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Malignant rhabdoid tumour of the brain

J M Abdullah, Z A A Rahman, A R M Ariff, H Jaafar, K S Phang

ABSTRACT

Rhabdoid tumour is a rare childhood tumour with poor prognosis. We report a 13-month-old Malay girl suffering from this tumour that was located at the left fronto-temporo-parietal region of the brain. Computed tomography showed a large irregular enhancing mass that caused obstructive hydrocephalus. The tumour did not reduce in size after three operations and finally the patient succumbed to the disease four months after diagnosis.

Keywords: cerebral tumour, computed tomography, rhabdoid tumour, surgery

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INTRODUCTION

Rhabdoid tumour was first described in the kidneys and was initially thought to be a sarcomatous variant of Wilms tumour[1]. The exact cell of origin is still not known. Renal rhabdoid tumour has been described as a solid tumor that is seen only in childhood, with most cases occurring in the first year of life. These tumours are usually large at presentation. The existence of extrarenal rhabdoid tumours has been controversial, partly because of the broad spectrum of primary sites, including the extremities, brain, and heart. However, growing numbers of case reports has led to its being widely accepted as a discrete entity[2]. The present case report describes a case of primary malignant rhabdoid tumour of the brain in a 13-month-old girl.

CASE REPORT

A 13-month-old Malay girl was admitted for jerky movements of the right lower leg and loss of consciousness for four days prior to admission. She had a history of macrocephaly since the age of eight months. On examination, she was in decorticate posture with a Glasgow Coma Scale of 8/15. The circumference of the head was greater than the 97th percentile. Physical examination of the patient's abdomen revealed no palpable masses that may indicate enlarged kidneys. Contrast-enhanced computed tomography (CT) showed an irregular enhancing intra-axial mass with central necrosis in the left fronto-temporo-parietal region. It measured 9cm by 8cm. The frontal horn and body of the ipsilateral lateral ventricle were compressed. The mass caused a midline shift to the right and obstructive hydrocephalus (Fig. 1). CT of the thorax and abdomen revealed no other abnormality.

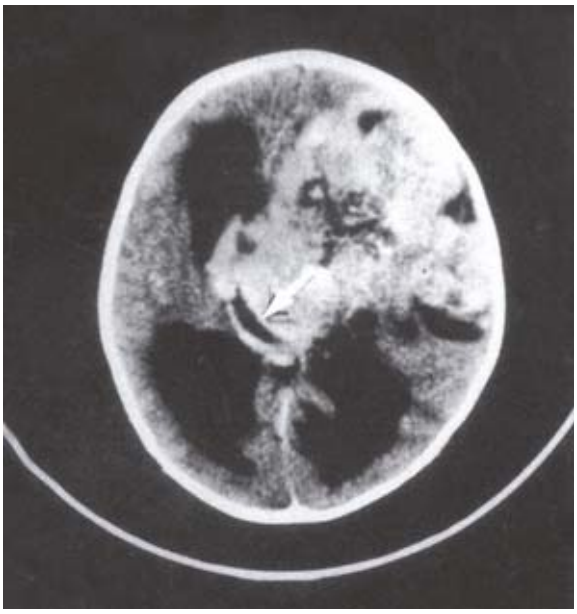

Fig. 1 Enhanced axial CT image shows an irregular enhancing mass in the left fronto-temporo-parietal lobe. Note the compressed ipsilateral lateral ventricle (arrow) and obstructive hydrocephalus.

A semi-urgent craniotomy and tumour debulking was performed. The solid tumour had cystic areas, and measured 10cm by 9cm in size. Post-operative CT taken three months later showed a large residual enhancing mass with marked perifocal oedema and herniation of the brain parenchyma. The brain had herniated through the craniectomy. The size of the tumour had not changed. The patient underwent two other semi-urgent subtotal resections of the tumour in another university hospital, due to increasing neurological deficit and decreased level of consiousness. Unfortunately, she became comatose two weeks after the last surgery and passed away.

Department of Neuroscience
School of Medical Sciences
Universiti Sains Malaysia
16150 Kubang Kerian
Kelantan
Malaysia

J M Abdullah, MD, PhD
Professor and Head

A R M Ariff, MD, MMed
Senior Lecturer

Department of Pathology

H Jaafar, MD, MPath
Associate Professor

Division of Neurosurgery
Faculty of Medicine
Universiti Kebangsaan Malaysia
56000 Cheras
Kuala Lumpur
Malaysia

Z A A Rahman, MD, MS
Associate Professor

Department of Pathology

K S Phang, MBBS, DCP
Senior Lecturer

Correspondence to:
Prof Jafri Malin Abdullah
Tel: (60) 9 7664240
Fax: (60) 9 7648613
Email: unitneurosains ppspusm@yahoo.com

