Cutaneous malignant rhabdoid tumor in the palm of an adult

Masaki Fujioka,[1] Kenji Hayashida,[1] Chikako Murakami,[1] Masanori Hisaoka,[2] Yoshinao Oda,[3] Masahiro Ito[4]

[1]Department of Plastic and Reconstructive Surgery, Clinical Research Center and National Organization, Nagasaki Medical Center, Nagasaki; [2]Department of Pathology and Oncology, School of Medicine, University of Occupational and Environmental Health, Fukuoka; [3]Department of Anatomic Pathology, Graduate School of Medical Sciences, Kyushu University, Fukuoka; [4]Department of Pathology, Clinical Research Center and National Organization, Nagasaki Medical Center, Nagasaki, Japan

Abstract

Malignant rhabdoid tumor is a rare tumor occurring mostly in the neonatal kidneys and central nervous system. Cutaneous malignant rhabdoid tumors are extremely rare in adults. The aim of the study was to report on the clinical, histologic, and immunophenotypic characteristics of this cutaneous malignant rhabdoid tumor which developed in an adult. A 27-year-old male complained of a right palm neoplasm that had been present for 6 months, which was initially diagnosed as an epithelioid sarcoma by biopsy. However, detailed investigation with immunohistochemistry enabled us to make a diagnosis of a rhabdoid tumor. The patient underwent radical abrasion, chemotherapy, and irradiation, and has survived for 1 year without relapse. Only 20 adult cases have been reported thus far in the English literature. We are reporting the 21st case, who remains disease-free at 12 months. Complete resection and local irradiation may increase survival, because there is no standard and reliable curative chemotherapeutic regimen.

Introduction

Malignant rhabdoid tumor is a rare and highly aggressive malignancy of undetermined origin.[1] This tumor occurs most commonly in infants and young children.[2,3] We present a case of malignant rhabdoid tumor developed on the right palm. Our patient is the 21st reported with this tumor developing primarily within the skin in an adult.

Case Report

A 27-year-old male consulted our Medical Center complaining of a right palm neoplasm that had been present for 6 months. On examination, a 4.0×4.5 cm, hard, round, protuberant tumor was found on the ulnar side of the right palm (Figure 1). Magnetic resonance imaging (MRI) showed that the mass adhered to the 5th metacarpal bone and invaded the hypothenar muscle and flexor tendons, but no mass in the head and abdomen. Histological analysis of a biopsy specimen suggested epithelioid sarcoma. Computed tomography revealed right axillary lymph node metastasis.

Surgery consisted of radical excision of the palm tumor along with axillary lymph node resection. *En bloc* resection of the tumor with a skin margin of 2 cm, including the 5th metacarpal bone, 4th intrinsic muscles, 5th flexor tendons, and neuro-vascular bundle, was performed. The 4th metacarpal bone-exposing wound was covered with artificial dermis.

After verification that the surgical margins were free from tumor invasion by histological analysis, secondary reconstruction with the nerve graft and free skin graft was performed 2 weeks later. The patient received 4 chemotherapies with doxorubicin (30 mg) and ifosfamide (5 g), and radiotherapies with a total dose of 60 Gy to the palm and axilla.

After 12 months, the patient showed no tumor relapse, and regained pinch and grip function and showed resolved sensory disturbance of the ring finger.

Pathological findings

Macroscopically, the tumor was located in the subcutis, measuring approximately 4.0×4.5 cm in size. It was grossly recognized by its white-yellow color with poor delimitation between the tumor and surrounding soft tissue. Microscopically, the sections showed the proliferation of round or oval tumor cells with vesicular rounded nuclei, prominent nucleoli, and eosinophilic cytoplasm in a sheet, accompanied by a focal myxoid matrix. Abundant rhabdoid cells with cytoplasmic inclusion bodies were observed (Figure 2). Mitotic figures were occasionally noted. Immunohistochemically, the tumor cells were positive for S-100 protein, GFAP, beta-catenin, and EMA (Figure 3), but negative for cytokeratins (CAM5.2, AE1/AE3, CK 14), alpha-SMA, desmin, HHF35, p63, HMB45, Melan A, MiTF, CD34, HLIB45, glypican 3, and myogenin. The loss of SMARCB1 (SWItch/Sucrose NonFermentable-related, matrix-associated, actin-dependent regulator of chromatin, subfamily b, number 1)/INI1 immunoreactivity was also recognized. The feature was that of an SMARCB1/INI1-deficient tumor, highly suggestive of an extrarenal malignant rhabdoid tumor.

