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Case Reports & Case Series

Case report: The unusual case of a sellar/suprasellar Extrarenal Rhabdoid tumour in an adult

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ABSTRACT

We present the case of a 33 year old lady presenting with progressive cranial nerve palsies caused by an Extrarenal Rhabdoid tumour manifesting as a sellar/suprasellar mass. We share our experience of managing the tumour and review the existing literature.

1. Case presentation

A previously well 33 year old Caucasian female presented with an intractable global headache, diplopia, progressive left III and left VI nerve palsies and reduced visual acuity of 4/6 in the right eye which she stated had developed over the course of one week. Biochemical analysis revealed: Prolactin: 1990(mu/L), mono-prolactin 1416(mu/L), IGF-1:41 (nmol/L), raised Anti B2 glycoprotein 1 Ab IgM at 68.5(MPLU/ml). Cortisol, FSH, LH and GH were normal.

2. Imaging investigations

An urgent CT scan of the Head suggested possible lymphocytic hypophisitis. An MRI and MR venogram (Fig. 1) was arranged and these confirmed a sellar mass with suprasellar and left cavernous sinus extension and adjacent leptomeningeal enhancement.

3. Histopathology and immunohistochemistry

A biopsy was taken via an endoscopic, transphenoidal approach and showed friable tissue fragments measuring 15 × 10 × 5 mm which had been biopsied from the region of the pituitary. These were composed entirely of a solid, cellular tumour replacing the normal anterior gland. The tumour cells demonstrated enlarged nuclei with abundant amounts of clear, vacuolated cytoplasm and occasional abnormal mitotic figures. Necrotic areas were present with a sparse, scattered chronic reactive inflammatory cell infiltrate. There was no granulomatous inflammation and no pronounced desmoplastic stromal response (Fig. 2).

Immunohistochemistry demonstrated negative staining for neuroendocrine markers, markers of pituitary differentiation and melanoma markers in the tumour cells. There was loss of nuclear staining for INI1 (SMARCB1) observed in tumour cells with the positive internal control of positive nuclear staining in the infiltrating lymphoid cells. In addition, tumour cells also demonstrated positive staining for markers to CD99, CD31 and strong vimentin expression and focal membranous positivity for EMA. MIB-1 was markedly elevated and also showed a cell proliferation index of 30%. There were also diffuse cytoplasmic and paranuclear dot-like patterns of cytokeratin staining (AE1/AE3). There was also some focal positive staining with SALL-4 and Glypican-3 but this was considered to be non-specific (Fig. 3).

Abbreviations: INI-1, Integrase interactor 1; SMARCB1, SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily b, member 1; AE1/AE3, Anti-pan cytokeratin antibody; EMA, Epithelial membrane antigen; AFP, Alpha Fetoprotein; PLAP, Placental like alkaline phosphate; SALL-4, Splat like transcription factor; OCT3/4, Octamer-binding transcription factor 4; GH, Growth Hormone; FSH, Follicle stimulating hormone; LH, Luteinizing hormone; ACTH, Adrenocortiocotrophic hormone; TSH, Thyroid stimulating hormone; Gy, Grays (units for Radiotherapy)

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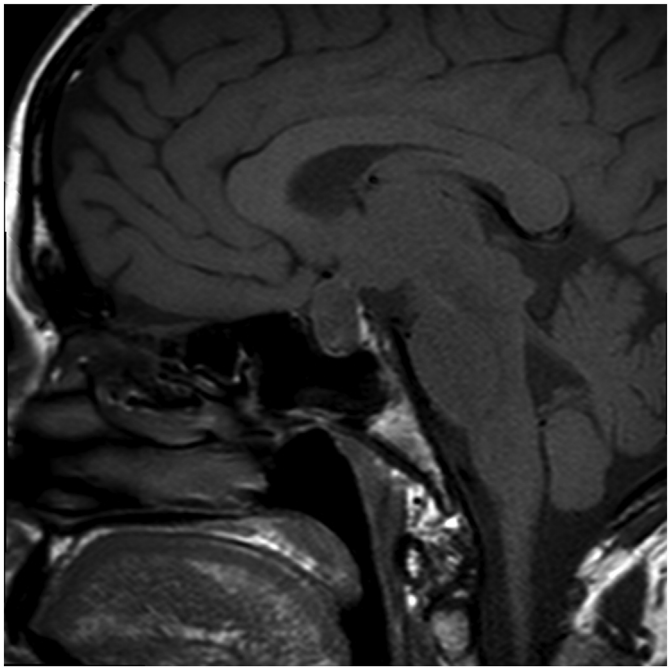

Fig. 1.

