

Nasal spindle cell tumor with rhabdomyoblastic features: A rare and diagnostically difficult case

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Abstract

Nasal spindle cell rhabdomyosarcoma is very rare. The tumor is sometimes confused with other spindle cell tumors. We herein report a case of nasal spindle cell tumor in a 62-year-old woman. The patient first presented herself to a medical doctor's office after an episode of left epistaxis. An intranasal tumor was found and resected. The tumor was composed of spindle cells, and she was diagnosed with desmoid-type fibromatosis. Five years after the initial episode, an intranasal tumor was found again. The tumor showed a fascicular growth pattern with high cellularity and was predominantly composed of spindle cells. Scattered eosinophilic rhabdomyoblasts were also observed. She was diagnosed with spindle cell rhabdomyosarcoma. This is a unique case report not only because nasal spindle cell rhabdomyosarcoma is very rare but also because the tumor was initially diagnosed as desmoid-type fibromatosis. It is important to consider spindle cell rhabdomyosarcoma as a differential diagnosis of nasal spindle cell tumors.

Keywords

Spindle cell rhabdomyosarcoma, rhabdomyosarcoma, spindle cell tumor, desmoid-type fibromatosis, nasal

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Background

Rhabdomyosarcoma is a common soft tissue sarcoma in adolescents and young adults but rarer in patients aged above 40 years.¹ Spindle cell rhabdomyosarcoma is a minor subtype of rhabdomyosarcoma, accounting for approximately 5%–10% of all rhabdomyosarcoma.¹ It affects both children and adults and occurs more frequently in males than in females.² The tumor commonly arises in the paratesticular region in pediatric patients or in head and neck deep soft tissue in adults; however, nasal spindle cell rhabdomyosarcoma is very rare.^{2–4} The tumor predominantly consists of spindle cells. Scattered rhabdomyoblasts may be observed; however, when rhabdomyoblastic features are not clear, the tumor can be mistaken for other benign or malignant tumors such as fibromatosis, leiomyosarcoma, and fibrosarcoma.^{1,5–7} Although the spindle cell variant generally has a better prognosis than other subtypes of rhabdomyosarcoma,¹ the outcome in adults is worse than in pediatric patients, with a recurrence and metastasis rate of 40%–50%,² and MYOD1-mutant spindle cell and sclerosing rhabdomyosarcoma show a poor prognosis, irrespective of age.⁸

We herein report a case of recurrent nasal spindle cell rhabdomyosarcoma that was initially diagnosed with desmoid-type fibromatosis.

Case presentation

A 62-year-old female first presented herself to a medical doctor's office 5 years previously after an episode of left

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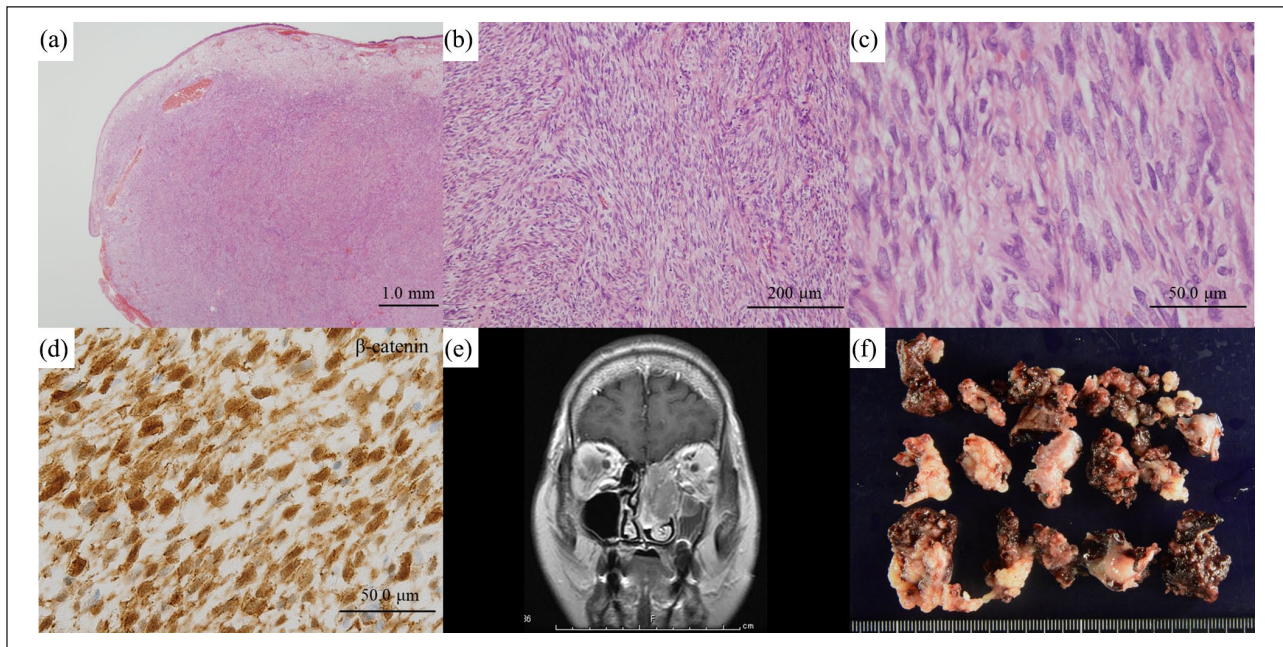


