

Case Report

Unusual radiological characteristics of teratoid/rhabdoid brain tumor in children.

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Abstract:

We report a case of atypical teratoid rhabdoid brain tumor for 4 months old male child, who presented with unusual radiological findings, that can be confused with other brain tumors, so we highlight these unusual imaging features to aid in making correct diagnosis.

Keywords: atypical teratoid–rhabdoid tumor, brain tumor, children, medulloblastoma.

A typical teratoid/rhabdoid tumor (AT/RT) of the central nervous system is an extremely rare and highly aggressive tumor of early childhood¹. AT/RT of the brain was described as a unique entity in the late 1980s².

It usually affects very young children, although it has been reported in adults as well. It is a highly aggressive neoplasm, often has central nervous system dissemination, does not respond to therapy and typically is fatal within 1 year³.

The differential diagnosis includes medulloblastoma, primitive neuroectodermal tumor (PNET), choroid plexus carcinoma, and malignant glioma⁴. This is of clinical importance because the prognosis of a patient with an AT/RT is worse than that of a patient with PNET/medulloblastoma despite aggressive surgical treatment, with or without adjuvant chemotherapy and radiation therapy⁵.

Primitive neuroectodermal tumors and AT/AR are childhood CNS neoplasm that may display similar characteristics on routine histological analysis and on neuroimaging. There are no specific or pathognomonic imaging features found in the literature for intracranial AT/RT that can clearly differentiate these tumors from other pediatric brain tumors, and neuroradiologists rarely mention AT/RT in their differential diagnosis.

Pediatric neurology showed that primitive neuroectodermal tumors are radiology less heterogeneous than AT/RT and occur in younger ages, usually below two years. However, a high tendency towards large size, hyperdense solid component on the CT scan with calcification, hemorrhage, necrosis, and subarachnoid spread suggest that AT/RT should be considered in the differential diagnoses.

According to the current WHO classification, diagnosis and distinction is made by immunohistochemical methods. Diagnosis of AT/RT is based on morphologic criteria that is, the presence of rhabdoid tumor cells and immunohistochemical methods. Rhabdoid tumor cells almost always express epithelial membrane antigen (EMA) and vimentin, and to a variable extent smooth muscle extent, glial fibrillary acidic protein, neuronal antigens, cytokeratin (CK) and for BAF47 antibody against the hSNF5/INI1 protein. This unique immunohistochemical profile of an AT/RT helps to distinguish it from other CNS tumors with specific morphological and different immunological characteristics⁶. However, application of INI1 immunostaining is also very helpful for distinguishing PNET from AT/RT as well⁷. Primitive neuroectodermal tumors and AT/RT are childhood CNS neoplasm that may display similar characteristics on routine histological analysis and on neuroimaging. Pediatric neurology showed that primitive neuroectodermal tumors are radiology less heterogeneous than AT/RT and occur in younger age, usually below two years.

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This case presents unusual radiological findings of AT/RT

Case Report:

A male child of four month had been presented to the pediatric clinic with complaints of sudden onset of bulging anterior fontanel. This child, who came after normal vaginal delivery, was previously healthy, but two weeks later he developed squint. Apart from that clinical examination was unremarkable.

Brain CT scan showed 9.5x5.3X7cm mixed density- mainly hypo dense- mass lesion in the RT temporoparietal region (fig 1).

