Eur Spine J
DOI 10.1007/s00586-014-3445-1

CASE REPORT

Atypical teratoid rhabdoid tumour of the spine: report of a case and literature review

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Received: 1 December 2013 / Revised: 28 June 2014 / Accepted: 29 June 2014

Abstract Atypical teratoid rhabdoid tumour (ATRT) is a rare and highly aggressive malignant neoplasm of the central nervous system (CNS), which occurs predominantly in children less than 2 years of age. There are less than 50 cases described in adult. We report a case of primary spinal ATRT in a 65-year-old male who presented to us with cauda equina syndrome. To the best of our knowledge, our patient is the (1) second oldest patient to be diagnosed with ATRT and only the third case of adult spinal ATRT report in the literature; (2) first reported case of CNS ATRT occurring in a patient with non-rhabdoid renal cancer; (3) first adult patient of ATRT to present with cauda equina syndrome.

Keywords Atypical teratoid rhabdoid tumour · Intradural extramedullary tumour · INI 1 protein · Leptomeningeal dissemination · Cauda equina syndrome

Case history

A 65-year-old Caucasian male presented to us with 5 weeks history of gradually progressive pain and weakness in the legs and 1-day history of painless urinary retention. Past medical history included right-sided nephrectomy for renal carcinoma, 10 years ago and ankylosing spondylitis. He had seen a rheumatologist 3 weeks ago for his symptoms and was started on steroids as his symptoms were attributed to progression of ankylosing spondylitis. On examination, he had 4/5 power in right lower limb and 3/5 in left lower limb and had perianal numbness. Magnetic resonance imaging (MRI) of whole spine showed a well-defined diffusely enhancing intradural extramedullary mass at T12 vertebral level. The lesion appeared isointense on T1 and hypointense on T2-weighted sequences (Fig. 1). The differential diagnosis was either a benign lesion such as meningioma or a metastasis given the history of renal carcinoma. Computer tomography (CT) chest abdomen pelvis and magnetic resonance imaging/magnetic resonance angiography (MRI/MRA) of renal visceral system did not show any abnormality.

He underwent T11–L1 laminectomy and subtotal resection of intradural extramedullary tumour. Intraoperatively on opening the dura, the tumour was found to be stuck and infiltrating the nerve roots of the left side and mixed in with cauda equina which was matted together. Post-operative period was uneventful. At the time of discharge to the rehabilitation unit, he had power 4/5 in the both the legs except a left foot drop (power 2/5) and was self-catheterising. Post operatively, he received external beam radiotherapy as adjuvant treatment (1.67 grey/fraction; 5 fractions/week; 30 fractions in total). He re-presented to our department 1½ years later with increasing back pain and leg weakness. MRI spine showed a recurrence at T4 (Fig. 2) for which he underwent T4/T5 laminotomy and resection of intradural extramedullary tumour. Post operatively he again received adjuvant radiotherapy. He re-presented again after 5 months with increasing

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Published online: 06 November 2014

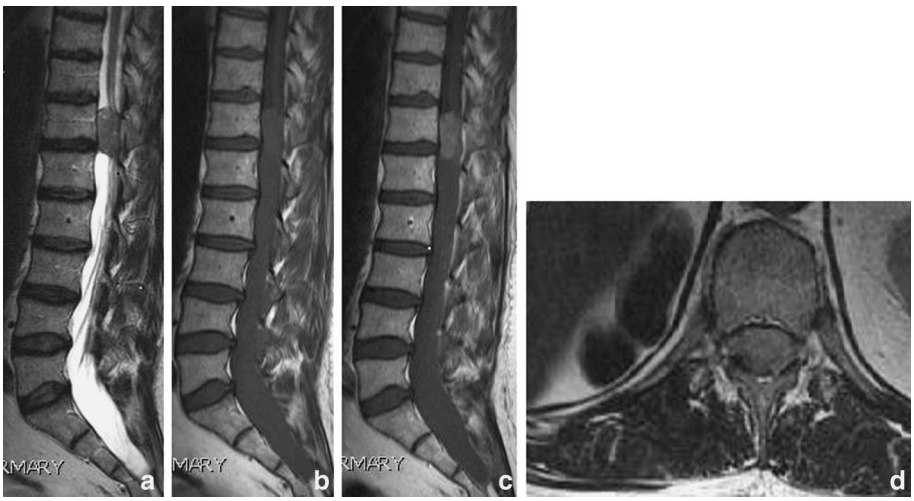

Fig. 1 **a** Sagittal T2-weighted MRI of the spine, **b** sagittal T1-weighted MRI of the spine, **c** sagittal T1-weighted MRI of the spine with gadolinium, **d** axial MRI of the spine

