

A Rare Case of Atypical Teratoid/ Rhabdoid Tumor of Supratentorial Region in an Adolescent

Hegde S*, Mahajan A, Krithikaa, Ajaikumar BS and Lohith G

HCG hospital, Bangalore, India

*Corresponding author: Hegde S, HCG hospital, Bangalore, India, Tel: +917338303676, E-mail: sweekruti_hegde@yahoo.com

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Abstract

We hereby present you a case of a 15-year-old girl came with the complaints of deviation of angle of the mouth who was evaluated with a supratentorial lesion, and underwent craniotomy for the same. Postsurgery histopathological examination was suggestive of atypical teratoid/rhabdoid tumor. She was planned and treated with external beam radiation (craniospinal irradiation) with tomotherapy to a dose of 36Gy and 18Gy as a boost.

We further discuss about this rare tumor of the brain and how it is common in children below 3 years of age. There are varied methods to diagnose this condition using MRI or PET scan (we have used fibroblast associated PET imaging) but the best method is immunohistochemistry. Many different treatment options are available but nevertheless atypical teratoid/rhabdoid tumor remains a tumor with bad prognosis and most of the patients succumb to this tumor within a year of diagnosis.

Keywords: AT/RT; Spectroscopy; Craniospinal Irradiation; Rhabdoid Cells

Case Summary

A 15-year-old girl came with the complaints of deviation of angle of the mouth to the right side which was intermittent in nature. She had also developed focal twitching of left eye lasting for a few minutes, progressive in nature that increased in frequency and had 3-4 episodes in the day eventually daily in the month of May 2020. She also had weakness and clumsiness of left hand in the month of June 2020. On examination, she was conscious, oriented and with intact memory. She had left upper motor palsy with mild left pronator drift with finger clumsiness.

Her CT (contrast) scan brain showed large slight hyperdense well defined multilobulated mass in right fronto-parietal region with minimal surrounding edema seen as hypodensity. Post contrast study showed inhomogenous bright enhancement. This lesion was reaching upto the dural surface – possibly suggests that the tumor was either glioblastoma or ependymoma.

Her MRI scan suggested a supratentorial lesion, well circumscribed intensely enhancing cortical based lesion of about 5.8 X 4.1 X 3.8 cm seen in the right temporo-parietal region with mass effect on the adjacent insular cortex and adjacent cortical sulci and midline shift of 5mm towards the left. The lesion demonstrates mild perilesional edema and mild to moderate diffuse restriction. There was gyral swelling in the adjacent parietal cortex. On MR spectroscopy, there was increased choline peak and decreased NAA peak with increased choline/creatinine ratio? oligodendroglioma or embryonal tumor.

She underwent right frontal craniotomy (awake) and tumor decompression under general anaesthesia in July. Histopathology examination and Immunohistochemistry reported grey brown soft tissue measuring 3X3X3 cm with spindled, polygonal and rhabdoid cells arranged in sheets over a scant myxoid matrix. Cells showed round to irregular nuclei with pleomorphism and brisk mitotic activity and loss of INI-1, P53 positive, ATRX- retained expression, MIB-1 – 20 to 22%. Final impression of immunohistochemistry was suggestive of Atypical teratoid/rhabdoid tumor- WHO grade 4 of right posterior frontal region.

Post op CT scan done in august showed burr holes in right parietal region with extra axial fluid in the small air pocket right parietal area- post craniotomy site and diffuse dural thickening at post craniotomy site, irregular hypodensities in the right frontal, right temporal and right parietal – post craniotomy changes and effacement of right cerebral sulci with mild midline shift of 4.5mm to the left.

PET CT was done in this adolescent female which helped us to contour brain tumours appropriately which was required to plan radiation treatment.