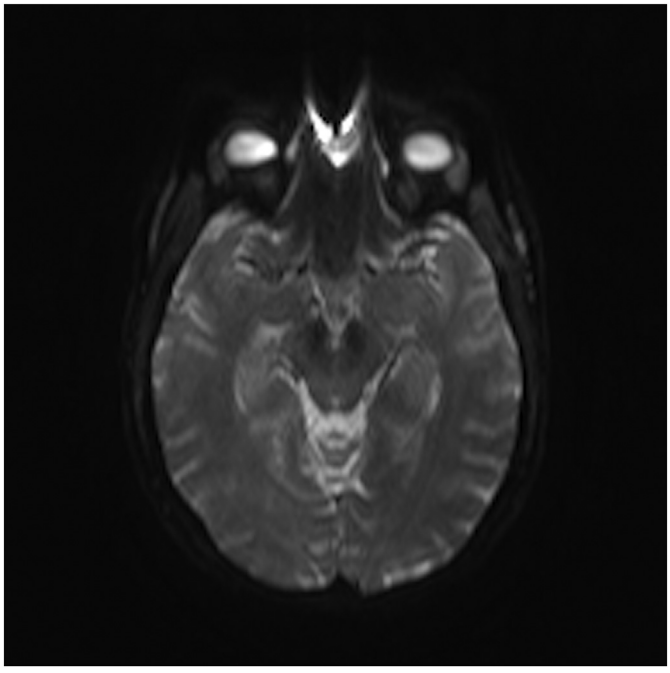

Fig. 2.

There were low numbers of single scattered tumour cells showed Desmin positivity, but additional muscle makers including H-Caldesmon and Myogenin were negative. Germ cell markers show negative staining with OCT 3/4, CD117, CD30, AFP and PLAP and neuroendocrine markers Synaptophysin and CD56 were negative as well. BRAF staining was negative in the tumour cells, but showed some cross-reactivity with entrapped pituicytes. The panel of anterior pituitary hormone markers was negative in tumour cells and with an admixed component of entrapped native pituicytes showing perihormonal expression for GH, FSH, LH, Prolactin, ACTH and TSH. The eventual diagnosis was of an Extrarenal Rhabdoid tumour displaying the characteristic loss of positive nuclear expression of INI-1 (Fig. 4).

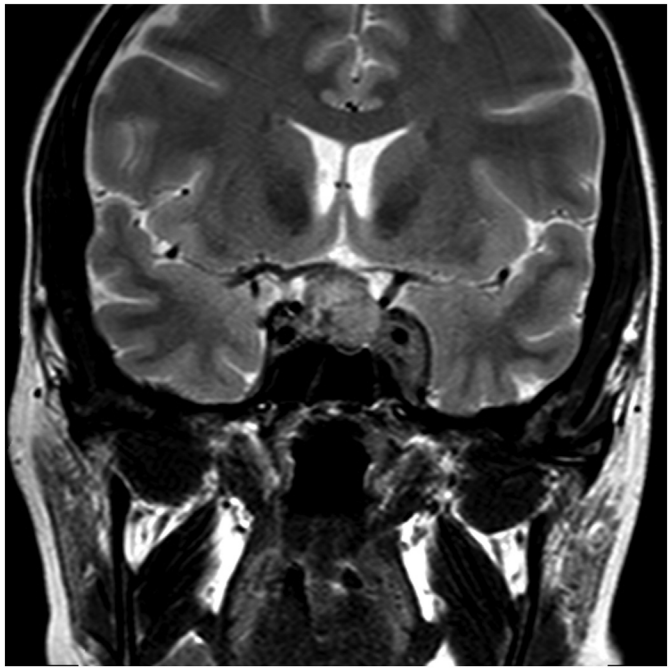

Fig. 3.

4. Management

The patient was initially managed with a trial of Hydrocortisone and Cabergoline but did not show improvement in symptoms. She underwent a biopsy and then a trans-sphenoidal decompression of the lesion. She was well post-operatively with resolution of her symptoms and was discharged home soon after. The patient represented two weeks later with a new right sided VIth nerve palsy. A repeat MRI Head was demonstrated further extension into the left cavernous sinus and new involvement of the right cavernous sinus.

