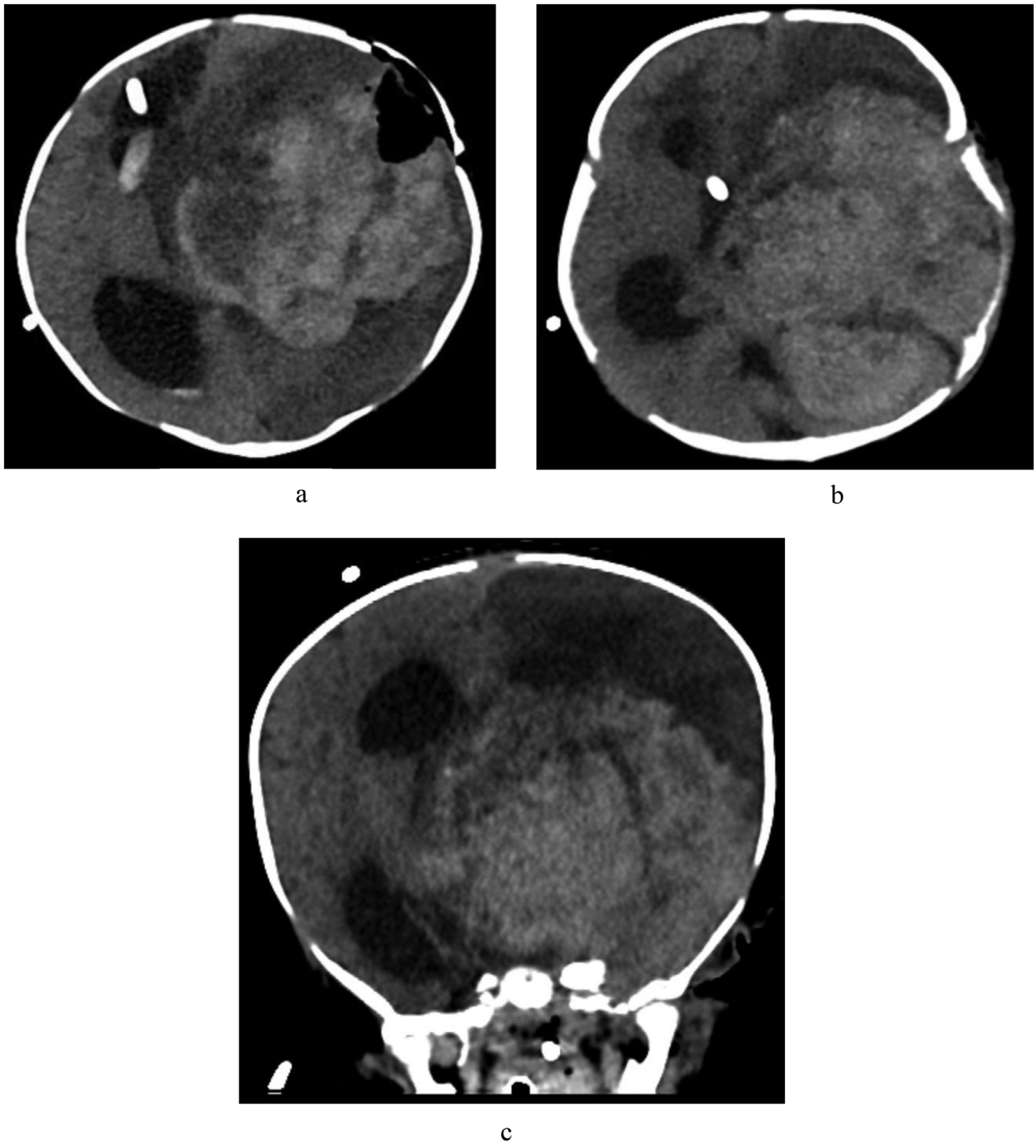

Fig. 5 – Postsurgical selected cross-sectional axial and coronal images without contrast showed further increase in the tumor size, worsening of the hydrocephalus and midline shift

left facial weakness documented at the time of birth, which indicates that an underlying lesion was likely present at that time.

Currently, there is no universally standard therapeutic regimen for AT/RT due to its rarity and being a relatively new recent pathology. However, the central component of the treatment has been surgical removal of the tumor followed by radiation and chemotherapy. Radiation treatment, including proton therapy, has demonstrated a favorable outcome in the treatment of AT/RT by limiting local and metastatic tumor progression, especially if implanted early [20].

To best of our knowledge, there is no prior case report of unusual AT/RT behavior with features of petrous involvement, namely the inner and middle ear structures, with rapid progression in size complicated by cerebral infarction. AT/RT carries poor prognosis when compared with other pediatric brain tumors such as medulloblastoma, other embryonal tumors, and ependymoma [21]. Age below 2 years has been demonstrated to be a poor prognostic factor likely due to different treatment options feasibility in an older age group. Moreover, tumor size, brain parenchymal invasion, and metastasis are determinants of the resectability, and hence prognostic determinants [22].