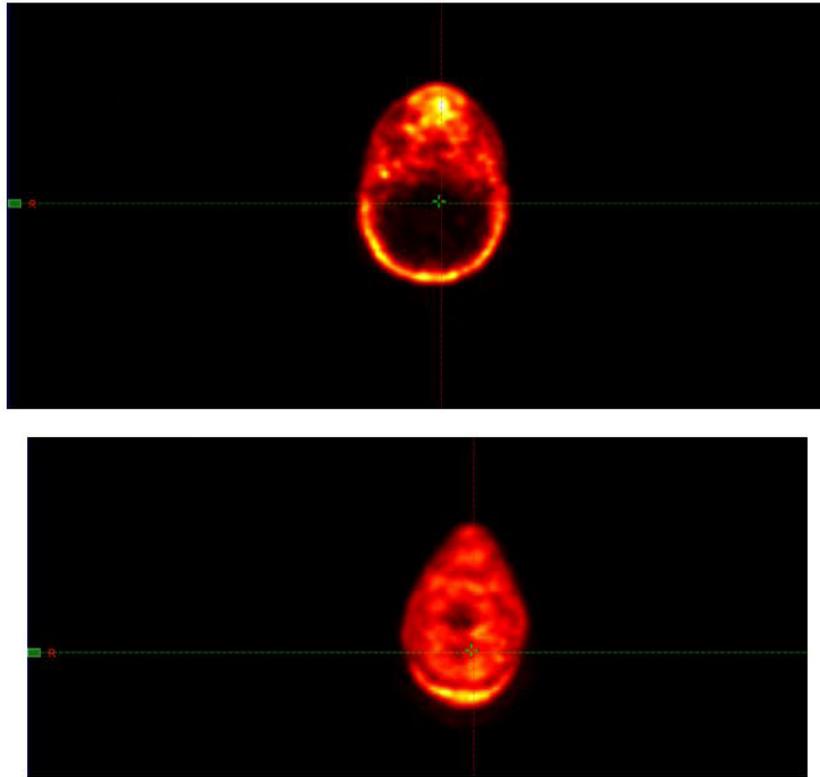


Figure 1: Fibroblast mediated pet scan showing uptake in frontal lobe & posterior fossa

She received radiation treatment with external beam therapy with tomotherapy (craniospinal irradiation) 36Gy to the post op tumor bed and 18Gy to the posterior fossa as the boost.

Discussion

Atypical teratoid/rhabdoid tumors (AT/RTs) are rare, highly malignant brain tumors of the central nervous system. This tumor has been recognised as a distinct entity since 1987 as prior to this they were misdiagnosed as primary neuroectodermal tumor or medulloblastoma due to their similar microscopic appearance.

It occurs almost exclusively in infants and young children especially less than 2 years of age and this rarity of tumor makes it extremely difficult to diagnose in adolescents and adults. prevalence is about 1 to 2% of all the childhood tumors and male predominance is seen. The rapid progression of this tumor provides an average survival of only 6-11 months.

Atypical teratoid rhabdoid tumor most commonly occurs in the posterior fossa most common being cerebellar hemispheres and cerebellopontine angle but can also occur anywhere in the central nervous system. Supratentorial location is relatively less common, though an incidence of 47% has been reported in some case series. Supratentorial locations include the suprasellar, pineal and temporal locations. Other sites where rhabdoid tumor is common are kidneys and few soft tissue sites. The tumor can show CSF dissemination and drop metastases.

Clinical presentation of this tumor is highly nonspecific and depends on the site involved but usually present with raised intracranial pressure. Some common symptoms are lethargy, irritability, macrocephaly and seizures. Loss of balance, trouble with coordination, asymmetric eye and face movements are also present in few cases. Congenital tumors may even present with macrocrania.

Imaging includes CT scan which shows heterogenous enhancement of the tumor, calcification and obstructive hydrocephalus may also be noted. MRI scan shows necrosis, multiple foci of cyst formation and haemorrhage. MR Spectroscopy shows choline elevated and NAA decreased. leptomeningeal seeding has been described in up to 15-30% of cases and so post-contrast imaging of the entire neuroaxis should be considered in suspected AT/RTs. Fibroblast mediated PET CT, a recent evolution in the imaging also shows the uptake of the radiotracer in the brain and spinal cord in case of metastases.