The findings were discussed at the multi-disciplinary team meeting and it was decided that the patient would be best managed with a combination of radiotherapy and chemotherapy. The patient received 36Gy in 20 fractions of Craniospinal irradiation followed by 18Gy in 10 boosts to tumour bed along with concurrent chemotherapy with an aim for alternating ICE–DOX-VAC chemotherapy regimen. ICE: Carboplatin 400 mg/m2 day 1, Etoposide 100 mg/m1 day 1, 2 and 3, Ifosamide 5000 mg/m2 day 1; VAC: Vincristine 1.5 mg/m2 day 1, Actinomycin 0.75 mg/m2 day 1 + 2, Cyclophosphamide 1500 mg/m2 day 1. The Actinomycin was omitted since the patient was on radiotherapy. The patient received one cycle of ICE then one VAC and then one further ICE before chemo was suspended due to liver toxicity and low cell counts due to additional bone marrow toxicity of craniospinal radiotherapy. These normalised following the completion of treatment. The patient responded quite well to this with complete resolution of her symptoms and has now returned to her normal lifestyle.

5. Discussion

Extrarenal Rhabdoid tumours (ERRT), also known as Malignant Rhabdoid Tumours (MRT) are quite rare and predominantly diagnosed in infancy or childhood and rarely seen in adulthood. They are aggressive tumours with a relatively poor prognosis which makes early diagnosis and treatment imperative [1]. Rhabdoid tumours are solid tumours, initially thought to be sarcomatous variants of Wilm's tumours but have over time become established entities of their own accord. These tumours are usually large at presentation and show necrosis and calcification more commonly than the previously thought of as ancestral Wilm's tumours which were first described in 1978 by Palmer

