

A newborn with malignant Rhabdoid tumors: A visual diagnosis case

Yu-Cheng Liu¹, Yung-Sen Chang², Wei-Xin Liang³, Kong-Sang Wan^{4,*}

¹⁻⁴Consultant, ^{1,2,4}Dept. of Pediatrics, ³Dept. of Gynecology and Obstetrics, Taipei City Hospital, Renai, Taiwan, China

***Corresponding Author: Kong-Sang Wan**

Email: gwan1998new@hotmail.com

Abstract

Malignant rhabdoid tumor (MRT) is a rare tumor and is first thought a variant of Wilms' tumor in the kidney. It is recognized as presenting at renal, central nervous system (CNS) and other extra-renal primary sites. A full term baby boy with multiple different sizes bluish skin nodules and masses were found on the surface of his body, which was consistent with MRT. Under the emergency skin biopsy, the result showed that the histologic diagnosis of malignant rhabdoid tumor. The presence of metastasis at diagnosis of MRT seems to be the only prognostic factor of outcome.

Keywords: Rhabdoid tumor, Newborn, Cutaneous, Treatment.

Introduction

Malignant rhabdoid tumor (MRT) is now recognized as an entity separate from a Wilms tumor. In contrast, MRT of the kidney is characterized by the early of local and distant metastases and resistance to chemotherapy. It is recognized as presenting at renal, central nervous system (CNS) and other extra-renal primary sites. The incidence of MRT in most countries has not been report. Male to female ratio is about 1.4:1. The mean age at presentation is 10.6 months with a mean age of 15 months. It is uniformly of very poor prognosis and occasionally, MRTs are diagnosed at or immediately after birth.¹

Case Report

A full term baby boy was born by a 32-year-old G1P1 healthy mother via C/S because of transverse position. He was born in 2950 gm with meconium stain. Mild respiratory distress was noted just after delivery but the dyspnea was relieved by intubations suction. The Apgar score was 8 in 1 minute and 10 in 5 minutes. However, multiple different sizes skin nodules were found on the surface of his body. (Fig. 1-2)

Upon physical examination, the baby was generally well. His ears, nose and throat were unremarkable. His chest showed symmetrical expansion, with clear breath sounds bilaterally. Heart rate and rhythm were regular without any audible murmur. Abdomen was soft with no palpable masses, and liver and spleen were not palpable. He moved all for extremities freely and there was no edema. Unfortunately, about 20 skin nodules and thumb-sized masses distributed over his face, body and extremities. Laboratory examination shows that biochemistry including liver function, renal function and electrolytes are within normal limit and matched with age. The complete blood count reveals WBC 20.16K/ μ L, RBC 5.57M/ μ L, Neut. 76.0%, Eos. 2.6%, Baso. 0.2%, Mono. 5.1%, Lym.11.0%. Chest plain film shows infiltration and haziness at right lung, equivocal at left lower lung and suspicious borderline gaseous distention of bowel loops. Thus, the boy is admitted to the hospital under the impression of meconium aspiration syndrome with cutaneous hemangioma for further evaluation and treatment. After the

boy admitted to observation unit, prophylactic antibiotics were given for meconium aspiration. Brain echo, renal echo, and tissue echo were performed, which showed no space-occupying lesion in brain or kidney and only vascular supply lesion was found in bluish skin nodules. Moreover, renal CT was done and no renal mass was seen. Dermatology consultation reported that multiple infantile hemangioma was suspected and 50% may resolve by 5 years old, 70% by 7 year of age. The cardiovascular consultation also suggested to role out hemangioma but skin biopsy was recommended. Also the ophthalmology consultation reported the skin masses over the eyelid are suspected to be tumor or inflammatory abscess. High resolution image study is recommended to examine the extent and nature of the lesion. Under the emergency skin biopsy, the result showed that the histologic diagnosis of malignant rhabdoid tumor depends on identification of characteristic rhabdoid cells large cells with eccentrically located nuclei and abundant, eosinophilic cytoplasm and immunohistochemistry with antibodies to vimentin, keratin and epithelial membrane antigen. (Fig. 3-5) According to the above information, cutaneous malignant rhabdoid tumor was confirmed.