REFERENCES

[1] Kleihues P. Pathology and genetics of tumours of the nervous system (2000), Lyon: IARC Press; 2000. ISBN 92 83 22409 4.
[2] Rorke LB, Packer RJ, Biegel JA. Central nervous system atypical teratoid/rhabdoid tumors of infancy and childhood: definition of an entity. J Neurosurg 1996;85:56–65.
[3] von Hoff K, Hinkes B, Dannenmann-Stern E, Bueren AO, Warmuth-Metz M, Soerensen N, et al. Frequency, risk-factors and survival of children with atypical teratoid rhabdoid tumors (AT/RT) of the CNS diagnosed between 1988 and 2004, and registered to the German HIT database. Pediatr Blood Cancer 2011;57:978–85.
[4] Woehrer A, Slavc I, Waldhoer T, Heinzl H, Zielonke N, Czech T, et al. Incidence of atypical teratoid/ rhabdoid tumors in children: a population-based study by the Austrian Brain Tumor Registry, 1996 –2006. Cancer 2010;116:5725–32.
[5] Reddy AT. Atypical teratoid/rhabdoid tumors of the central nervous system. J Neuro-Oncol 2005;75:309–13.
[6] Yun-Sik Dho S-KK, Lee JY, Phi JH. Investigation of the location of atypical teratoid/rhabdoid tumor. SpringerLink: Springer Berlin Heidelberg; 2015.
[7] Gheyi V, Hui FK, Doppenberg EM, Todd W, Broaddus WC. Glioblastoma multiforme causing calvarial destruction: an unusual manifestation revisited. AJNR Am J Neuroradiol 2004;25:1533–7.
[8] Murphy MN, Korkis JA, Robson FC, Sima AA. Gliosarcoma with cranial penetration and extension to the maxillary sinus. J Otolaryngol 1985;14:313–16.
[9] Osborn RE, Ley CE. Astrocytoma with calvarial erosion. AJNR Am J Neuroradiol 1986;7:178.
[10] Schuss P, Ulrich CT, Harter PN, Tews DS, Seifert V, Franz K. Gliosarcoma with bone infil- tration and extracranial growth: case report and review of litera- ture. J Neuro-oncol 2011;103:765–70.
[11] Woodruff WW Jr, Djang WT, Voorhees D, Heinz ER. Calvarial destruction: an unusual manifestation of glioblastoma multiforme. AJNR Am J Neuroradiol 1988;9:388–9.
[12] Lefkowitz IB, Rorke LB, Packer RJ, Sutton LN, Siegel KR, Katnic RJ. Atypical teratoid tumor of infancy: definition of an entity. Ann Neurol 1987;22:448–9.
[13] Rickert CH, Paulus W. Epidemiology of central nervous system tumour in childhood and adolescence based on new WHO classification. Childs Nerv Syst 2001;17:503–51.
[14] Fenton LZ, Foreman NK. Atypical teratoid/rhabdoid tumour of the central nervous system in children: an atypical series and review. Pediatr Radiol 2003;33:554–8.
[15] Kanoto M, Toyoguchi Y, Hosoya T, Kuchiki M, Sugai Y. Radiological image features of the atypical teratoid/rhabdoid tumor in adults: a systematic review. Clin Neuroradiol 2014.
[16] Arslanoglu A, Aygun N, Tekhtani D, Aronson L, Cohen K, Burger PC, Yousem DM. Imaging findings of CNS atypical teratoid/rhabdoid tumours. Am J Neuroradiol 2004;25:476–80.
[17] Woodruff WW Jr, Djang WT, Voorhees D, Heinz ER. Calvarial destruction: an unusual manifestation of glioblastoma multiforme. AJNR Am J Neuroradiol 1988;9:388–9.
[18] Warmuth-Metz M, Bison B, Gerber NU, Pietsch T, Hasselblatt M, Frühwald MC. Bone involvement in atypical teratoid/rhabdoid tumors of the CNS. AJNR Am J Neuroradiol 2013;34(10):2039–42. doi:10.3174/ajnr.A3554.
[19] Isaacs H Jr. Fetal and neonatal rhabdoid tumor. J Pediatr Surg 2010;45(3):619–26. doi:10.1016/j.jpedsurg.2009.12.011.
[20] Nesvick Cody L, Rao Amulya A Nageswara, Raghunathan Aditya, Biegel Jaclyn A, Daniels David J. Case-based review: atypical teratoid/rhabdoid tumor. Neuro-Oncology Practice June 2019;6(Issue 3):163–78. Pages https://doi.org/10.1093/nop/npy037 .
[21] MacDonald TJ, Rood BR, Santi MR, Vezina G, Bingaman K, Cogen PH, et al. Advances in the diagnosis, molecular genetics, and treatment of pediatric embryonal CNS tumors. Oncologists 2003;8:174–86.
[22] Dufour C, Beaugrand A, Le Deley M, Bourdeaut F, André N, Leblond P, et al. Clinicopathologic prognostic factors in childhood atypical teratoid and rhabdoid tumor of the central nervous system. Cancer 2011;118(15):3812–21.