

Letter to Editor

Rhabdoid melanoma: a case report with review of the literature

Mitsuaki Ishida, Munee Iwai, Keiko Yoshida, Akiko Kagotani, Hidetoshi Okabe

Department of Clinical Laboratory Medicine and Division of Diagnostic Pathology, Shiga University of Medical Science, Shiga, Japan

Received November 21, 2013; Accepted December 17, 2013; Epub January 15, 2014; Published February 1, 2014

It is well known that malignant melanoma sometimes shows a variety of cytomorphological and architectural features, such as balloon cell, small cell, signet-ring cell, myxoid, and adenoid (pseudoglandular) types [1-4]. Rhabdoid melanoma is a relatively rare variant of malignant melanoma, which is histopathologically characterized by the presence of large sheets or nests of polygonal tumor cells with abundant cytoplasm containing eosinophilic inclusions and a peripherally-located nucleus [2]. Herein, we describe an additional case of rhabdoid melanoma and review the clinicopathological features of this type of tumor.

A 57-year-old Japanese female presented with a nodule in her right cheek, which had been detected approximately 6 months earlier. She had undergone tumor resection in the same region more than 10 years earlier at a local outpatient dermatological clinic, and no pathological examination was performed at that time. Physical examination revealed a relatively well-circumscribed reddish nodule, measuring 18 x 15 mm in diameter, in her right cheek. Biopsy was performed, and the pathological diagnosis was malignant melanoma. Subsequently, she underwent total resection of the nodule with 1 cm margins and sentinel lymph node dissection.

Histopathological study of the resected specimen revealed a relatively well-circumscribed elevated nodular lesion involving the dermis and subcutis (the tumor thickness was 9 mm) (**Figure 1A**). The tumor was composed of proliferation of sheets or nests of polygonal neoplas-

tic cells (**Figure 1B**). These neoplastic cells had peripherally-located large round to oval nuclei containing conspicuous nucleoli and rich eosinophilic cytoplasm (**Figure 1C**). The characteristic feature of the tumor cells was the presence of eosinophilic inclusions (**Figure 1C**). No melanin pigments were observed within the cytoplasm of the tumor cells (**Figure 1B, 1C**). Myxoid material was also present around some of the tumor nests (**Figure 1D**). Mitotic figures were occasionally observed (3/10 high-power fields). All tumor cells showed the above-mentioned rhabdoid features, and no conventional malignant melanoma component was detected. Moreover, no intraepidermal atypical melanocytic proliferation was noted (**Figure 1B**). Sentinel lymph node had absence of metastasis.

Immunohistochemical studies were performed using an autostainer (Ventana) by the same method as previously reported [5-8]. Diffuse positive immunoreactivity for S-100 protein in the tumor cells was observed (**Figure 2A**). The characteristic feature was positive immunoreactivity for vimentin in the intracytoplasmic inclusions of the neoplastic cells (**Figure 2B**). Only a few Melan-A-positive tumor cells were noted. However, HMB-45, cytokeratin (AE1/AE3 and CAM5.2), desmin, glial fibrillary acid protein, nestin, peripherin, and alpha-internexin were not expressed in the tumor cells.

Accordingly, an ultimate diagnosis of rhabdoid melanoma was made.

Rhabdoid melanoma was first described by Bittesini *et al.* in 1992 [9]. Since then, this type