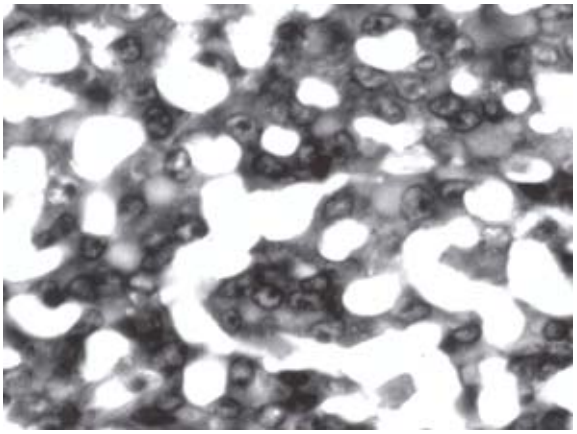

Fig. 2 Photomicrograph of the rhabdoid tumour shows abundant pleomorphic tumour cells with eccentric nuclei, and prominent nucleoli. Characteristic eosinophilic cytoplasmic inclusions are seen in some cells. (Haematoxylin & eosin, X 400).

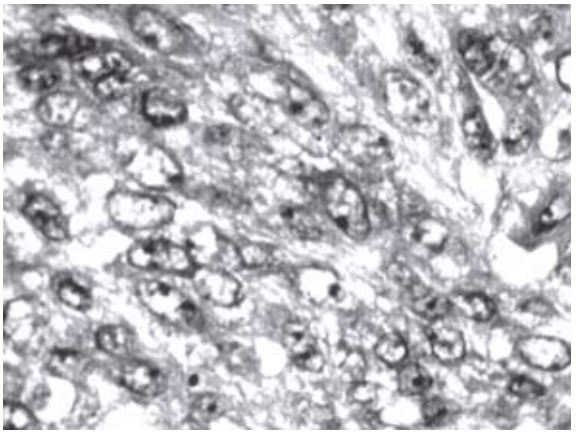

Fig. 3 Photomicrograph shows pleomorphic spindle cells of the mesenchymal component of the malignant rhabdoid tumour. (Haematoxylin & eosin, X 400).

Histopathological examination of the tumour revealed malignant cells infiltrating the brain tissue in loose clusters and singly. In several areas, the tumour cells were floating in a mucopolysaccharide-like substance. The tumour cells expressed large central to eccentric vesicular nuclei, prominent nucleoli, and abundant pink cytoplasm. Prominent paranuclear cytoplasmic inclusions comprising pale bodies with a pinkish margin and eosinophilic bodies were present (Fig. 2). About 25% of the tumour cells exhibited mesenchymal features with pleomorphic plump spindle-shaped nuclei arranged in fascicles (Fig. 3). Cytoplasmic striation was not seen. There were no small cell primitive neuroectodermal tumour-like area or epithelial component seen. Mitosis was brisk and necrosis was extensive. The cells did not stain positive for glycogen or mucin. Immunomarkers for vimentin, S-100 protein and epithelial membrane antigen were strongly positive. The tumour cells also stained positive for actin. Stains for glial fibrillary acid protein, cytokeratin, desmin, myoglobulin, neurofilament and synaptophysin were negative.

DISCUSSION

Malignant rhabdoid tumour (MRT) is a rare tumour that occurs mostly in soft tissue and is highly aggressive. In one large series, the mean survival of patients with central nervous system involvement was 10 months after diagnosis[2]. Numerous papers have reported on both CT and magnetic resonance (MR) imaging appearances of rhabdoid tumours of the central nervous system[3,4]. These papers have described various features such as calcification and bleeding on unenhanced CT, and post-contrast enhancement of the solid component, as well as bony involvement[3,5,6]. In our patient, MR imaging was not done due to the acute emergency admission into both hospitals and parents' refusal for MR imaging. Other papers have also reported recurrent growth of this tumour persisted despite surgery[4,6].

Histologically, MRT of the brain typically comprises of discohesive round to polygonal cells with distinct cell borders. Prominent hyaline inclusions seen in the perinuclear region of the cytoplasm are characteristic of MRT (rhabdoid cells). These features are similar to those of a MRT of the kidney. Apart from rhabdoid cells, it is important to consider other factors in diagnosing MRT. These include its clinical presentation in infancy and in early childhood, histological features of diffuse sheets of cells with vesicular nuclei, sharp nuclear borders, prominent nucleoli and abundant pink cytoplasm, and immunohistochemical profile of tumour cells expressing embryonal, mesenchymal and epithelial cell differentiation.

