CASE REPORT

Primary intraocular synovial sarcoma in the post retinal detachment operative state

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SUMMARY

Synovial sarcoma generally arises in the deep soft tissue, although it has been described at virtually every anatomic site except in the eyeball. We report the case of a 48-year-old woman who had a history of retinal detachment surgery and who had undergone vitrectomy and the insertion of a solid silicon explant 24 years previously. She reported a visual field defect. Funduscopy and MRI revealed a tumour just behind the iris in the left eyeball, and enucleation was performed. Microscopic examination of the tumour revealed uniform spindle cells in a fascicular arrangement with frequent mitotic figures. Immunohistochemistry showed that the tumour was positive for TLE1 and epithelial membrane antigen and fluorescent in situ hybridisation revealed that the tumour had a rearrangement of the SYT gene. Reverse transcription (RT)-PCR confirmed the presence of a SYT-SSX2 fusion transcript. On the basis of these histomorphological and molecular features, the diagnosis of poorly differentiated synovial sarcoma was rendered.

BACKGROUND

This is the first reported case of intraocular synovial sarcoma, and we highlight its distinctive histological and molecular features.

CASE PRESENTATION

A 48-year-old Japanese woman presented with a 2-month history of a left eye visual field defect. She had a medical history of rhegmatogenous retinal detachment of the left eye and had undergone vitrectomy with lens extraction and scleral explant buckling with a solid silicone tyre and encircling band 24 years previously. Funduscopy and MRI revealed a mass just behind the iris in the left eyeball, which showed isointensity in T1-weighted image and slightly high intensity in T2-weighted image, which were enhanced by gadolinium (figure 1A), and 18F-fluorodeoxyglucose positron emission tomography and CT revealed 18F-fluorodeoxyglucose uptake in the mass. The intraocular pressure of the left eye was found to be as high as 44 mm Hg. No other mass or lesion was detected on a whole body scan. Since a malignant tumour was suspected, but fundus examination and MRI were not compatible with malignant melanoma, enucleation of the eye was performed. Grossly, a solid mass measuring 15×10×7 mm was located in the eyeball (figure 1B). Under the equivalent site of the silicon band, the tumour showed invasion to the stroma of ciliary body and lack of continuity of choroid membrane (figure 1C, D). Histologically, the tumour was composed of uniform spindle cells

with a fascicular arrangement, and it contained frequent mitotic figures (up to 30/10 high-power fields; figure 1E). Epithelial nests or glandular structures were absent. Most of the tumour adhered to the surface of the ciliary body and retina, and there was limited infiltration into the stroma of the ciliary body and iris. Under the site of the removed silicon band, the tumour showed clear invasion of the stroma of the ciliary body, and the choroid membrane lacked continuity. The tumour had not infiltrated the optic nerve and sclera. The tumour was graded according to the FNCLCC grading system (stage IIa, T1, N0, M0 and G3). Immunohistochemically, the tumour cells were diffusely positive for TLE1 (figure 1F) and Bmi1 and focally positive for cytokeratin AE1/AE3 and epithelial membrane antigen. Staining for S100 protein, SMA, desmin, caldesmon, myogenin, CD34, HMB45 and melan A was negative. The Ki-67 labelling index of the tumour cells was approximately 50%. Fluorescent in situ hybridisation (FISH) revealed rearrangement of the SYT gene (figure 2A), and SYT-SSX2 (but not SYT-SSX1) fusion transcripts were detected using RT-PCR (figure 2B). The RT-PCR primers and FISH probes have been previously described.^{1 2} On the basis of these findings, the tumour was diagnosed as a poorly differentiated synovial sarcoma.

OUTCOME AND FOLLOW-UP

The patient was tumour free at the 6-month follow-up examination.

DISCUSSION

Synovial sarcoma is a rare soft tissue sarcoma of uncertain differentiation. It occurs most commonly in the deep soft tissue of the extremities. Although there have been six reports of synovial sarcomas in the eye, they occurred either in the orbit (n=5)^{3 4} or the conjunctiva (n=1)⁵; none were intraocular. The case we report here was confined to the eyeball and showed limited infiltration into the stroma of the ciliary body, choroid and iris, and no other lesions were detected by systemic imaging examinations before surgery. Therefore, we consider this tumour to be the first reported case of a primary intraocular synovial sarcoma.

Although most patients with synovial sarcomas have no definitive history of antecedent trauma, some have a history of trauma or previous therapeutic irradiation in the same region.⁶ It is likely that trauma is coincidental rather than a predisposing factor. In our case, the relationship between synovial sarcoma and the patient's history of retinal

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Figure 1 Primary intraocular synovial sarcoma arising after surgery for retinal detachment. (A) MRI revealing a mass in the left eye (gadolinium-enhanced T1-weighted imaging). (B) The gross cut surface of the left eye. Under the silicone band, the lesion had scar tissue (arrowhead) and a greyish-white, firm tumour had adhered to the surface of the retina in the eyeball without necrosis or haemorrhage. *There was a silicone tyre around the sclera that had been inserted in the previous surgery for retinal detachment. (C) Low power. (D) High-power views of a H&E stained section revealing that the tumour is composed of uniform spindle cells in a fascicular arrangement. **Beneath the site from which the silicon band was removed, the tumour had invaded the stroma of the ciliary body, and the choroid membrane lacked continuity. (E) Mitotic figures were frequently encountered. (F) TLE1 immunohistochemical staining of the tumour showing diffuse positivity in the tumour nuclei.

detachment and its treatment is unclear. Cryotherapy or diathermy in the retinal detachment surgery may contribute to form this tumour, but the episcleral silicone band itself had no relationship with tumourigenesis, because the tumour cells did not infiltrate to the sclera, and the intact sclera separated the tumour and silicone band. Synovial sarcomas are difficult to distinguish from malignant peripheral nerve sheath tumours and

fibrosarcomas, as both may share similar morphological and immunohistological properties. However, the t(X; 18) translocation and the SYT-SSX fusion gene are specific markers for synovial sarcoma, and this finding is helpful in confirming the diagnosis. Our case successfully exemplified the usefulness of molecular genetic studies in the final diagnosis of synovial sarcoma arising at unusual anatomical site.

Figure 2 RT-PCR and fluorescent in situ hybridisation (FISH) confirmed the presence of a specific SYT gene fusion. RNA was extracted using the RNeasy formalin-fixed paraffin-embedded tissue RNA Isolation Kit (Qiagen SAS, Courtabeuf, France). (A) The electrophoresis image of RT-PCR products from the SYT-SSX chimeric transcripts. The expected product sizes are 120 bp for the SYT-SSX common site, 87 bp for $\beta 2$ microglobulin (B2M), and 331 bp for both SSX1 and SSX2. The results show that this tumour carried a SYT-SSX2 fusion gene. M: Marker; Lane 1: SYT-SSX; Lane 2: SYT-SSX1; Lane 3: SYT-SSX2; Lane 4: B2M. (B) FISH using the SYT break-apart probe showing an SYT split signal, indicating rearrangement of the SYT gene.

Learning points

- ▶ Synovial sarcoma is a rare soft tissue sarcoma of uncertain differentiation.
- ▶ We consider this tumour to be the first reported case of a primary intraocular synovial sarcoma.
- ▶ Synovial sarcomas are difficult to distinguish from malignant peripheral nerve sheath tumours and fibrosarcomas, as both may share similar morphological and immunohistological properties. However, the t(X; 18) translocation and the SYT-SSX fusion gene are specific markers for synovial sarcoma, and this finding is helpful in confirming the diagnosis.

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