Rhabdoid melanoma

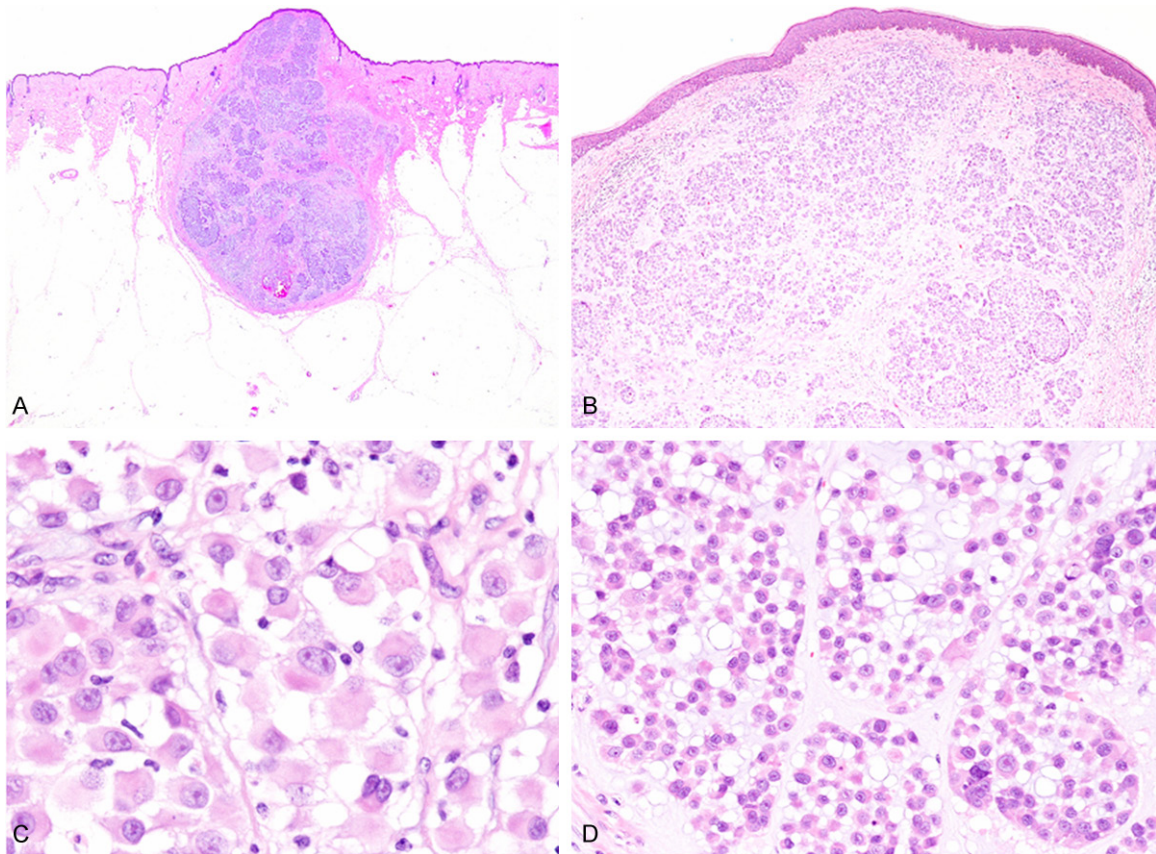


Figure 1. Histopathological features of the cheek tumor. A: Panoramic view showing a relatively well-circumscribed elevated nodular lesion involving the dermis and subcutis. B: Proliferation of nests or sheets of polygonal neoplastic cells. No atypical melanocytic proliferation within the epidermis. HE, x 40. C: The neoplastic cells have eccentrically-located large round to oval nuclei containing conspicuous nucleoli and eosinophilic inclusions within the rich cytoplasm. HE, x 400. D: Myxoid material is present around some of the tumor nests. HE, x 200.

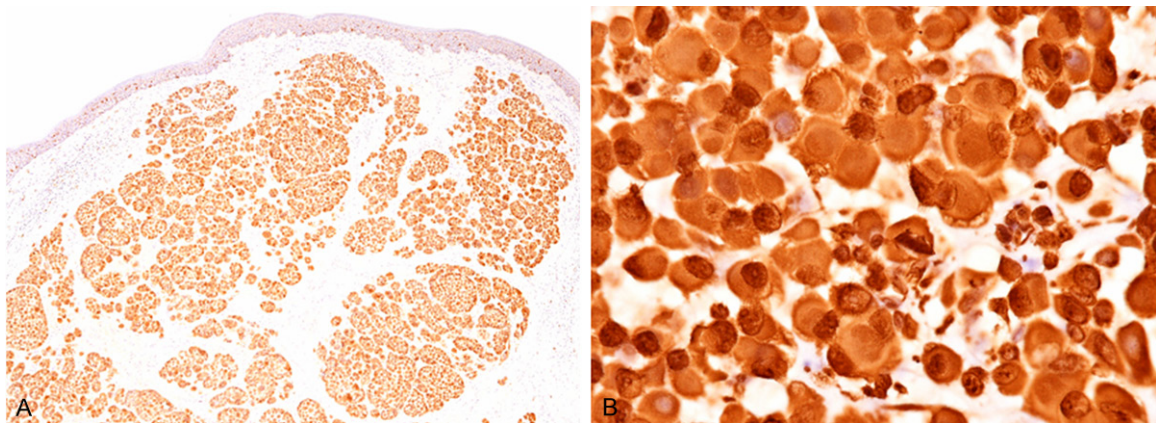


Figure 2. Immunohistochemical findings of the cheek tumor. A: S-100 protein is diffusely expressed, x 40. B: Vimentin is expressed in the intracytoplasmic inclusions, x 400.