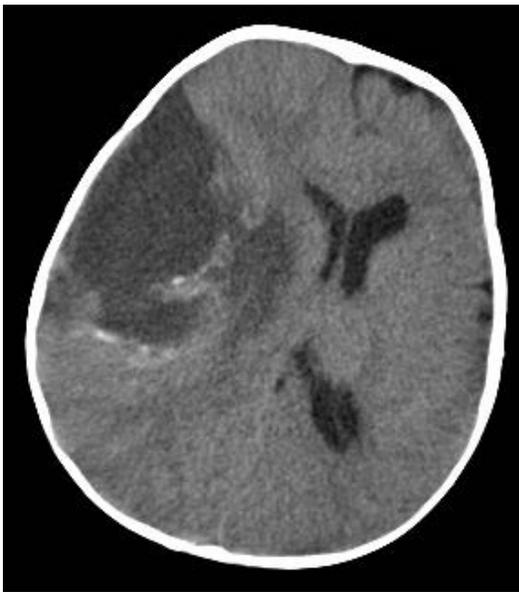


Fig1: axial CT scan showed mixed density mass lesion in the RT temporoparietal

After 50 ml intravenous injection of non ionic contrast media (iopamiro300), this mass lesion showed variable heterogeneous pattern of enhancement, the small solid part showed enhancement that contained a small hyper dense areas which may represent hemorrhage. In our case, the patient underwent preoperative MRI using 1.5-T units. The

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applied MRI protocol included T2- and T1-weighted images in different imaging planes, with and without IV gadopentetate dimeglumine (Gd). MRI images of the entire neuraxis, with Gd-enhanced sequences were obtained. Two pediatric neuroradiologists evaluated MRI and CT scan examinations. Spinal MRI should be performed to exclude tumor dissemination through the whole neural axis Preoperative precontrast MRI images showed mixed intensity mainly hypo intense in T1WI and flair (fig 2, 3) and hyperintense in T2WI (fig 4), filled with fluid of C.S.F intensity.

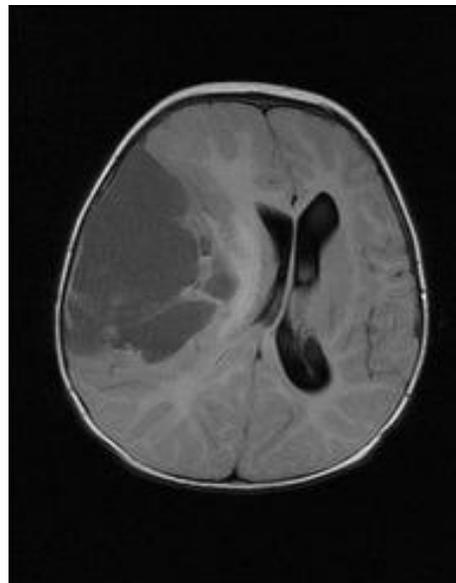


Fig 2 : axial MRI FLAIR sequence, showed Mass effect on the RT lateral ventricle.

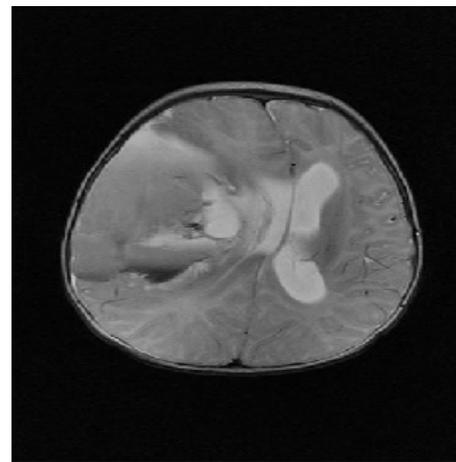


Fig 3 : axial T2WI, showed cystic component of the tumor.

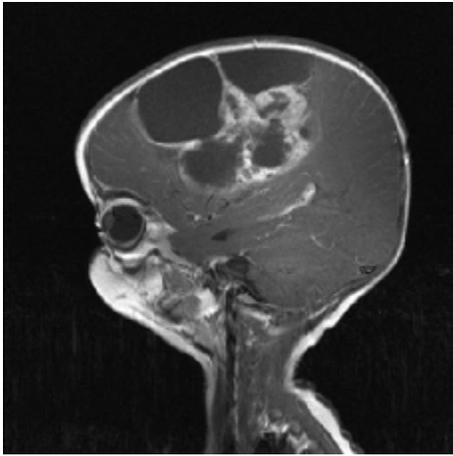


Fig : Sagittal TIWI, showed Non-enhancement of the cystic component in postcontrast study.