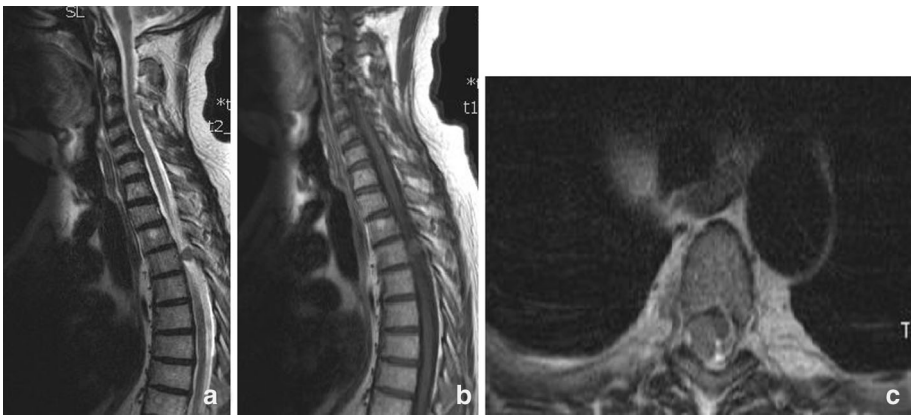

Fig. 2 **a** Sagittal T2-weighted MRI of the spine, **b** sagittal T1-weighted MRI of the spine, **c** axial MRI of the spine

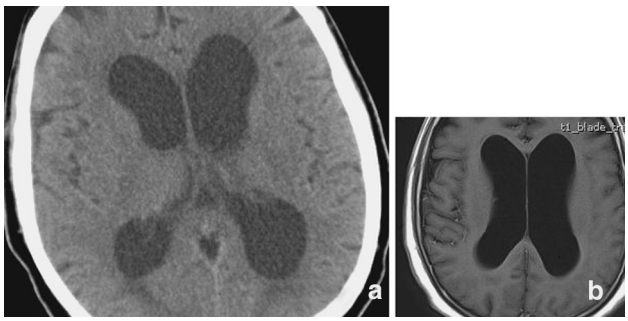

Fig. 3 **a** Axial CT of the head, **b** axial MRI of the brain

confusion and worsening mobility. On examination, his Glasgow Coma Score (GCS) was E3V4M6. He had a CT head which showed enlarged ventricles with periventricular lucency. However, MRI brain did not show any evidence of leptomeningeal dissemination (LMD) of the spinal disease (Fig. 3). He had a ventriculoperitoneal (VP) shunt inserted to manage his hydrocephalus; however, post shunt he continued to be confused. Repeat MRI of his spine showed tumour involvement of his sacrum and pelvis. Cerebrospinal fluid (CSF) obtained from shunt reservoir tap to rule out possible infection showed atypical cells similar in morphology to original spinal tumour, indicating infiltration by malignant tumour cells into the CSF. In view of the above findings and after extensive discussion with the family, the patient was referred for palliative treatment and he finally succumbed to the disease a month later.

Neuropathology

Macroscopic appearance

Fragments of light and dark brown slightly firm tissue were seen.

Microscopic appearance

The tumour consisted of sheets of pleomorphic large cells with rhabdoid and epithelial features. Cells had well-defined cell membrane, abundant eosinophilic and clear cytoplasm and large oval to round vesicular nuclei with

prominent nucleoli. Mitoses were common focally. No necrosis, whorl formation, psammoma bodies or papillary architecture was seen. Under electron microscope, intermediate filaments were seen within the cytoplasm with some vacuoles. These filaments were similar to myofilaments in thickness and arrangement. There was no basement membrane around the cells and no desmosomes. The differential diagnosis were: (1) metastatic carcinoma, especially given the history of renal cell carcinoma; (2) rhabdoid meningioma; (3) ATRT; (4) rhabdomyosarcoma of the meninges; (5) unusual lymphoid tumour. Other less likely differentials were: (1) melanoma; (2) angiosarcoma or proximal epithelioid sarcoma; (3) plasmacytoma; (4) metastatic adrenocortical carcinoma; (5) epithelioid malignant peripheral nerve sheath tumour.

Immunohistocytochemistry

Vimentin was strongly positive in the majority of tumour cells. Epithelial membrane antigen (EMA), cell adhesion molecule (CAM), pancytokeratin (PanCK) and melanoma antigen (Melan-A) were positive in dispersed cells. Staining for renal cell carcinoma (RCC), cytokeratin 7 and 20 (CK7, CK20), human melanoma black (HMB 45), S-100, smooth muscle actin (SMA), CD10 and desmin were negative (Fig. 4).

Cytogenetics report

Fluorescent in situ hybridization (FISH) and other chromosomal studies did not reveal any deletion of either 1p36 or 22q. INI1 protein expression was present in the original tumour from the kidney, but there was loss of INI1 protein expression in the current sample.

The current resected sample was compared to previous renal cell carcinoma. The renal cell carcinoma was a papillary carcinoma of low to intermediate grade, whereas the current tumour showed no evidence of papillary configuration. Also the tumour cells in current sample were very different from those seen in the previous renal carcinoma, in that now, the nuclei were large with prominent nucleoli and large accumulations of eosinophilic material in the cytoplasm whereas there were no rhabdoid cells in the renal carcinoma. The nephrectomy for renal carcinoma was almost 10 years ago and whilst late metastases from renal carcinoma is a well-described phenomenon in low and intermediate grade tumours [1, 2], the metastasis almost invariably looks like the primary morphologically, which was not the case in our patient. Whilst some cells in current sample were positive for the pancytokeratin markers (MNF and CAM), the results for individual keratins were negative. Also the pattern of positivity for MNF and CAM was unusual for de-differentiated epithelial tumour with rhabdoid features as the rhabdoid appearance in those circumstances is the result of disorganisation of the cytoskeleton with accumulation of intermediate filaments, most of which are keratin. The eosinophilic inclusions in this case were also cytokeratin negative. Furthermore, epithelial tumours that are sufficiently de-differentiated to show rhabdoid change are usually highly proliferative, but immunostaining for Ki-67 showed that this was not the case here. There was background non-specific positivity for EMA with only focal membrane-associated specific positivity. Rhabdoid meningioma was ruled out as morphology of cells, and immunostaining results were not in favour of this diagnosis. Desmin was negative so rhabdomyosarcoma was unlikely. CD31 and CD34 were negative, making the diagnosis of angiosarcoma or proximal epithelioid sarcoma unlikely too. Melanoma was considered; however, immunostaining for S-100 and HMB45 were negative. Also absence of any deep extradural soft tissue tumour made the diagnosis of proximal epithelioid sarcoma with INI 1 loss unlikely. Plasmacytoma was considered; however, CD138, CD45 and CD79a were negative. Immunostaining for inhibin was negative, making diagnosis of adrenal carcinoma unlikely. Epithelioid malignant peripheral nerve sheath tumour with INI1 loss was ruled out because of morphology and complete lack of S-100 staining.