of tumor has been recognized as a distinct histopathological subtype of malignant melanoma. Most cases of rhabdoid melanoma appear

as a metastatic or recurrent form, and only a few cases of primary rhabdoid melanoma have been documented [10-16]. Rhabdoid melano-

ma, whether primary or metastatic, appear to have no sex predilection [11, 13]. This type of tumor affects mainly middle-aged to elderly persons [11, 13]. Primary forms can arise in the scalp, trunk, and extremity [10, 11, 16]. Chang *et al.* analyzed the clinicopathological features of thirty-one specimens from 29 patients of metastatic malignant melanoma with rhabdoid features [14]. The most common metastatic site of this type of tumor was lymph nodes (16 cases), followed by soft tissue (10), and liver (2) [14]. Interestingly, primary melanocytic malignancies failed to be documented in three of the 29 cases [14]. The extent of rhabdoid features among neoplastic cells of rhabdoid melanoma varies. Fifteen of 31 specimens were composed nearly exclusively of rhabdoid tumor cells. In the remaining 16 specimens, the neoplastic cells with rhabdoid features constituted less than 25% of the tumor [14]. Moreover, 61% of the specimens were exclusively amelanotic and included 15 tumors with a purely rhabdoid pattern [14]. In the present case, the tumor was a pure rhabdoid melanoma and amelanotic. The patient had a clinical history of skin tumor at the same site more than ten years earlier, although histopathological analysis was not performed at that time. It is difficult to determine whether the present tumor is primary or metastatic, however, it is highly possible that it was a recurrent tumor because rhabdoid melanoma frequently occurs as a recurrent form of malignant melanoma, and no intraepidermal component was present.

Immunohistochemical characteristics of rhabdoid melanoma also show heterogeneity. S-100 protein was diffusely positive in 61% of the specimens, and completely negative in 16% of the largest case series of metastatic rhabdoid melanoma as reported by Chang *et al.* [14]. HMB-45 was diffusely positive in 42% of the specimens, and completely negative in 19% [14]. On the other hand, vimentin was positive in all cases with diffuse or globular staining, which correlated well with the distribution of eosinophilic inclusions, as seen in the present case [14]. The loss of melanocytic markers in rhabdoid melanoma may render the diagnosis difficult, particularly when the tumor is mainly composed of rhabdoid cells and/or with no identified primary site [13]. Positive immunoreactivity for S-100 protein solely leads to the diagnosis of rhabdoid melanoma in some cases

[16]. However, diagnosis of cases showing loss of expression of both S-100 protein and HMB-45 with only presence of rhabdoid features without conventional histopathological features of malignant melanoma is extremely challenging [13, 17]. Laskin *et al.* reported a case of rhabdoid melanoma with loss of expression of S-100 protein and HMB45, in which the clinical information regarding history of malignant melanoma led to an ultimate diagnosis of metastatic rhabdoid melanoma [17].

Ultrastructural analysis revealed that intracytoplasmic inclusions consisted of whorls of intermediate filaments with entrapped rough endoplasmic reticulum and mitochondria [14, 15]. Our immunohistochemical study clearly demonstrated that intracytoplasmic inclusions of the neoplastic cells of rhabdoid melanoma had vimentin, but not other intermediate filaments such as cytokeratin, desmin, glial fibrillary acid protein, nestin, peripherin, and alpha-internexin. Accordingly, intracytoplasmic inclusions, the characteristic feature of rhabdoid melanoma, are composed of accumulation of vimentin.

In conclusion, we describe here an additional case of rhabdoid melanoma. This type of tumor is a relatively rare variant of malignant melanoma and appears mainly as a metastatic or recurrent form. Amelanotic cases and loss of expression of melanocytic markers as shown by immunohistochemical analysis are not uncommon. Therefore, rhabdoid melanoma must be included in the differential diagnostic considerations of rhabdoid tumors, especially metastatic lesions of unknown primary sites.

Disclosure of conflict of interest

None.