Correspondence: Masaki Fujioka, Department of Plastic and Reconstructive Surgery, National Hospital Organization Nagasaki Medical Center, 1001-1 Kubara 2 Ohmura City, 856-8562 Japan.
Tel. +81.095.7523121 - Fax: +81.095.7540292
E-mail: mfujioka@nmc.hosp.go.jp

Key words: malignant rhabdoid tumor, adult, extrarenal rhabdoid tumor, non-central nervous system rhabdoid tumor, epithelioid sarcoma.

Contributions: MF, guarantor of integrity of the entire study, study concept and design, literature research, clinical studies, manuscript preparation and editing; MI, study concept and design, literature research, manuscript preparation; CM, KH, MH, YO, clinical studies.

Conflict of interests: the authors declare no potential conflict of interests.

Received for publication: 11 January 2013.
Revision received: 13 March 2013.
Accepted for publication: 13 March 2013.

Rare Tumors 2013; 5:e36
doi:10.4081/rt.2013.e36

Discussion

Cutaneous malignant rhabdoid tumor is characterized by the presence of rhabdoid cells, and usually occurs in infants and young children with a highly aggressive biological behavior.[1,4,5]

Kodet *et al.* investigated 26 patients with this tumor, and reported that 11 patients were less than 1 year old, with a median age of 2.9 years.[6]

Thus, a malignant rhabdoid tumor with primary cutaneous involvement in adults is rare, and this is the 21st report.[1]

The most difficult neoplasm to distinguish from a rhabdoid tumor is epithelioid sarcoma.[1] Histologically, it showed a solid arrangement of large, rhabdoid-like cells.[7] Distinction from proximal-type epitheloid sarcoma is difficult, because both of these mesenchymal tumors exhibit epithelial profiles such as immunohistochemical expression of cytokeratin and EMA.[8] They also share ultrastructural features such as specialized junctions, tonofilaments, and microvilli indicative of epithelial differentiation.[9] Our patient was initially diagnosed with an epithelioid sarcoma based on its clinical features and findings on biopsy. However, detailed investigation with immunohisto-

chemistry enabled us to make a diagnosis of a rhabdoid tumor. The most important immunohistochemical markers of the proximal subtype of an epithelioid sarcoma were reported to be vimentin, epithelial membrane antigen, cytokeratin, CD34, and desmin.[7] Epithelioid sarcoma was less of a possibility in our case, because of a lack of reactivity for desmin and CD34. INI (integrase interacting protein) 1 is one member of the BRG (Brahma/SWI2-related gene)-associated factor (BAF) or SWI/SNF (SWItch/Sucrose NonFermentable) complex, which are important in chromatin remodeling.[10] The loss of SMARCB1/INI1 immunoreactivity was also recognized in our specimen. The feature was that of an SMARCB1/INI1-deficient tumor, highly suggestive of an extrarenal malignant rhabdoid tumor.[11]

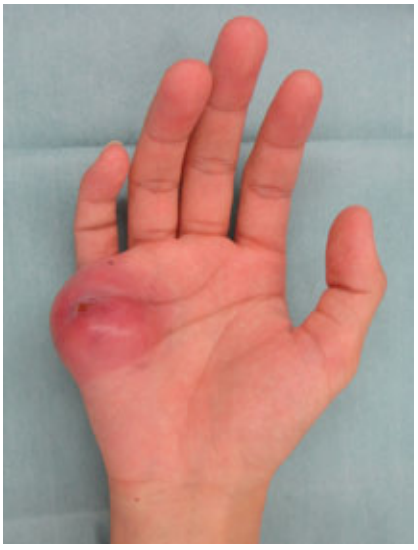

Figure 1. A 4.0×4.5 cm, hard, round, protuberant tumor was found on the right palm.

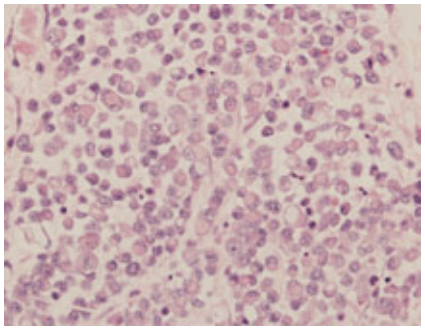

Figure 2. The tumor cells had eccentric nuclei and round and deeply eosinophilic cytoplasms with inclusion bodies, displaying a rhabdoid appearance.

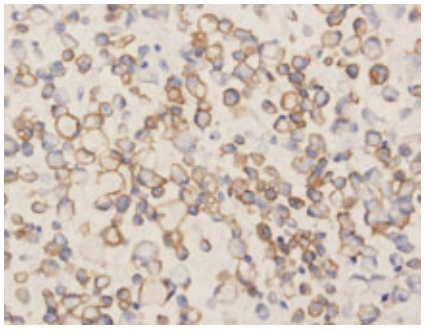

Figure 3. Immunohistochemically the tumor cell were positive for S100 and EMA.