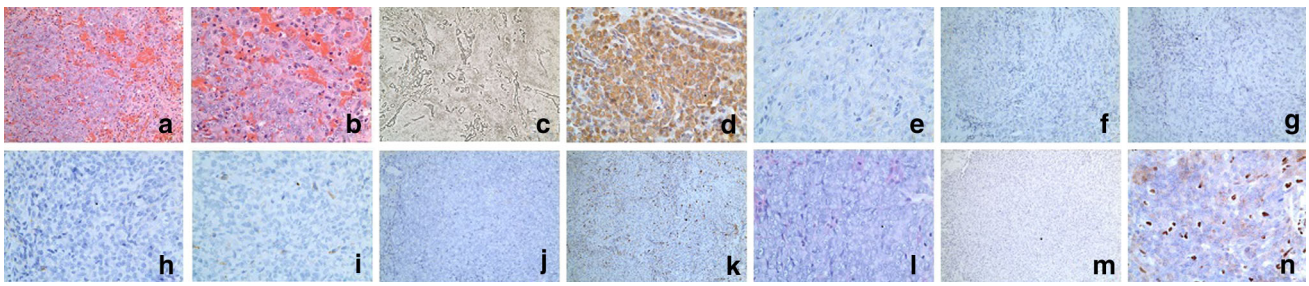

Fig. 4 **a** Hematoxylin and eosin stain ×20, **b** hematoxylin and eosin stain ×40, **c** poor reticulin around tumour cells ×20, **d** vimentin +ve tumour cells ×20, **e** desmin −ve tumour cells ×20, **f** epithelial membrane antigen +ve tumour cells ×20, **g** low Ki-67 ×20, **h** CAM 5.2 −ve tumour cells ×20, **i** synaptophysin −ve tumour cells ×40, **j** CD34 −ve tumour cells ×20, **k** CD31 −ve tumour cells ×20, **l** PAS −ve tumour cells ×40, **m** S100 −ve tumour cells ×20, **n** lack of INI 1 protein expression in tumour cells ×20

Given the histological, Immunohistocytochemistry and loss of INI1 protein expression in current sample, it was diagnosed as primary spinal ATRT despite no evidence of chromosome 22q deletion.

Discussion

ATRT is a rare and highly aggressive malignant neoplasm of the CNS, which occurs predominantly in children less than 2 years of age. It constitutes 2–3 % of all paediatric brain tumours and around 10 % of all CNS tumours in infants [3]. ATRT was first documented by Beckwith and Palmer [4] in 1978, and in 1981 Haas et al. [5] coined the term malignant rhabdoid tumour to describe a variant of Wilms tumour, which resembled rhabdomyosarcoma. Intracranial disease was first described by Lefkowitz et al. [6] in 1987, and finally in 1996 Rorke et al. [7] defined ATRT as a distinct CNS neoplasm. The term "teratoid" was used as the tumour usually containing different cell types as seen in teratoma, and the term "rhabdoid" used as cell morphology was similar to rhabdomyosarcoma. However, there is still debate about the tissue of origin of ATRT. Among the paediatric population, it occurs most commonly in the posterior fossa (61 %), followed by the cerebral hemispheres (20 %), suprasellar and third ventricular region (5 %), pineal region (5 %) and the spinal cord (1 %). It can be multifocal at presentation too (5 %) [8]. Whereas in adults, ATRT is most commonly located in the cerebral hemispheres (53 %), followed by sella (17 %), cerebellum (13 %) and spinal cord (7 %) [9]. It may even involve the cranial vault or show extracranial extension [10]. In patients with ATRT, the main cause of death is either tumour recurrence or leptomeningeal dissemination (LMD) with median survival being 21 months in adults as compared to 6–16.75 months in the paediatric population [7, 11, 12]. Most studies describe the rate of LMD at diagnosis to be around 20–40 % [7, 10, 11, 13–16] though some studies have described much lower rate of 10–15 % [17–19]. ATRTs are polyphenotypic tumours and may be positive for vimentin, EMA, SMA, synaptophysin, CD99, GFAP, S-100 and cytokeratin. They are typically negative for desmin and germ cell markers such as human chorionic gonadotropin and alpha-fetoprotein.