Fig. 1: Multiple different sizes bluish skin nodules on the surface of the newborn body



Fig. 2: Bluish skin nodule on the back of the newborn body

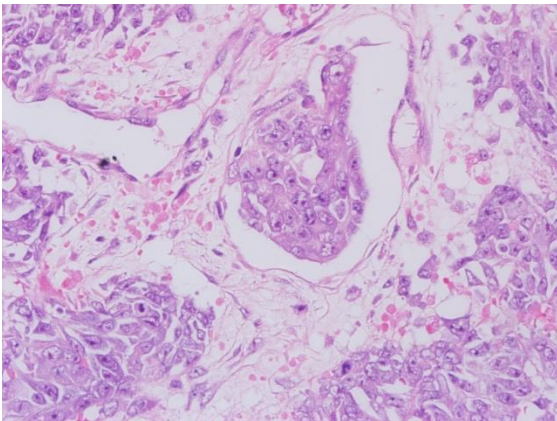


Fig. 3: The tumor cells are focally positive for cytokeratin (AE1/3), and negative for desmin

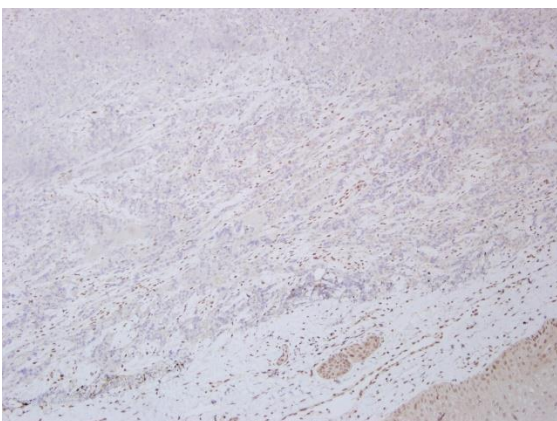


Fig. 4: The tumor cells are showing loss of nuclear stain in INI-1

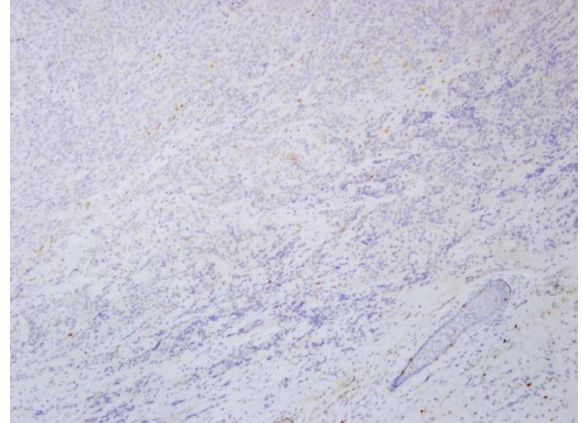


Fig. 5: The tumor cells are negative for S-100 protein

Discussion

MRT is increasingly recognized in young children, probably as a consequence of advances in accurate histological diagnosis rather than a true increase in frequency. Moreover, extrarenal tumors outside the CNS in children less than 12 months of age are now well recognized. Also, MRTs can present as local or disseminated neoplastic disease involving the orbit and should be considered in the differential diagnosis of rapidly progressing orbital lesions presenting in early infancy.²

The histological hallmark is the present of a round-epithelioid cell morphology and a bland immunophenotype, but a distinctive ultrastructure dominated by paranuclear whorls of intermediated filaments, most usually of vimentin.³ Inactivation of SMARCB1 tumor-suppressor gene was originally described as highly specific for rhabdoid tumors.⁴ One-third of newly diagnosed patients with rhabdoid tumor (RT) have an underlying genetic predisposition, therefore, families may demonstrate incomplete penetrance and gonadal mosaicism, which must be considered when counseling families of patients with RT. Also, there is a strong correlation between the loss of INI1 protein immunostaining and presence of an INI1 mutation suggesting that the former is a reliable marker for MRT and atypical teratoid/rhabdoid tumors (AR/RT) in children.⁵ The diagnosis of RT was supported by CT or MRI, but detailed investigation with light microscopy, immunohistochemistry, and electron microscopy enabled us to mark a diagnosis. Moreover, CT findings are not pathognomonic; a renal mass seen on CT in child is unlikely to represent RT of the kidney regardless of its CT features. Therefore, supplemental imaging of the brain should be based on clinical findings or tissue diagnosis.⁶

