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An Atypical Case of Atypical Teratoid Rhabdoid Tumor (ATRT)

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Introduction

- Atypical Teratoid Rhabdoid tumors are rare pediatric tumors that usually occur at age <3 years[1]
- These tumors are scarcely seen in adults, with the first adult case appearing in 1992[2]
- Since then, about 64 cases have been reported in the literature[3]
- As such, much what has been learned about adult ATRT cases has come from case reports and extrapolation from pediatric cases
- The loss of INI1 (SMARCB1) or BRG1 (SMARCA4) genes are implicated in pathogenesis of ATRT[3]
- Specifically, the INI1/SMARCB1 gene is classified as a tumor-suppressor gene that encodes a core subunit protein of the ATP-dependent SWI/SNF chromatin remodeling complex[4]

Case Presentation

- A 62-year-old Caucasian right-handed female with history of hypertension and sinusitis presented with 2-month history of bilateral, frontal headaches reaching 5/10 in severity with associated nausea, emesis, polydipsia and polyuria.
- Polyuria occurred every hour while polydipsia included drinking (15-20) 16 oz. bottles per day
- She presented to a local hospital and was found to have hypernatremia (Sodium 154 mEq/mL)
- Non-contrast brain Computerized Tomorgraphy (CT) revealed a 1.2cm x 1.1cm x 1.7 cm sellar mass with suprasellar extension
- Urine studies diagnosed central diabetes insipidus, responsive to D-amino D-arginine vasopressin (DDAVP). However, she developed seizures and Abducen's nerve palsy.
- Magnetic resonance imaging (MRI) of the brain demonstrated intraventricular and subarachnoid hemorrhage along with optic nerve edema
- Prior to surgery, she was found on the floor of her room in pool of urine with incoherent speech and a sluggish pupillary reflex on right side.
- STAT non-contrast head CT revealed 2.7 x 1.8 x 2.5 cm extension of hemorrhage into interpeduncular cisterns and ventricles with associated 3rd ventricle & lateral ventricle. There was no midline shift or impending herniation
- She received bi-coronal craniotomy with excision of sellar mass and right frontal external ventricular drain placement
- Hydrocephalus and hemorrhage improved on subsequent MRI a few days later
- The pathology report came back positive for a malignant epithelioid neoplasm, specifically sellar ATRT, WHO grade V. Tumor was shown to be SMARCB1/INI1 deficient and no metastatic lesions were found.
- Recommendations were made for craniospinal radiation and chemotherapy afterward

Histology

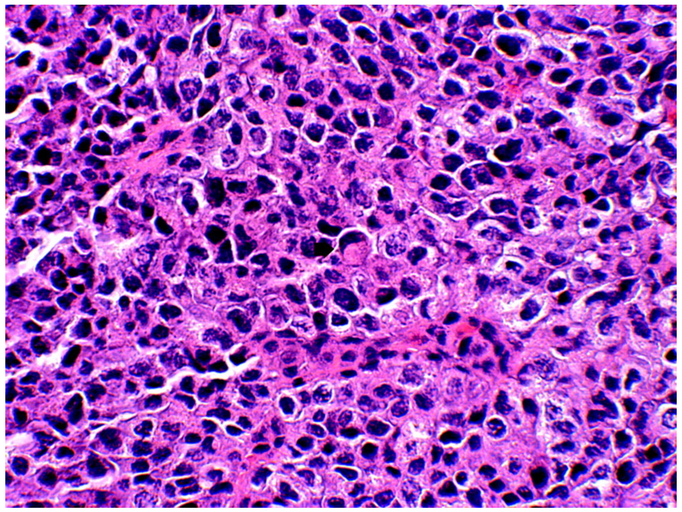
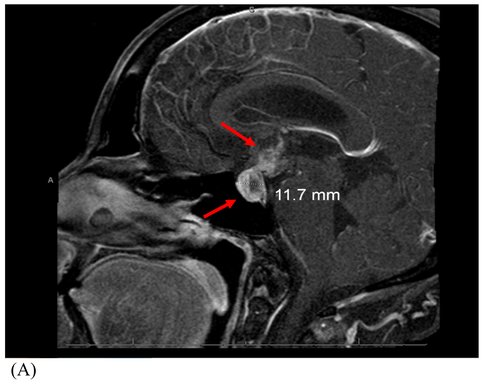
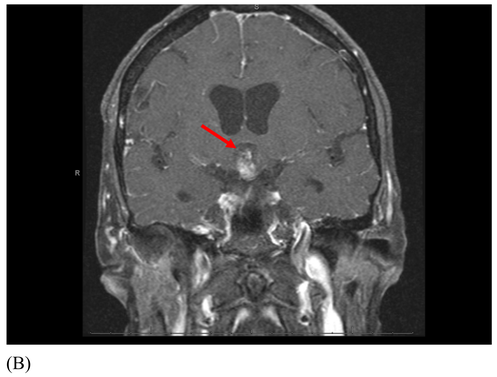


Figure 1: Rare cells with rhabdoid features (arrow), eccentric nuclei, and eosinophilic cytoplasmic hyaline inclusions (hematoxylin-eosin, ×600).[9]

MRI



(A)



(B)

Figure 2: MRI showing hemorrhage of sellar mass (Red arrows) from (A) Sagittal view and (B) Coronal view.

Conclusion

- She was discharged to facility for radiation therapy. During rehab however, she was noted to be less responsive than usual.
- CTH showed worsening hydrocephalus with IVH and she was transferred to HFH
- She underwent Left Frontal VP shunt with EVD placement
- Initial GCS on admission was E2V1TM3
- Her shunt was externalized to facilitate drainage
- There was a deterioration in neurological status and CT head showed new ICH related to tumor
- Goals of care discussion lead to terminal extubation

Discussion

- ATRTs remain rare and aggressive brain tumors seen in both the pediatric and adult population
- The management of ATRT remains a difficult challenge with multimodal approaches to treatment remaining the mainstay
- Resection followed by radiation and chemotherapy has been shown to significantly increase 5-year overall survival rate[5] , yet median time to progression remains in the range of 6-10 months[6].
- Much more standardization is required in the treatment of disease, since patients continue to get variable approaches to treatment
- Additionally, radiation doses and optimal chemotherapy regimens have yet to be determined[7]
- A promising step towards these answers have been in-vitro studies of Insulin-growth factor receptor (IGF-1R) inhibition in sensitizing the tumor to chemotherapy and radiation[8].

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