POOR OUTCOME OF ATYPICAL TERATOID RHABDOID TUMOR OF BRAIN IN AN ADULT PATIENT TREATED WITH PAEDIATRIC PROTOCOL

KARTHIK S UDUPA
Department of Medical Oncology,
CANCER INSTITUTE (W I A)

Abstract:
Atypical teratoid rhabdoid tumor (ATRT) of the CNS is an extremely rare and aggressive tumor of early childhood with generally poor outcome even with multimodality treatment 1, 2. Usually it presents in children less than 2 years with posterior cranial fossa being the most common site 2. ATRT is extremely rare in adults with just over 30 cases reported in literature 3. Here we present a case of ATRT in an adult patient who presented with tumor in temporal region. Even with gross total excision of tumor and multimodality treatment he had progressive disease and succumbed to his illness. This case is presented for rarity of disease and to highlight difficulties in implementation of pediatric protocol for treatment of adult patients with ATRT.

Keyword: Atypical teratoid rhabdoid tumor (ATRT), adult ATRT, gross total excision, multimodality treatment

CASE REPORT

26 year old gentleman was evaluated for generalized tonic clonic seizures, underwent MRI brain which revealed $3.2 \times 2.5 \times 2.2$ cm well defined mass lesion with internal hemorrhage in right temporal lesion (Fig 1a). Patient underwent right temporal craniotomy and gross total excision of tumor. Histopathology showed tumor composed of sheets of cells with rhabdoid morphology which were round with abundant pale pink cytoplasm (Fig 1d). These cells diffusely expressed vimentin with variable expression of EMA. Ki 67 labeling was very high (70-80%). Loss of expression of INI-1 was seen in tumor cell nuclei.
overall picture was suggestive of atypical teratoid rhabdoid tumor WHO grade 4. Post excision whole body PET CT scan did not show any uptake (Fig 1b) and CSF cytology did not show any atypical cells. He was started modified IRS-3 protocol which included 54 gy RT to local site(4). During chemotherapy patient had 2 episodes of life threatening febrile neutropenia requiring icu admission . Hence the protocol was modified and actinomycin D was given for 3 days instead of 5 days from week 30 onwards. He also had vincristine(VCR) induced sensory motor neuropathy because of which VCR was omitted from week 33. On week 42 of protocol treatment, he presented with diplopia and examination revealed right 3rd cranial nerve palsy and bilateral papilledema. MRI showed recurrence of lesion in left parieto-occipital region with transmeningeal spread and leptomeningeal enhancement suggestive of relapse (Fig 1c). He had rapid progression of disease and expired after 1 month.

**DISCUSSION**

ATRT is a rare childhood tumor and first described by Biggs et al. in 1987[5]. The name was derived as the gross tumor appearance was similar to a rhabdomyosarcoma; but the cells morphologically and immunohistochemically were different than rhabdomyosarcoma [6]. Atypical teratoid/rhabdoid tumor (AT/RT) shows the mixtures of rhabdoid, epithelial,primitive neuroepithelial, mesenchymal elements [7].ATRT is usually seen in infants and is extremely rare in adults. ATRT usually occurs in posterior fossa, and cerebellum (cerebellopontine angle) being most common site of presentation accounting for upto 52% CNS tumors [8].Monosomy of chromosome 22 or deletion of chromosome 22q11.2 is the most common aberration found in these cases and mutation of the tumor suppressor gene, hSNF5/INI1, is pathognomonic in diagnosis of ATRT [9]. Prognosis of ATRT used to be extremely poor(mean overall survival of 8.5 months)[10].But recently, aggressive pediatric protocol in children with ATRT have resulted in improved survival with 2 year progression free survival and overall survival 53% and 70 % respectively[4]. The extent of surgical resection has a major impact on survival with patients undergoing gross total resection showing significantly better survival [11]. Our patient is an adult who had gross total excision of right temporal tumor and was stated on modified IRS-3 protocol due to the encouraging results in paediatric age group [4]. But our Patient had poor chemotolerance including severe myelosuppression and febrile neutropenia requiring frequent ICU admissions , significant peripheral neuropathy and also severe mucositis which required modification of treatment regimen and significant treatment delay. inspite of gross total excision and incorporation of such an aggressive protocol patient had disseminated CNS relapse and had poor outcome.In summary ATRT in adults is extremely rare and treatment is a major challenge to physicians as aggressive pediatric protocols are generally not well tolerated by adults. An ideal treatment regimen is yet to be described which is less toxic and achieves optimum Cure.

**Reference:**


**Figure legends**

1a. MRI brain which revealed 3.2 x 2.5 x 2.2 cm well defined mass lesion with internal hemorrhage in right temporal lesion.

1b. Post surgery PET CT showing only right temporal hypodensity, with no PET uptake.

1c. Recurrent parenchymal lesion in left parieto-occipital region with transmeningeal spread and leptomeningeal enhancement.

1d. Histopathology showed tumor composed of large round to oval cells with rhabdoid morphology abundant pale pink cytoplasm.