Figure 1. Microscopic findings of the tumor resected 5 years previously, and an MRI scan and macroscopic findings of the recurrent tumor. (a) A low-power view of the tumor revealed a poorly circumscribed lesion. Bar = 1.0 mm (H&E staining; original magnification: $\times 12.5$). (b) The tumor showed a fascicular pattern with moderate cellularity. Bar = 200 μm (H&E staining; original magnification: $\times 100$). (c) The tumor was composed of spindle cells with mild to moderate nuclear atypia. Bar = 50.0 μm (H&E staining; original magnification: $\times 400$). (d) The tumor cells were considered to be reactive for nuclear β -catenin. Bar = 50.0 μm (original magnification: $\times 400$). (e) Enhanced T1-weighted MRI showed a 43 mm tumor in the left nasal cavity. The orbit adjacent to the tumor was slightly unclear. (f) The tumor tissue resected from the nasal cavity, weighed 16 g (in total) and had a tan color.

epistaxis. An intranasal tumor was found and resected. A microscopic study of the tumor revealed a poorly circumscribed lesion (Figure 1(a)). The tumor showed a fascicular fashion with moderate cellularity (Figure 1(b)). The tumor was composed of spindle cells with mild to moderate nuclear atypia (Figure 1(c)). Immunohistochemically, the tumor cells were positive for vimentin, focally positive for S-100 protein, and negative for cytokeratin AE1/AE3, α -smooth muscle actin (α -SMA), and CD34. They were considered to be reactive for nuclear β -catenin (Figure 1(d)) and negative for desmin. The Ki-67 labeling index was less than 5%. Based on these findings, she was diagnosed with desmoid-type fibromatosis.

There were subsequently no symptoms for 5 years after the initial episode; however, she noted nasal hemorrhage again 4 months ago and was admitted to a hospital. Magnetic resonance imaging (MRI) revealed a 43 mm tumor in the left nasal cavity with suggestion of orbit involvement (Figure 1(e)). Positron emission tomography-computed tomography (PET-CT) showed no metastasis. Recurrence of the desmoid-type fibromatosis was clinically considered, and the tumor was surgically removed, weighing 16 g in total and showing a tan-colored cut surface (Figure 1(f)). Microscopically, the lesion was poorly circumscribed and intermingled with hemorrhage (Figure 2(a)). The tumor displayed a fascicular growth pattern with high cellularity and was predominantly

composed of atypical spindle cells with elongated nuclei and scant cytoplasm (Figure 2(b) and (c)). In addition, scattered eosinophilic rhabdomyoblasts and cross-striations in the tumor cells were present (Figure 2(d) and (e)).

Immunohistochemistry revealed that the tumor cells were not reactive for nuclear β -catenin (Figure 2(f)) but were positive for myogenin (Figure 2(g)), myoglobin, desmin, MyoD1 (Figure 2(h)), α -SMA, HNF-35, cytokeratin CAM5.2, and epithelial membrane antigen (EMA). They were negative for cytokeratin AE1/AE3, S-100 protein, HMB-45, and melan A. The Ki-67 labeling index was 9%, especially in hot spots. As these findings suggested spindle cell rhabdomyosarcoma as a diagnosis, the re-evaluation of the initial tumor was performed, revealing that the initial tumor had not been reactive for nuclear β -catenin but was positive for myogenin, myoglobin, and desmin.