To the best of our knowledge, there are only two other cases of adult spinal ATRT described in the literature. Also, it is a well-recognised phenomenon that intrinsic as well as metastatic spinal tumours can present with cauda equina syndrome [20]; however, this is the first reported case of ATRT in an adult presenting with cauda equina syndrome.

Histologically, ATRT may be composed entirely of rhabdoid cells (13 %) or mixed with primitive neuroepithelial (67 %), mesenchymal (31 %) or epithelial (25 %) tissue [7]. Our patient had rhabdoid cells mixed with mesenchymal and epithelial tissue. The rhabdoid component is characterised by large oval cells with eccentric nuclei, prominent nucleoli and eosinophilic cytoplasm. The primitive neuroepithelial component consists of small poorly differentiated cells with deeply basophilic nuclei, whereas mesenchymal component consists of loosely arranged spindle-shaped cells and epithelial component is characterised by poorly differentiated adenomatous or papillary areas [7, 21].

On CT, ATRT typically appears as hyperdense lesion with heterogeneous enhancement. On T1-weighted MRI it appears as hypo- or isointense, whereas on T2-weighted MRI it may appear as hypo- to hyperintense. It enhances heterogeneously after gadolinium administration [19, 22]. Cyst, calcification, haemorrhage or necrosis may be present in the lesion. MRI has been shown to be more accurate than CSF cytology for early detection of leptomeningeal dissemination in patients with ATRT [15]; however, in our patient the MRI failed to show any leptomeningeal dissemination whereas CSF showed presence of tumour cells. There are isolated care reports describing metastasis of a cranial ATRT to peritoneum via a VP shunt [15]; however, Berger et al. [23] studied 415 patients with primary CNS tumours of whom 152 had shunts and found that shunts does not increase the rate of peritoneal metastasis.

ATRT is the first brain tumour for which a named tumour suppressor gene has been identified. About 70 % of ATRTs are associated with monosomy 22 or deletion/mutation of chromosome band 22q11.2 leading to inactivation of INI1 gene. INI1 is a tumour suppressor gene and member of ATP (adenosine triphosphate)-dependent SW1-SNF (SWItch/Sucrose NonFermentable) chromatin remodelling complex, which regulates cell cycle, growth and differentiation [24–26]. Though monosomy or deletion/mutation of chromosome 22 is seen in only 70 % of the patients, absence of INI 1 protein expression is consistently seen in all cases of ATRT [21, 24]. Our patient too did not have any chromosome 22 anomalies; however, diagnosis of ATRT was confirmed by the absence of INI1 protein expression in the tumour cells along with other Immunohistocytochemistry tests. Although loss of INI1 expression has been reported in other tumours such as epithelioid sarcomas [27] and some renal-collecting duct carcinomas [28], immunohistocytochemistry and demonstration of loss of INI1 protein expression is still considered to be a gold standard for the diagnosis of ATRT [29, 30].

On literature review, we found 43 cases of ATRT described in adults (Table 1). Of these, 22 were male, 20 female and in 1, sex of the patient was not specified. The median age of the patient was 31 years (18–61 years). The

Table 1 Case reports describing the management of atypical teratoid rhabdoid tumour in adults reported in the literature

References	Journal	Age/ sex	Location	IHC	Cytogenetic	Treatment	Survival
Horn et al. (1992)	*Acta Neuropathologica*	21/ M	Lt temporal	Vim, EMA, α1-antichymotrypsin	–	PR + Rx. Recurrence treated with Sx + Cx	72 months
Cossu et al. (1993)	*Virchows Archiv. A, Pathological Anatomy and Histopathology*	18/ M	Lt frontal	Vim, EMA, CK, NSE	–	GTR + CX. Recurrence treated with Sx	18 months
Fisher et al. (1996)	*The Canadian Journal of Neurological Sciences*	32/ M	Lt frontal	Vim, S-100, GFAP	–	Bx + shunt	1 month
Ashraf et al. (1997)	*Medical and Pediatric Oncology*	34/ M	Lt parietal	Vim	–	Bx + Rx. 2 weeks after Rx, had resection of lesion	6 months
D Byram (1999)[a]	*International Journal of Radiation Oncology Biology Physics*	35/ M	Lt temporal	Vim, EMA, CK	–	Sx + Rx. Recurrence treated with Sx	60 months
Sugita et al. (1999)	*Pathology International*	27/ M	Pineal	Vim, EMA, SMA, S-100, chromogranin A, synaptophysin, NSE,	–	PR + Cx + Rx	24 months
Kuge et al. (2000)[b]	*No Shinkei Geka*	32/F	Suprasellar	Vim, EMA, SMA, CK	–	STR + Rx + Cx	24 months
Arrazola et al. (2000)	*Neuroradiology*	20/ M	Lt parietal	Vim, EMA, CK, S-100	–	GTR. Recurrence treated with Sx and Rx	Alive at 24 months
Lutterbach et al. (2001)	*Journal of Neuro-Oncology*	30/F	Cerebellar vermis and both cerebellar hemisphere	Vim, EMA, CK, S-100, GFAP, NFP	–	GTR + Rx. Recurrence treated with radiosurgery and Cx.	11 months
Bruch et al. (2001)	*Human Pathology*	21/F	Spine	–	22q del	–	6 months
		34/F	Parietal	–	22q del	–	6 months
Pimentel et al. (2003)	*Journal of Neuro-Oncology*	31/F	Rt parietal	Vim, NFP, HHF-35, GFAP, S-100, α1-antichymotrypsin, α1 antitrypsin, CD68	–	PR. LMD treated with Cx + Rx	6 months
Kachhara et al. (2003)	*Neurology India*	35/ M	Rt thalamus	Vim	–	PR. During Rx tumour recurred	–
Kawaguchi et al. (2004)	*Acta Neurochirurgica*	22/ M	Cerebellum with LMD to brainstem and whole of spine	Vim, SMA, EMA, NFP, synaptophysin, CK	–	PR + Rx + Cx	Alive at 24 months
Cheng et al. (2005)	*Acta Radiologica*	25/ M	Lt cerebellum	–	–	–	–
Erickson et al. (2005)	*Journal of Neuro-Oncology*	20/F	Rt occipital	Vim, GFAP CAM5.2, EMA, SMA,	No Ch 22 anomaly	GTR + Rx	Alive at the time of publication duration[b]