Histopathologically, AT/RTs are classified as WHO grade IV neoplasms. The tumor was composed of rhabdoid cells and undifferentiated small cells mixed with epithelial or mesenchymal components or mimicking PNET/medulloblastoma. Immunohistochemical examination is essential for distinguishing AT/RT from PNET/medulloblastoma. Vimentin, epithelial membrane antigen, cytokeratin, smooth muscle actin, glial fibrillary acid protein, S100, and synaptophysin were positive in varying proportions. The presence of mesenchymal and neuronal marker and the absence of papillary structures are helpful for the diagnosis.

Rhabdoid cells are the hallmark of AT/RT, but only comprise a fraction of the tumor. Other portions of the tumor are indistinguishable on imaging and histology from a medulloblastoma or embryonal tumor with multilayered rosettes. Mutation or deletion of both copies of the *hSNF5/INI1* gene that maps to chromosome band 22q11.2 is observed in approximately 70% of primary tumors. An additional 20 to 25% of tumors have reduced expression at the RNA or protein level, indicative of a loss-of-function event. The INI1 protein is a component of the SWI/SNF chromatin-remodeling complex. Rhabdoid cells are considered as the histological hallmark of AT/RT. They are described as large, pale cells with oval, polygonal or elongated nuclei, and eosinophilic or pale cytoplasm. The nuclei have open chromatin pattern and small to moderately prominent nucleolus .

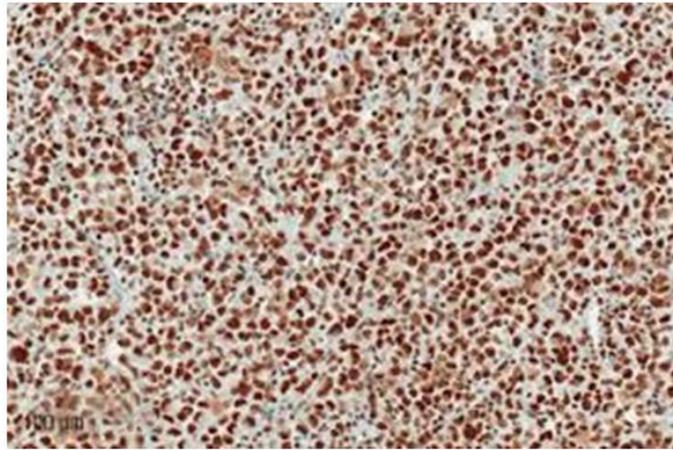


Figure 2: INI1 immunohistochemical stain. Note the strong and uniform nuclear positivity in the rhabdoid tumor cells

Treatment is now focusing on radical surgery followed by aggressive chemotherapy and radiotherapy. High dose of chemotherapy is active against AT/RT and may be associated with a high chance of cure but must be approached with caution due to its potential toxicity. Radiotherapy is classically delayed for young children under 3 years due to its neurotoxicity such as cognitive, motor, visual, and hearing impairment, but some authors recommend immediate postoperative focal irradiation particularly for children without metastatic disease. Athale et al. reported in a meta-analysis that there was a trend towards greater mean survival time (18.4 months vs. 9.5 months, $p=0.097$) with RT . Buscariollo et al. similarly reported survival benefit for ATRT patients given RT ($p=0.02$).

A significant portion of patients die of local recurrence in spite of aggressive surgery and chemotherapy and they succumb usually within a year of diagnosis.

Conclusion

This is a rare case observed in an adolescent girl and has shown good response to both surgery and radiation treatment. Atypical teratoid and rhabdoid tumor remains a rare entity especially in adolescents as most cases are reported within the age of 3 years. There are many ways to diagnose ATRT, involving imaging and recent advances such as fibroblast mediated PET CT but nevertheless immunohistochemistry is the most powerful diagnostic tool in this tumor. Despite the aggressive treatment with surgery, chemotherapy and radiation treatment, patients usually succumb to this tumor within a year of diagnosis.

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