She was finally diagnosed with recurrent spindle cell rhabdomyosarcoma. There was no metastasis, and relapse was not reported for 4 years after the second resection.

Discussion

This is a unique case report not only because nasal spindle cell rhabdomyosarcoma is very rare^{3,4} but also because the tumor was initially diagnosed as desmoid-type fibromatosis.

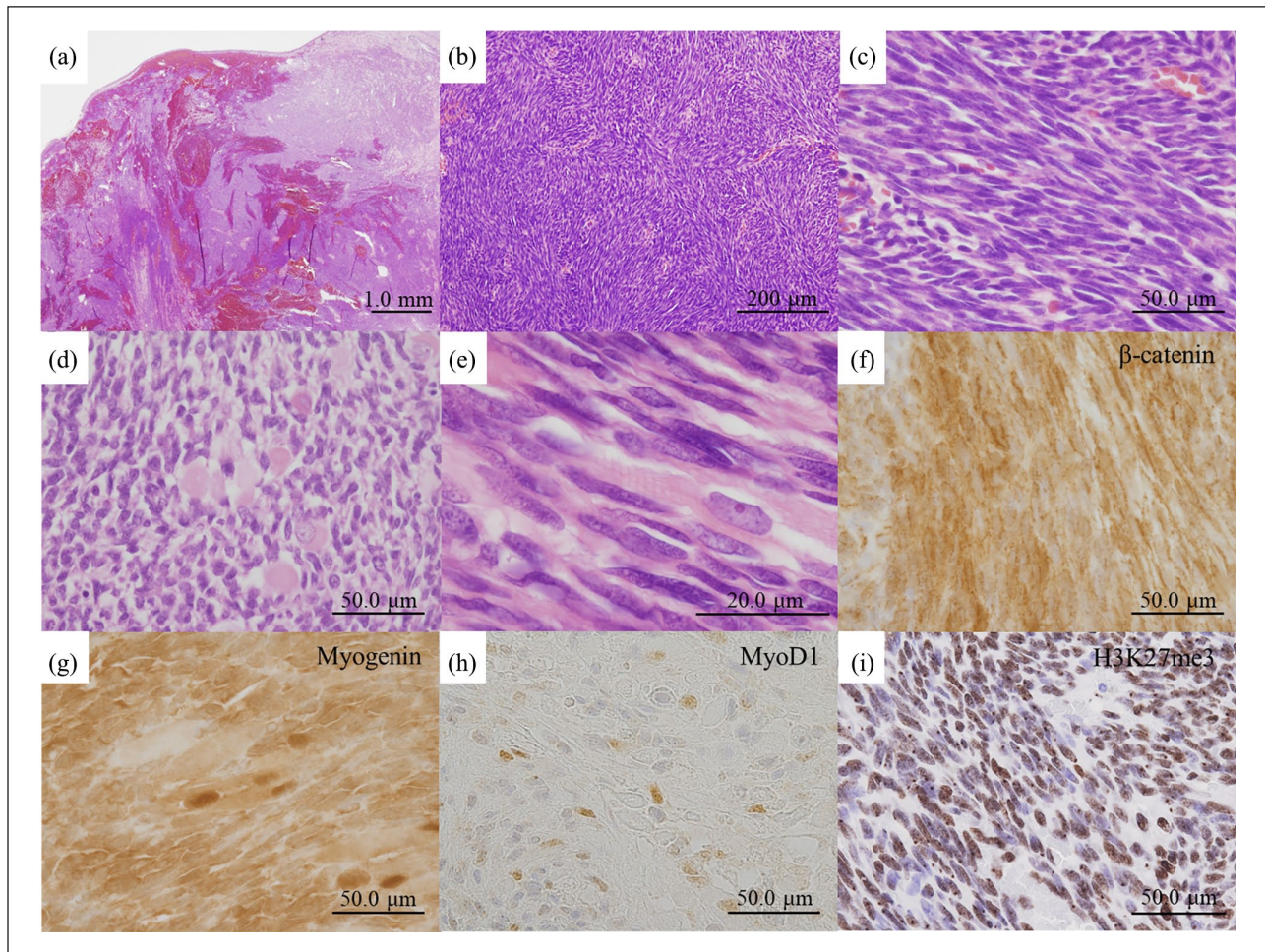


Figure 2. Microscopic and immunohistochemical findings of the recurrent tumor. (a) A low-power view showed a poorly circumscribed lesion intermingled with hemorrhage. Bar = 1.0 mm (H&E staining; original magnification: $\times 12.5$). (b) The tumor showed a fascicular growth pattern with high cellularity. Bar = 200 μm (H&E staining; original magnification: $\times 100$). (c) The tumor was predominantly composed of atypical spindle cells with elongated nuclei and scant cytoplasm. Bar = 50.0 μm (H&E staining; original magnification: $\times 400$). (d) Scattered eosinophilic rhabdomyoblasts were present. Bar = 50.0 μm (H&E staining; original magnification: $\times 400$). (e) A high-power view of tumor cells with cross-striations. Bar = 20.0 μm (H&E staining; original magnification: $\times 1000$). ((f)–(h)) The tumor cells were not reactive for (f) nuclear β -catenin but were positive for (g) myogenin and (h) MyoD1. Bar = 50.0 μm (original magnification: $\times 400$). (i) The tumor was positive for H3K27me3. Bar = 50.0 μm (original magnification: $\times 400$).

In the initial tumor, compared with the recurrent one, rhabdomyoblastic features were not clear. In addition, cytoplasmic strong reactivity for β -catenin of the tumor cells was retrospectively considered to have been confused with nuclear reactivity. In that sense, multiple samplings of tumor tissue may lead to its correct diagnosis. Moreover, nuclear reactivity for β -catenin was also reported in other spindle cell tumors in the sinonasal or oral region such as sinonasal glomangiopericytoma, solitary fibrous tumor, and synovial sarcoma.^{9–11} Our case indicates that nuclear positivity for β -catenin alone is not useful in the investigation of nasal spindle cell tumors and that a broader immunohistochemical evaluation is necessary, including myogenin and MyoD1.

Our second tumor morphologically showed rhabdomyoblastic features and was immunohistochemically positive for