Table 1 continued

References	Journal	Age/sex	Location	IHC	Cytogenetic	Treatment	Survival
Raisanen et al. [21]	*Brain pathology*	20/F	Sellar	Vim, EMA, CK, loss of INI1 expression	Small del around .22q11.2	Sx + Rx + Cx. Recurrence treated with Cx	Alive at 28 months
		31/F	Sellar with suprasellar extension	Vim, EMA, CK, loss of INI1 expression	Small del around .22q11.2	Sx + Rx	9 months
		45/M	Rt cerebellar	Vim, EMA, CK, loss of INI1 expression	Small del around .22q11.2	Sx + Cx. Cx stopped because of bleeding into tumour bed. Received Rx then	Alive at 15 months
Chen et al. (2006)	*International Journal of Radiation Oncology Biology Physics*	19/M	Post fossa	–	–	GTR + Rx	56.5 months
Rezanko et al. (2006)	*Neuropathology*	27/M	Rt frontal	Vim, EMA, S-100	–	GTR + Rx	4 months
Ingold et al. (2006)	*Acta Neuropathologica*	45/F	Pineal	Vim, SMA, EMA, CK. Loss of INI 1 expression	22q11.2 del	Sx + Rx + Cx + VP shunt. Recurrence treated with Sx	7½ months
Chacko et al. (2007)	*Journal of Neuro-Oncology*	23/M	Rt Frontal	Vim, EMA, SMA, CD34 loss of INI1 expression	22q11.2 del and INI 1 mutation	PR + Rx. 1 month into Rx, he developed progression of disease for which he had GTR and Rx	1.5 months
Zarovnaya et al. [30]	*Journal of Neuro-Oncology*	43/F	C4–C6	EMA, loss of INI1 expresion	Monosomy 22	PR + Rx. Had Sx + Rx + Cx for LMD	30 months
Arita et al. (2008)	*Acta Neurochirurgica*	56/F	Sellar	EMA, NFP, Vim. Loss of INI 1 expression	–	PR + stereotactioc irradiation. LMD treated with Rx	23 months
Makuria et al. (2008)	*Journal of Neuro-Oncology*	23/M	Lt Temporal	Vim, CK, SMA, EMA, MSANF, synaptophysin. Loss of INI1 expression	–	Sx + Rx Cx	Alive at 30 months
		25/F	Rt frontal	Vim, SMA, Synaptophysin, CD34, NFP. Loss of INI1 expression	–	GTR. Recurrences treated with gamma knife and Sx + Rx + Cx	Alive at 17 years
		42/M	Rt fronto parietal	Vim, EMA, CK. Loss of INI1 expression	–	Sx + Rx + Cx	Alive at 18 months
		37/M	Rt fronto parietal	Vim, No Loss of INI1 expression	–	Sx + Rx	–
Samaras et al. [12]	*Clinical Neuropathology*	18/M	Rt fronto temporal	Vim, EMA, SMA, CK, synaptophysin, NFP, GFAP. Loss of INI1 expression	Homozygous del of INI1 gene	GTR + Rx	4 months
Chi et al. [13]	*Journal of Clinical Oncology*	19.5/	Supratentorial	Loss of INI 1 expression	–	Bx + Cx + Rx	2.1 years
Takei et al. (2010)	*Journal of Neurosurgery*	33/F	Pineal	Vim, EMA, CK, GFAP, S-100. Loss of INI1 protein	Del of INI 1 gene	STR + Rx + Cx + VP shunt	Alive at 13 months