Address correspondence to: Dr. Mitsuaki Ishida, Department of Clinical Laboratory Medicine and Division of Diagnostic Pathology, Shiga University of Medical Science, Tsukinowa-cho, Seta, Otsu, Shiga, 520-2192, Japan. Tel: +81-77-548-2603; Fax: +81-77-548-2407; E-mail: mitsuaki@belle.shiga-med.ac.jp

References

- [1] Banerjee SS, Harris M. Morphological and immunophenotypic variations in malignant melanoma. *Histopathology* 2000; 36: 387-402.

- [2] Margo CM, Crowson AN, Mihm MC. Unusual variants of malignant melanoma. *Mod Pathol* 2006; 19: S41-S70.
- [3] Ishida M, Iwai M, Yoshida K, Kagotani A, Okabe H. A distinct histopathological variant of a malignant melanoma with perivascular pseudorosettes: A case report. *Oncol Lett* 2013; 6: 673-675.
- [4] Ishida M, Iwai M, Yoshida K, Kagotani A, Okabe H. Signet-ring cell melanoma with sentinel lymph node metastasis: a case report with the immunohistochemical analyses and review of the clinicopathological features. *Oncol Lett* 2014 Jan; 7: 65-68.
- [5] Ishida M, Iwai M, Yoshida K, Kagotani A, Okabe H. Sarcomatoid carcinoma with small cell carcinoma component of the urinary bladder: A case report with review of the literature. *Int J Clin Exp Pathol* 2013; 6: 1671-1676.
- [6] Ishida M, Mori T, Umeda T, Kawai Y, Kubota Y, Abe H, Iwai M, Yoshida K, Kagotani A, Tani T, Okabe H. Pleomorphic lobular carcinoma in a male breast: case report with review of the literature. *Int J Clin Exp Pathol* 2013; 6: 1441-1444.
- [7] Nakanishi R, Ishida M, Hodohara K, Yoshida T, Yoshii M, Okuno H, Horinouchi A, Iwai M, Yoshida K, Kagotani A, Okabe H. Prominent gelatinous bone marrow transformation presenting prior to myelodysplastic syndrome: a case report with review of the literature. *Int J Clin Exp Pathol* 2013; 6: 1677-1682.
- [8] Ishida M, Mochizuki Y, Iwai M, Yoshida K, Kagotani A, Okabe H. Pigmented squamous intraepithelial neoplasia of the esophagus. *Int J Clin Exp Pathol* 2013; 6: 1868-1873.
- [9] Bittesini L, Dei Tos AP, Fletcher CD. Metastatic malignant melanoma showing a rhabdoid phenotype: further evidence of a non-specific histological pattern. *Histopathology* 1992; 20: 167-170.
- [10] Chung BY, Ahn IS, Cho SI, Kim HO, Kim KH, Park CW, Lee CH. Primary malignant rhabdoid melanoma. *Ann Dermatol* 2011; 23: S155-S159.
- [11] Tallon B, Bhawan J. Primary rhabdoid melanoma with clonal recurrence. *Am J Dermatopathol* 2009; 31: 200-204.
- [12] Nakamura T, Matsumine A, Kato H, Kusuzaki K, Nishimura K, Murata T, Shiraishi T, Oda Y, Tsuneyoshi M, Uchida A. Malignant melanoma with a rhabdoid phenotype exhibiting numerous solid tumor masses: A case report. *Oncol Rep* 2009; 21: 887-891.
- [13] Gavino AC, Gillies EM. Metastatic rhabdoid melanoma: report of a case with a comparative review of the literature. *J Cutan Pathol* 2008; 35: 337-342.
- [14] Chang ES, Wick MR, Swanson PE, Dehner LP. Metastatic malignant melanoma with "rhabdoid" features. *Am J Clin Pathol* 1994; 102: 426-431.
- [15] Abbott JJ, Amirkhan RH, Hoang MP. Malignant melanoma with a rhabdoid phenotype: histologic, immunohistochemical, and ultrastructural study of a case and review of the literature. *Arch Pathol Lab Med* 2004; 128: 686-688.
- [16] Borek BT, McKee PH, Freeman JA, Maguire B, Brander WL, Calonje E. Primary malignant melanoma with rhabdoid features: a histologic and immunocytochemical study of three cases. *Am J Dermatopathol* 1998; 20: 123-127.
- [17] Laskin WB, Knittel DR, Frame JN. S100 protein and HMB-45 negative "rhabdoid" malignant melanoma: a totally dedifferentiated malignant melanoma? *Am J Clin Pathol* 1995; 103: 772-773.