Malignant rhabdiod tumor has a poor prognosis. Review from the German HIT database showed 77% of patients with rhabdoid tumors died of disease.[12] Local recurrences are common and the appearance of distant metastases is almost invariably rapidly fatal. Complete resection increases patient chance to survive.[1,6] There is no standard curative chemotherapeutic regimen. Partial responses to ifosfamide in combination with carboplatin and etoposide in children have been reported; however, complete remission is uncommon.[13] Our patient underwent neo-adjuvant chemotherapy with doxorubicin and ifosfamide, which was recommended for the treatment of high-risk soft-tissue sarcomas in adult patients, and has survived for one year after surgery.[14] The role of adjuvant radiotherapy following complete surgical resection is controversial. Ulutin *et al.* recommends postoperative adjuvant radiotherapy for close surgical margins (margins less than 5 cm) and high-grade tumors.[15] Our patient underwent postoperative adjuvant radiotherapy following these suggestions.[8]

Conclusions

Cutaneous malignant rhabdoid tumors are extremely rare in adults. Our patient is alive more than 1 year after undergoing local radiation and chemotherapy following radical resection of the tumor without recurrence. Complete resection and local irradiation may increase survival, because there is no standard and reliable curative chemotherapeutic regimen.

References

1. Albregts AE, Hebert AA, Aboul-Nasr RA, Raney RB. Malignant rhabdoid tumor presenting as a hemangioma. Pediatr Dermatol 1996;13:468-71.
2. Sangueza OP, Meshul CK, Sangueza P, Mendoza R. Rhabdoid tumor of the skin. Int J Dermatol 1992;31:484-7.
3. Sajedi M, Wolff JE, Egeler RM, et al. Congenital extrarenal non-central nervous system malignant rhabdoid tumor. J Pediatr Hematol Oncol 2002;24:316-20.
4. Morgan MB, Stevens L, Patterson J, Tannenbaum M. Cutaneous epithelioid malignant nerve sheath tumor with rhabdoid features: a histologic, immunohistochemical, and ultrastructural study of three cases. J Cutan Pathol 2000;27:529-34.
5. Shiratsuchi H, Oshiro Y, Saito T, et al. Cytokeratin subunits of inclusion bodies in rhabdoid cells: immunohistochemical and clinicopathological study of malignant rhabdoid tumor and epithelioid sarcoma. Int J Surg Pathol 2001;9:37-48.
6. Kodet R, Newton WA Jr, Sachs N, et al. Rhabdoid tumors of soft tissues: a clinicopathologic study of 26 cases enrolled on the Intergroup Rhabdomyosarcoma Study. Hum Pathol 1991;22:674-84.
7. Rekhi B, Gorad BD, Chinoy RF. Clinicopathological features with outcomes of a series of conventional and proximal-type epithelioid sarcomas, diagnosed over a period of 10 years at a tertiary cancer hospital in India. Virchows Arch 2008;453:141-53.
8. Narendra H, Ray S, Rao L, Geetha V. Malignant extrarenal rhabdoid tumor of the vulva in an adult. J Can Res Ther 2010;6:82-5
9. Argenta PA, Thomas S, Chura JC. Proximal-type epithelioid sarcoma vs. malignant rhabdoid tumor of the vulva: A case report, review of the literature, and an argument for consolidation. Gynecol Oncol 2007;107:130-5.
10. Venneti S, Le P, Martinez D, et al. p16INK4A and p14ARF tumor suppressor pathways are deregulated in malignant rhabdoid tumors. J Neuropathol Exp Neurol 2011;70:596-609.
11. Biegel JA, Perry A, Rhodes CH, et al. Atypical teratoid/rhabdoid tumor of the spine in an adult: case report and review of the literature. J Neurooncol 2007;84:49-55.
12. von Hoff K, Hinkes B, Dannenmann-Stern E, et al. Frequency, risk-factors and survival of children with atypical teratoid rhabdoid tumors (AT/RT) of the CNS diagnosed between 1988 and 2004, and registered to the German HIT database. Pediatr Blood Cancer 2011;57:978-85.
13. Gururangan S, Bowman LC, Parham DM, et al. Primary extracranial rhabdoid tumors. Clinicopathologic features and response to ifosfamide. Cancer 1993; 71:2653-9.
14. Gortzak E, Azzarelli A, Buesa J, et al. A randomised phase II study on neo-adjuvant chemotherapy for high-risk adult soft-tissue sarcoma. Eur J Cancer 2001;37:1096-103.
15. Ulutin HC, Zellars RC, Frassica D. Soft tissue sarcoma of the vulva: a clinical study. Int J Gynecol Cancer 2003;13:528-31.