Malignant rhabdoid tumor is an uncommon tumor that rarely occurs outside of renal and CNS. MRT of the kidney is a childhood tumor that is associated with uniformly aggressive behavior, but it shows a wide spectrum of histological, immunophenotypic, and cytogenetic findings.⁷ The CNS rhabdoid tumors are a highly malignant group of neoplasms usually occurring in children under 2 years of ages with characteristic histopathologic findings but unclear histogenesis and almost uniformly fatal outcome. Cutaneous RT can present as bluish skin nodules producing the

'blueberry muffin baby'-like appearance, sometimes it may misdiagnosed as hemangioma. It share the same chemosensitivity, early recurrence and poor prognosis with lowest survival rate, 4%.⁸

In MRT, no survival differences were observed between those treated with or without radiation, or with or without chemotherapy. Surgical excision had a 74% improvement in survival; especial patient with localized disease.⁹ Older ages was associated with improved survival. The 4-year survival comparing patients less than 2-years-old versus older than 2 at diagnosis was 11% versus 35%. Moreover, the outcome of metastatic RT at diagnosis was very poor with a 5-year overall survival of 11%.¹⁰ In cranial RTs, brain cancer stem cells that suppress the immunity of patients and are resistant to conventional chemotherapy and radiation therapy. It proposed that intensified therapy with autologous bone marrow transplantation and intrathecal chemotherapy may improve the prognosis. According to the above information, the presence of metastasis at diagnosis seems to be the only prognostic factor of outcome.

Funding: The authors received no financial support for the research and /or authorship of this article.

Conflict of Interest: Nil.

References

1. Cobb AR, Sebire NJ, Anderson J. Congenital malignant rhabdoid tumor of the scalp. *J Craniomaxillofac Surg* 2012;40(8):e258-260.
2. Gottlieb C, Nijhawan N, Chorneyko K. Congenital orbital and disseminated extrarenal malignant rhabdoid tumor. *Ophthal Plast Reconstr Surg* 2005;21(1):76-79.
3. Fabre A, Eyden B, Ali HH. Soft-tissue extrarenal rhabdoid tumor with a unique long-term survival. *Ultrastruct Pathol* 2004;28(1):49-52.
4. Rizzo D, Freneaux P, Brisse H. SMARCB1 deficiency in tumors from the peripheral nervous system: a link between schwannomas and rhabdoid tumors? *Am J Surg Pathol* 2012;36(7):964-972.
5. Wu X, Dagar V, Algar E, et al. Rhabdoid tumor: a malignancy of early childhood with variable primary site, histology and clinical behaviour. *Pathol* 2008;40(7):664-670.
6. Agrons GA, Kingsman KD, Wagner BJ. Rhabdoid tumor of the kidney in children: a comparative study of 21 cases. *AJR Am J Roentgenol* 1997;168(2):447-451.
7. Wick MR, Ritter JH, dehner LP. Malignant rhabdoid tumors: a clinicopathologic review and conceptual discussion. *Semin Diagn Pathol* 1995;12(3):233-248.
8. Isaacs H Jr. Cutaneous metastases in neonates: a review. *Pediatr Dermatol* 2011;28(2):85-93.
9. Horazdovsky R, Manivel JC, Chen EY. Surgery and actinomycin improve survival in malignant rhabdoid tumor. *Sarcome* 2013;2013:315170.
10. Reinhard H, Reinert J, Beier R. Rhabdoid tumors in children: prognostic factors in 70 patients diagnosed in Germany. *Oncol Rep* 2008;19(3):819-823.

How to cite this article: Liu Y. C, Chang Y. S, Liang W. X., Wan K. S. A newborn with malignant Rhabdoid tumors: A visual diagnosis case. *Int J Med Paediatr Oncol*. 2018;4(4):167-169.