Contrast CT study showed large intra-axial complex RT temporoparietal mass lesion that was partially solid but predominantly cystic, surrounded by a small white matter edema containing multiple eccentric cysts causing mass effect on the ipsilateral ventricular system and shift of midline structures about 13.5mm from the midline with nodular dural enhancement, suggestive of dural infiltration Figure(5).

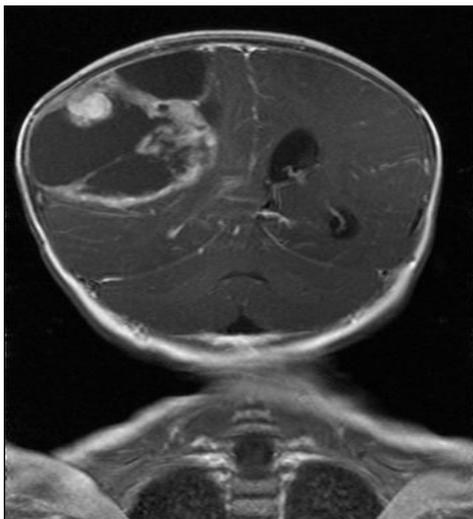


Fig.5: Coronal pos-contrast T1 weighted image shows enhancement of the solid component.

MRI and CT features were not straight forward for teratoid rhabdoid tumors and the case was radiologically reported as primitive

neuroectodermal tumor. Surgery was done for this child and histopathological and immunohistochemical findings, proved the diagnosis of AT/RT.

Discussion:

Because of complexity and heterogeneity and high variability of neuroimaging characteristics, we should be aware of these unusual imaging characteristics that may be seen in children diagnosed to have AT/RT and seek distinguishing imaging features that can aid in preoperative diagnosis, as the radiological features for AT/RT may represent an imaging dilemma. AT/RT and PNET may be confused with each other and make the diagnosis of these tumors difficult⁸.

AT/RT is a highly malignant tumor of the central nervous system (CNS) in childhood, comprising up to 20% of malignant CNS tumors in patients < 3 years old⁹. In our case we had misdiagnosed this child as primitive neuroectodermal tumor, as MRI and CT scan findings of primitive neuroectodermal tumor can be similar to those of AT/RT. The most common site of origin for primitive neuroectodermal tumor is the posterior fossa for medulloblastomas and supratentorial location for ependymomas and germ cell tumors. Children with primitive neuroectodermal tumor usually present at older age compared to those with AT/RT (the median age at diagnosis of AT/RT is less than 2 years). The importance of distinguishing AT/RT from PNET lies in the lack of response of the former to standard therapy for medulloblastoma. Reviewing the literature we found that the radiological appearance of AT/RT usually more variable and heterogeneous. Going with that contrast enhanced CT and MR images of our patient showed patchy pattern of enhancement and striking heterogeneity and complexity of imaging features (cystic and solid component) with calcification and haemorrhage. To achieve the correct diagnosis we should completely be aware of these unusual imaging characteristics. The diagnosis of AT/RT should be included in the differential diagnosis of pediatric brain tumors, especially when the tumor is very heterogeneous and

the child age below 2 years. Tumors of heterogeneous nature should alert the radiologist to differential diagnosis of different brain tumors. Pathology plays a crucial role in diagnosis of teratoid rhabdoid and pathological review included histological and immunochemical analysis, which showed that AT/RT is composed mainly of rhabdoid cells and the presence of these cells, is diagnostic. In immunohistochemically almost all the tumor cells were strongly positive for vimentin. Children presenting with ATRT before the age of 3 years have a dismal prognosis, ATRT presenting in older patients can be cured using a combination of radiation and high-dose alkylating therapy¹⁰.

Conclusion: The striking heterogeneity, high variability and complexity of neuro imaging characteristics of AT/RT make the diagnosis of these tumors difficult, and radiologists should be aware of these unusual radiological features. However, combined efforts of the radiologist, pathologist and the clinician are important and can aid in making correct diagnosis of these tumors

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