myogenin, MyoD1, and desmin, which are sensitive markers for rhabdomyosarcoma, suggesting rhabdomyoblastic differentiation; however, rhabdomyoblastic differentiation is also seen in other spindle cell tumors.¹² Malignant peripheral nerve sheaths tumor may show rhabdomyoblastic features (malignant triton tumor), and on immunohistochemistry, the tumor is focally positive for S-100 protein; however, our second tumor was negative for S-100 protein. In addition, our re-evaluation of initial and second tumors showed that they were positive for H3K27me3 (Figure 2(i)), the complete loss of which can be seen in malignant peripheral nerve sheaths tumor. Biphenotypic sinonasal sarcoma is a rare low-grade sarcoma that was first described in 2012 as low-grade sinonasal sarcoma with neural and myogenic features, including rhabdomyoblastic differentiation.^{13,14} It characteristically

demonstrates rearrangement of PAX3^{13,14} and shows the immunohistochemical expression of PAX3 and S-100 protein.^{13,14} A study revealed that PAX8 is also positive for biphenotypic sinonasal sarcoma, possibly due to cross-reactivity with PAX3.¹⁵ As this might be a relatively new entity, it was not considered as a differential diagnosis of our tumors when they were diagnosed. We therefore re-evaluated our second tumor, revealing positivity for PAX8. We were not able to investigate the immunohistochemical PAX3 expression or the genetic profile of PAX3 due to the limitation of our study. The expression of PAX3 and PAX8 has also been reported in spindle cell rhabdomyosarcoma;¹⁵ however, we cannot exclude the possibility that our tumor was biphenotypic sinonasal sarcoma.

Conclusion

We reported a case of nasal spindle cell tumor that was initially considered as desmoid-type fibromatosis and finally diagnosed with spindle cell rhabdomyosarcoma. Although further investigation may be required, this notable and diagnostically difficult case is worth reporting. It is important to keep in mind that spindle cell rhabdomyosarcoma is a rare but possible differential diagnoses of nasal spindle cell tumors.

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Author contributions

K.M., M.K., and S.Y. were involved in article concepts and design; K.M. was involved in article preparation; M.K. and S.Y. were involved in article review; and K.M., M.K., A.A., S.T., S.I., H.M., T.N., and S.Y. were associated with the interpretation of this case.

Consent for publication

Consent was obtained for the publication of this case report.

Declaration of conflicting interests

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Ethical approval

Our institution does not require ethical approval for reporting individual cases or case series.

Informed consent

Written informed consent was obtained from the patient(s) for their anonymized information to be published in this article.

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Availability of data and material

The data set supporting the findings and conclusions of this case report is included within this article.

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