Table 1 continued

References	Journal	Age/sex	Location	IHC	Cytogenetic	Treatment	Survival
Umredkar et al. (2010)	*British Journal of Neurosurgery*	32/M	Lt frontal	Vim, EMA, GFAP	–	GTR + Rx + Cx	Alive at 6 months
Heras et al. [29]	*Pathology, Research and Practice*	46/F	Sellar	Vim, CD99, CD34, CK, Synaptophysin, SMA, NSE, Loss of INI 1 expression	–	Sx	
Han et al. (2011)	*American Journal of Neuroradiology*	50/F	Rt temporal	VIM5/5, EMA 4/5, GFAP4/5, SMA3/5, NFP 3/5	–	GTR + Rx	13 months
		25/M	Lt parieto-occipital		–	GTR + Rx	25 months
		24/M	Rt temporo-occipital		–	GTR + Rx	10 months
		35/F	Rt frontal		–	PR + Rx	20 months
		32/M	Rt frontal		–	GTR + RX 4 also had Cx	Alive at 32 months
Schneiderhan et al. [9]	*Neuropathology and Applied Neurobiology*	61/F	Sellar with supra sellar extension	GFAP,S-100, NFP, Synaptophysin, EMA, CK. Loss of INI1 expression	–	Sx. Redo Sx for neurological deterioration	3 months
		57/F	Sellar with parasellar extension	SMA, S100, CD99, P53, EMA, CK (8 and 18), thyroid transcription factor 1. Loss of INI 1 expression	–	GTR. Recurrence treated with Rx + Cx	Alive at 6 months
Takahashi et al. (2011)	*Brain Tumour Pathology*	27/F	Lt parietal	Vim, EMA. Loss of INI 1 expression	–	Sx + Rx. Recurrence treated with Sx + Cx	Alive at 9 years
Shonka et al. (2011)	*Journal of Clinical Medicine Research*	33/F	Pineal	EMA, SMA. Loss of INI 1 expression	–	STR + Cx + SCR +RX + VP shunt. Recurrence treated by changing the Cx	Alive at 18 months

Bx biopsy, *Cx* chemotherapy, *Ch* chromosome, *CK* cytokeratin, *Del* deletion, *EMA* epithelial membrane antigen, *GFAP* glial fibrillary acidic protein, *Lt* left, *LMD* leptomeningeal dissemination, *NFP* neurofilament protein, *NSE* neuron specific enolase, *PR* partial resection, *Rx* radiotherapy, *Rt* right, *SMA* smooth muscle antigen, *SCR* stem cell rescue, *STR* subtotal resection, *Sx* surgery, *VP Shunt* ventriculoperitoneal shunt, *Vim* vimentin

[a] Information from Lutterbach et al. (2001)

[b] Information from Rezanko et al. (2006)

location of the tumour was cerebral lobar in 24, sellar 7, posterior fossa 5, pineal 4 and spinal cord in 2. In one patient, location of the tumour was described as supratentorial only. Treatment information was available for 40 patients of whom 14 had gross total resection (GTR), 3 had subtotal resection (STR), 9 had partial resection (PR), 3 had biopsy and in 11 patients extent of surgery was not specified. 32 patients also had radiotherapy as primary treatment and 19 had chemotherapy as part of primary treatment. 4 patients also underwent VP shunt insertion at the time of primary surgery. Follow-up information was available for 39 patients, of which 25 had succumbed to the disease with median survival of 11 months (1–72 months). 14 patients were still alive with median follow-up of 24 months (6–204 months). From the literature review it is difficult to draw inference about how the extent of resection affects survival as only 42.85 % (6/14) of the patient who were still alive had GTR. Among this cohort of 14 patients who were alive at follow-up, 7 had radiotherapy and chemotherapy, 2 had radiotherapy only, 1 had chemotherapy and 3 did not receive any adjuvant treatment. In one patient, it was not possible to tell if he had chemotherapy in addition to radiotherapy. Also from the literature review, it is difficult to draw conclusions about the management of recurrence and LMD as various treatment modalities including surgery, radiotherapy and chemotherapy have been tried, either alone or in combination.

Similarly on literature review, we found 43 cases of spinal ATRT described in the literature (Table 2). Information about age was available for 35 patients with a median age of 24 months (2–516 months). 21 patients were male, 14 female and in 8, sex of the patient was not mentioned. The tumour was located in cervical spine in 14 patients, thoracic spine in 1, lumbar spine in 1, cervicothoracic spine in 5, thoracolumbar spine in 4, lumbosacral spine in 1 and thoracolumbosacral spine in 4. In 1 patient, there was diffuse involvement of the spine, whereas in 12 patients the location in spine was not specified. Treatment information was available for 28 patients of whom 3 had GTR, 5 STR, 8 PR and 8 had biopsy. Extent of surgery was not specified in four patients. 3 patients also had radiotherapy as primary treatment, 10 had chemotherapy as part of primary treatment, 12 received radiotherapy as well as chemotherapy. Follow-up information was available for 30 patients of which 25 had succumbed to the disease with median survival of 4 months (0.4–42 months). 5 patients were alive at the median follow-up of 34 months (3–60 months). Of these 5 patients, 2 had GTR, 2 had STR whereas in 1 patient the extent of resection was not specified. 4 of these patients also had radiotherapy and chemotherapy, whereas 1 only had radiotherapy (he was offered chemotherapy but family declined it).

Synchronous occurrence of ATRT in the brain and rhabdoid tumour in the kidney is a well-described phenomenon. Similarly delayed occurrence of cranial ATRT, post nephrectomy for malignant rhabdoid tumour has been described in the literature [31]. Also rhabdoid predisposition syndrome has been described in literature, where germline INI1 gene mutation can lead to increased tendency to develop rhabdoid tumour in the CNS and kidney along with supratentorial primitive neuroectodermal tumour (PNET), medulloblastoma and choroid plexus carcinoma [32]. Weeks et al. [33] showed that 13.5 % of patients with renal rhabdoid tumour had concurrent non-rhabdoid CNS tumour. However, our patient did not have INI1 gene mutation and even rhabdoid predisposition syndrome cannot explain occurrence of spinal ATRT, 10 years after nephrectomy for renal papillary carcinoma. Litman et al. [34] had described a case of synchronous malignant rhabdoid tumour occurring in the pelvis and the lung two decades after both sites were irradiated for Wilms' tumour. To the best of our knowledge, this is the first reported case of CNS ATRT in a patient with non-rhabdoid carcinoma of the kidney.