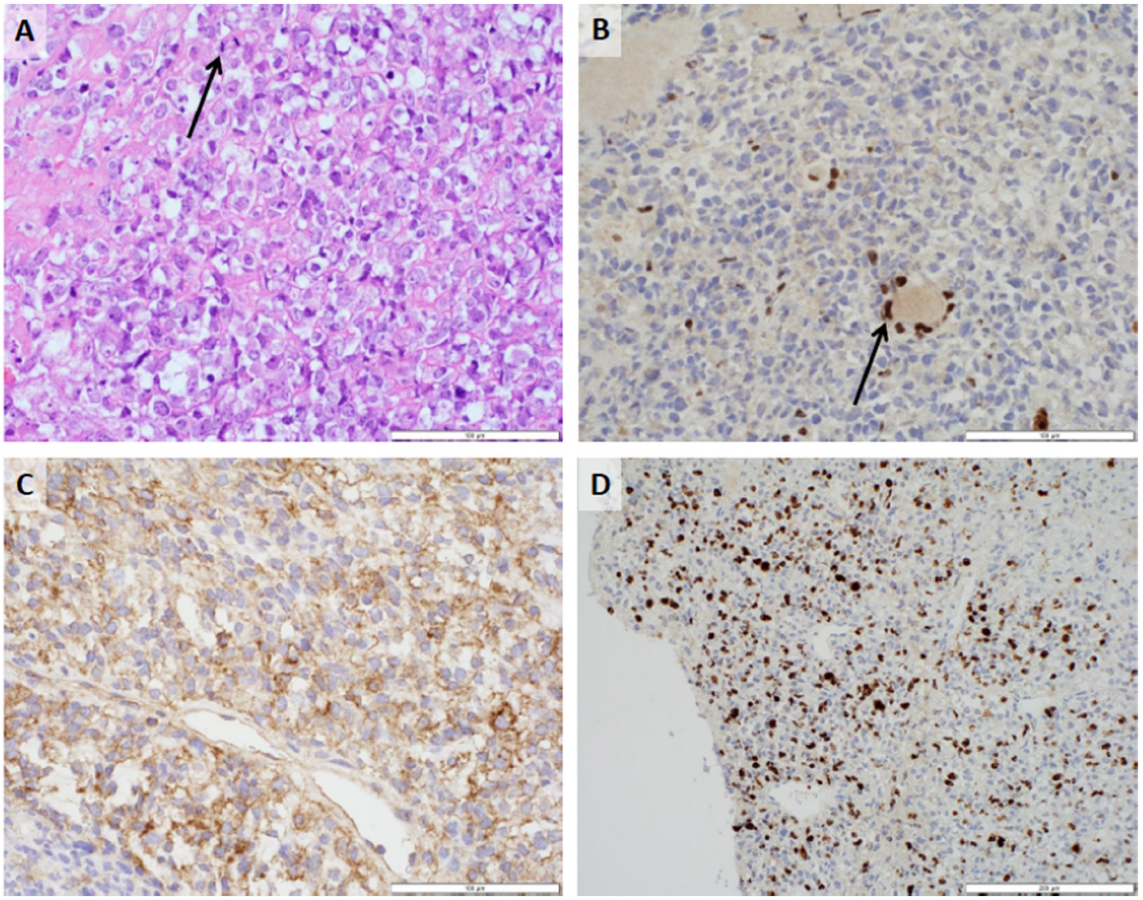

Fig. 4. Histology shows a cellular tumour composed of cells with vacuolated cytoplasm and enlarged, vesicular nuclei with prominent nucleoli. Mitotic activity is present (arrow in A). Loss of tumour cell nuclear expression of INI-1 (SMARCB) is seen, with positive staining of low numbers of infiltrating lymphocytes (arrow in B). Tumour cells display moderately strong cytoplasmic CD99 positivity (c) and MIB-1 demonstrates a high index of cell proliferation in the tumour cells (D). Scale bar represents 100 μM in A, B, C and 200 μM in D.

[1,11]. ERRT's are neoplasms composed of large non cohesive cells with keratin positive para-nuclear inclusions but is histologically identical to renal rhabdoid tumours. INI1 gene inactivation or loss of INI1 antigen expression is observed in epithelioid sarcomas, atypical teratoid rhabdoid tumours in infants and 50% of epithelioid malignant peripheral nerve sheath tumours [2].

Their diagnostic criteria is defined by evidence of sheets of poorly cohesive cells, large polygonal or spindled cells, abundant glassy eosinophilic cytoplasm, paranuclear eosinophilic inclusions and have large eccentric vesicular nucleus which can be round or bean shaped and large, prominent nucleoli and have a high mitotic rate [3,4]. Cytogenetic and molecular analyses have shown abnormalities in the long arm of chromosome 22 and alteration of the hSNF5/INI1 (SMARCB1) gene in renal, extra-renal and intracranial rhabdoid tumours [1,3]. This gene alteration has been thought to be a result of a specific molecular event. It is thought that these may be derived from a primitive pluripotential cell, such as neural crest or equivalent and may be categorized as one of the subsets of primitive neuroectodermal tumour [4].

The choice of imaging for these lesions is Computed Tomography (CT) and Magnetic Resonance Imaging (MRI) and these tumours are defined by their correspondence to Beckwith's restrictive morphological criteria for malignant rhabdoid tumours and tend to present as hypodense, heterogeneous tumours demonstrating contrast enhancement and areas of necrosis on CT and MRI [1] They are graded according to the French Federation of Cancer Center's System grading scheme for adult sarcomas and the TNM classification system. Anecdotal evidence sheds light on the varying distribution of these tumours including with a predilection for pelvic and gastrointestinal distribution in adults [7–9,11].

According to a study by Sultan et al. [10], MRT's of the central nervous system comprise approximately 35% of all ERRT's but this looks at the paediatric population only. The true prevalence and survival information for adult CNS ERRT's still remains somewhat unclear. To date, there are 17 reported cases of ERRT's [5,6]. A literature review was conducted on Pubmed looking at the MeSH terms: Pituitary and/or sellar, rhabdoid tumour and adult/s revealed 131 results in total of which we were able filter these down to 12 articles. We found that of the cases reported, the vast majority was female aged between 21 and 69 and only two of the reported cases were male [12,13]. 2 of the women were pregnant at the time of diagnosis [14,15]. Most of the cases reported in the literature report ERRT's arising in the sellar/pituitary regions primarily with a few exceptions where there was a background of previous CNS tumours or radiotherapy [16–18]. This prompts the questions why these tumours arise in the particular region and why it has a predilection for affecting females – these questions have yet to be explored and have scope for further research. The overall prognosis of this case is poor but survival of upto 6 years [17]. The reason our case is an asset to the existing literature is because despite its dire prognosis of being an aggressive tumour with poor early outcomes and high mortality rate, we were able to manage our patient and treat her symptoms using the treatment mentioned in the Management section thus demonstrating that positive clinical outcomes are possible and thus making this the third longest survival in a patient with an ERRT to date, to the best of our knowledge. Our patient whom we have presented had remained well and asymptomatic 3 years following her initial presentation before representing with extensive abdominal

metastasis. This rare and unusual tumour is best managed as part of a multidisciplinary team with a combination of radiotherapy, chemotherapy and debulking surgical intervention depending on the tumour size and the areas involved. There are ample opportunities to explore the pathogenesis of Extra Renal Rhabdoid tumours.