The WHO classification of tumours of the nervous system had defined rhabdoid tumour as a malignant embryonal central nervous system tumour manifesting in children and composed of rhabdoid cells, with or without fields resembling a classical primitive neuroectodermal tumour (PNET), epithelial tissue and neoplastic mesenchyme[7]. In our patient, two-thirds of the tumour were made up of rhabdoid cells, while the remaining areas were composed of sarcomatous elements. Small cell embryonal and epithelial components were not seen. This is in contrast to the report by Rorke et al[2] that noted the presence of small cell embryonal component and epithelial component in two-thirds and one-quarter of the 52 MRTs studied, respectively. Strong expression of vimentin and epithelial membrane antigen (EMA), typically seen and almost always present in MRT, were evident in our patient. These positivities are not found in PNET; in particular, EMA is always negative in PNET[6]. Electron microscopical study also supports the existence of MRT,

revealing the presence of cytoplasmic whorled or staked arrays of closely packed intermediate filaments in the paranuclear region.

Some authors have described deletion or monosomy of chromosome 22 in 90% of MRTs. The gene involved in MRT, hSNF5/INI1, maps to chromosome band 22q11.2[8,9]. In the brain, the differential diagnoses to be considered include rhabdomyosarcoma (RMS), neuroblastoma, PNET and lymphoma. Histologically, RMS has spindle cells with coarser and more irregular nuclei, and cross-striation is present. Prominent nucleoli are seldom seen when compared to MRT. Electron microscopical studies show definitive skeletal muscle differentiation in RMS, but not in MRT.

Neuroblastoma and PNET have small uniform blue cells with a scanty cytoplasm and a rosette formation. These tumours show neural differentiation. Lymphoma also has small blue cells with a scanty cytoplasm, but test positive with lymphoid markers such as leucocyte common antigen. Treatment in these patients include surgery alone, surgery with chemotherapy, and surgery with both chemotherapy and radiotherapy. All these treatment modalities have not decreased patient mortality and morbidity[1,10].

In conclusion, rhabdoid tumour of the central nervous system is a rare tumour in children. At present, there is no effective treatment for this disease. Our patient presented to two different university hospitals at two different periods with increasing neurological deficit and poor Glasgow Coma Scale. The second neurosurgeon did not know about the original histopathology of the patient as the patient absconded from the first university hospital to seek traditional treatment. The diagnosis of primary MRT of the brain can only be confirmed by histopathological examination. CT and MR imaging findings are non-specific, and do not help in determining a pre-operative diagnosis. The short life span of our patient, despite numerous treatment measures, is a feature that was also noted in other previous case reports[1,2,4,10].

REFERENCES

1. Beckwith JB, Palmer NF. Histopathology and prognosis of Wilms' tumor: results from the First National Wilms' Tumor study. Cancer 1978; 41:1937-48.
2. Rorke LB, Parker RJ, Biegel JA. Central nervous system atypical teratoid/rhabdoid tumours of the infancy and childhood: definition of an entity. J Neurosurg 1996; 85:56-65.
3. Caldemeyer KS, Smith RR, Azzarelli B, Boaz JC. Primary central nervous system malignant rhabdoid tumour: CT and MR appearance stimulates a primitive neuroectodermal tumour. Pediatr Neurosurg 1994; 21:232-6.
4. Chung YN, Wang KC, Shin SH, Kim N, Chi JG, Min KS, et al. Primary intracranial atypical teratoid/rhabdoid tumor in a child: a case report. J Korean Med Sci 2002; 17:723-6.
5. Evans A, Ganatra R, Morris SJ. Imaging features of primary malignant rhabdoid tumour of the brain. Pediatr Radiol 2001; 31:631-3.
6. Yoon CS, Chuang S, Jay V. Primary malignant rhabdoid tumor of the brain : CT and MR findings. Yonsei Med J 2000; 41:8-16.
7. Kleihues P, Louis DN, Scheithauer BW, Rorke LB, Reifenberger G, Burger PC, et al. The WHO classification of tumors of the nervous system. J Neuropathol Exp Neurol 2002; 61:215-25.
8. Biegel JA, Rorke LB, Packer RJ, Emanuel BS. Monosomy 22 in rhabdoid or atypical teratoid tumour of the brain. J Neurosurg 1990; 73:710-4.
9. Biegel JA, Zhou JY, Rorke LB, Stenstrom C, Wainwright LM, Fogelgren B. Germ-line and acquired mutations of INI1 in atypical teratoid and rhabdoid tumours. Cancer Res 1999; 59:74-9.
10. Ronghe MD, Moss TH, Lowis SP. Treatment of CNS malignant rhabdoid tumors. Pediatr Blood Cancer 2004; 42:254-60.