Our case highlights the fact that because of the rarity of this condition (1) it is hardly ever mentioned by the neuroradiologist in their differential diagnosis; (2) most of our experience is based on case reports and case series; (3) there is no standardised treatment and most of the treatment protocols for adults are derived from paediatric experience. Our case report also highlights the fact that because of non-specific clinical presentation and radiological appearance of ATRT, it is necessary to correlate the clinical findings with histopathology, immunohistocytochemistry and cytogenetics to arrive at the correct diagnosis. Though rare, in relevant circumstances, ATRT should be considered in the differential diagnosis of enhancing lesion-causing spinal cord compression. Ankylosing spondylitis is a common cause of back pain and can lead to neurological deterioration; however, at times there can be other cause of sudden deterioration in symptoms of such patients. We also recommend that all patients with ATRT should have imaging of their brain, spine and renal system once diagnosis has been established because of high incidence of LMD at presentation and presence of synchronous renal and CNS ATRT.

Conclusion

We have presented a patient with spinal ATRT in whom diagnosis of the condition was extremely difficult because of (1) rarity of the disease—only two previous cases of adult spinal ATRT has been described; (2) age of our patient—only one other case of ATRT above the

Table 2 Case reports describing the management of spinal atypical teratoid rhabdoid tumour in the literature

Author	Journal	Age/sex	Location	IHC	CYTO	Treatment	Outcome
Rosemberg et al. (1994)	*Clinical Neuropathology*	2 years/F	C6–T1 IDEM	Vim	–	PR + Cx	2 months
Rorke et al. [7]	*Journal of Neurosurgery*	Infant	Cervical IDEM	–	–	Sx	–
Howlett et al. (1997)	*Neuroradiology*	9 months/M	C5 and T5–T10 IM	Vim EMA, CAM5.2, GFAP	–	PR + Cx + Rx	4 months
Tamiya et al. (2000)	*Pediatric Neurosurgery*	7 months/F	T7 below IM	Vim, CK	No chromosome 22 anomaly	PR	12 days
Bruch et al. (2001)	*Human Pathology*	21 years/F	Spine (not specified)	–	22q del	NA	6 months
Bambakidis et al. (2002)	*Pediatric Neurosurgery*	17 years/M	Diffuse spinal	Vim, EMA, SMA	–	Bx + Rx	1 month
		22 months/M	T11–l3 IM	Vim, EMA	–	STR + Cx + ABMT. Recurrence treated with palliative Rx	10 months
Biegel et al. (2002)	*Clinical Cancer Research*	–	Spine (not specified)	–	Ch 22q del	–	–
		–	Spine (not specified)	–	Ch 22q del	–	–
Deshpande et al. (2004)	*Journal of Neuropathology and Experimental Neurology*	7 years/M	L3–S1 IDEM	Vim, EMA, SMA, GFAP, CD99	Ch 22q del	–	–
Chen et al. (2005)	*Neurosurgical Focus*	15 years/M	Spine (not specified) ID	–	–	STR + Cx + Rx	Alive at 34 months
Cheng et al. (2005)	*Acta Radiologica*	2 years/F	T12–S1 ID	–	–	–	2 months
Tekautz et al. [16]	*Journal of Clinical Oncology*	Less than 3 years old	Spine (not specified)		–	Sx + Cx	–
Bannykh et al. (2006)[a]	*Journal of Neuro Oncology*	4 years/M	T9–L1 IDEM	CK, Vim, EMA, SMA. Loss of INI 1 expression	–	Sx + Rx + Cx	Alive at 18 months
Haberler et al. (2006)	*The American Journal of Surgical Pathology*	3 years/M	Cervical	Loss of INI 1 expression	–	Sx + Cx + Rx + HDC (SCR)	2 years
		30 months/F[b]	Cervical	Vim, EMA, CK. Loss of INI 1 expression	Homozygous 22q11.2 del		6 months
		6 months/M[c]	Cervical				0.5 months
		23 months/M[d]	Cervical	Loss of INI 1 expression			
Tanizaki et al. (2006)	*Clinical Neuropathology*	10 months/F	T10 below	Vim, Desmin, SMA, EMA, NSE, CAM5.2	–	Bx + Cx + Rx	3 months