Conflicts of interest

None to declare.

References

[1] A. Abdullah, Y. Patel, T.J. Lewis, H. Elsamaloty, S. Strobel, Extrarenal malignant rhabdoid tumors: radiologic findings with histopathologic correlation, Cancer Imaging 10 (2010) 97–101 (PMCID: PMC2842174).

[2] K. Mularz, et al., Specificity and sensitivity of INI-1 labeling in epithelioid sarcoma. Loss of INI1 expression as a frequent immunohistochemical event in synovial sarcoma, Pol. J. Pathol. 3 (2012) 179–183.

[3] R. Kempson, R. Rouse, Extrarenal Rhabdoid tumor, [online], 2007. http://surgpathcriteria.stanford.edu/softmisc/extrarenal_rhabdoid_tumor/.

[4] S. Ota, Malignant rhabdoid tumor. A study with two established cell lines, Cancer 71 (9) (1993 May) 2862–2872.

[5] M. Almalki, A. Alrogi, Atypical teratoid/rhabdoid tumour of the sellar region in an adult with long term survival: case report and review of the literature, J. Clin. Med. Res. 9 (3) (2017) 216–220.

[6] European Rhabdoid Registry – International Society of Paediatric Oncology, (2009).

[7] Guidelines for Management of Atypical Teratoid Rhabdoid Tumours Version 2.2, (May 2011).

[8] T. Rezanko, M. Tunakan, Primary rhabdoid tumour of the brain in an adult, Neuropathology 26 (1) (2006 Feb) 57–61.

[9] J.B. Beckwith, N.F. Palmer, Histopathology and prognosis of Wilms tumors: results from the First National Wilms' Tumor Study, Cancer 41 (5) (1978 May) 1937–1948.

[10] I. Sultan, Age, stage, and radiotherapy, but not primary tumor site, affects the outcome of patients with malignant rhabdoid tumors, Pediatr. Blood Cancer 54 (1) (2010 Jan) 35–40.

[11] F. D'Amico, Extraordinary disease-free survival in a rare malignant Extrarenal Rhabdoid tumor: a case report and review of the literature, J. Med. Case Rep. 12 (1) (2018 Feb) 39.

[12] S. Nobosawa, J. Hirato, T. Sugai, Atypical teratoid/rhabdoid tumor (AT/RT) arising from ependymoma: a type of AT/RT secondarily developing from other primary central nervous system tumors, J. Neuropathol. Exp. Neurol. 75 (2) (2016 Feb) 167–174.

[13] W. Wu, W.L. Bi, Adult atypical teratoid/rhabdoid tumours, World Neurosurg. 85 (2016 Jan) 197–204, https://doi.org/10.1016/j.wneu.2015.08.076 (Epub 2015 Sep 4).

[14] M. Erickson, Malignant rhabdoid tumor in a pregnant adult female: literature review of central nervous system rhabdoid tumors, J. Neuro-Oncol. 74 (3) (2005 Sep) 311–319.

[15] G. Mirone, Primary malignant rhabdoid tumour of the brain in adults, J. Clin. Neurosci. 16 (11) (2009 Nov) 1495–1497.

[16] V. Barresi, S. Lionti, Pituitary atypical teratoid rhabdoid tumor in a patient with prolactinoma: a unique description, Neuropathology 38 (3) (2018 Jun) 260–267.

[17] K. Takahashi, Case of atypical teratoid/rhabdoid tumor in an adult, with long survival, Brain Tumor Pathol. 28 (1) (2011 Feb) 71–76.

[18] P. Gorayski, Radiation-induced primary cerebral atypical teratoid/rhabdoid tumour in an adult, J. Clin. Neurosci. 20 (10) (2013 Oct) 1466–1468.