Table 2 continued

Author	Journal	Age/sex	Location	IHC	CYTO	Treatment	Outcome
Kodama et al. (2007)	*Journal of Neuro-Oncology*	9 months/M	C4–T6 IDEM	Vim, SMA, EMA, S100, GFAP, CK	–	STR + Rx + Cx	20 months
Moeller et al. (2007)[a]	*American Journal of Neuroradiology*	9 years/M	T11–L2 IDEM	EMA, NSE. Loss of INI 1 expression	INI 1 gene mutation	GTR + Rx family declined Cx	Disease free at 3 months
Yang et al. (2007)	*Neuropathology*	7 years/M	L2–L4 IDEM	Vim, EMA, CK, CD99, NFP. Loss of INI 1 expression	22q del	Sx + Rx + Cx	7 months
Zarovnaya et al. [30]	*Journal of Neuro-Oncology*	43 years/F	C4–C6 partly IM and partly EM	EMA. Loss of INI1 expression	Monosomy 22	PR + Rx. Recurrence treated with Sx + Rx + Cx	30 months
Seno et al. (2008)	*Brain Tumour Pathology*	5 months/F	C4–T3 ED	EMA, SMA, CK, Vim	–	PR + Cx	2 months
Tinsa et al. (2008)	*Journal of Child Neurology*	4 years/F	Bulbomedullary junction to T1	Vim, GFAP, EMA	–	Decompression + Bx Cx offered but patient succumbed to disease	2 weeks
Warmuth-metz et al. [19]	*Neuroradiology*	6 months/M	Lower T–Spine (not specified)	–	–	–	–
Yano et al. (2008)	*Pediatric Neurosurgery*	1.75 years/F	C2–C6 ID	EMA, CK. Loss of INI 1 expression	–	GTR + Cx + RX + ABMT	Alive at 4 years of age
Fridley et al. (2009)	*Pediatric Neurosurgery*	13 months/F	Lower C–Spine IDEM	Vim. Loss of INI 1 expression	–	PR + Cx	4 months
Niwa et al. (2009)	*Magnetic Resonance in Medical Sciences*	6 years/M	C4–C6 IDEM	Vim, SMA, CK. Loss of INI 1 expression	–	PR	–
Heuer et al. (2010)	*Journal of Neurosurgery Pediatrics*	7 years/M	Clivus to C2 ED	EMA, CK. Loss of INI 1 expression	Homozygous 22q11.2 del	GTR + Cx + Rx. Recurrence treated with Sx	42 months
Mohapatra et al. [22]	*Neuropathology*	4½ years/M	C1–C2 IDEM	Vim, EMA, GFAP, loss of INI 1 expression	–	–	–
Nicolaides et al. (2010)	*Journal of Neuro-Oncology*	8 months/M	C4–C7	Vim, Loss of INI 1 expression	–	Bx + Cx	4 months
Stabouli et al. (2010)	*Hippokratia*	2 months/M	C1–C5 IDEM	Vim, EMA, CK, S-100, CD57	Ch 22 anomaly	Bx + Cx	6 months

Table 2 continued

Author	Journal	Age/sex	Location	IHC	CYTO	Treatment	Outcome
Woehrer et al. (2010)	*Cancer*	30 months/F[e]	Spine (not specified)	Loss of INI 1 expression	–	Bx + Cx + Rx	5.8 months
		3.4 years/M	Spine (not specified) spinal cord	Loss of INI 1 expression	–	Bx + Cx + Rx	22 months
Bruggers et al. (2011)	*Pediatric Blood and Cancer*	6 months/M	C–Spine	–	–	PR + Cx	3 months
		18 months/F	Spine (not specified)	–	–	Bx + Cx	2 months
Dufour et al. [14]	*Cancer*	1 < 2 years old and 3 patients between 2–8.5 years	Spine (not specified)	49/50 patients had loss of INI1 expression in the study	–	–	–
Imagama et al. (2012)	*Journal of Orthopaedic Science*	2 years/F	T12–S1 ID	Vim, SMA, EMA, NSE, GFAP	–	STR + Cx. Recurrence treated with Rx + Cx	9 months
Kelly et al. (2012)	*Journal of Neurosurgery Pediatrics*	4 years/M[f]	T10–L1 IDEM	Vim, EMA, SMA. Loss of INI 1 expression	–	STR + Rx + Cx	Alive at 5 years

ABMT autologous bone marrow transplant, *Bx* biopsy, *Cx* chemotherapy, *Ch* chromosome, *CK* cytokeratin, *Del* deletion, *EMA* epithelial membrane antigen, *ED* extradural, *GFAP, EM* extramedullary, glial fibrillary acidic protein, *HDC* (*SCR*) high dose chemotherapy(stem cell rescue), *ID* intradural, *IDEM* intradural extramedullary, *IM* intramedullary, *NFP* neurofilament protein, *NSE* neuron specific enolase, *PR* partial resection, *Rx* radiotherapy, *SMA* smooth muscle antigen, *SCR* stem cell rescue, *STR* subtotal resection, *Sx* surgery, *Vim* vimentin

[a] Presented with cauda equina syndrome

[b] Initially classified as Ewings sarcoma/ATRT in 1999. Reclassified as ATRT in 2005

[c] Initially classified as anaplastic ependymoma in 1982. Reclassified as ATRT in 2005

[d] Initially classified as sPNET in 1985. Reclassified as ATRT in 2005

[e] Initially diagnosed as Ewings sarcoma in 1999

[f] At the time of report patient was 9 years old

age of 65 years reported in the literature; (3) previous history of non-rhabdoid renal carcinoma—we had to re-examine the previously resected nephrectomy sample and run various immunohistochemistry and cytogenetic tests to make sure that it was not a rhabdoid tumour that had been misdiagnosed as a renal papillary carcinoma. Similarly, the current sample from the spine was sent to other specialist units to rule out an atypical metastasis from the previously resected renal carcinoma; (4) uncommon cytogenetics—our patient did not have monosomy or deletions of chromosome 22.

Acknowledgments We are grateful to Mr Darrach Crimmins, Ms Patricia Harnden and Dr Olaf Ansorge for their help in the management of this challenging case.

Conflict of interest None.

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