Primitive myxoid mesenchymal tumours of infancy: first case surrounding the optic nerve

Primitive myxoid mesenchymal tumour of infancy is a rare form of sarcoma presenting in infancy.[1] It was first described by Alaggio et al.[2] in 2006 as a primitive mesenchymal soft tissue sarcoma without any distinctive lineage of differentiation. The tumours described initially occurred on the trunk, head, neck, extremities, chest, scalp, abdomen, back, or limbs.[2,3] The tumours initially may be diagnosed as and placed under the broad umbrella of sarcomas, but immunohistochemical and genetic analyses have now categorized the tumour as a definite entity.[3]

We are reporting here a case of primitive myxoid mesenchymal tumours of infancy surrounding the optic nerve. This is the first case reported in this location. The primitive myxoid mesenchymal tumour occurring in association with the optic nerve has not been reported before.

A 1-year-old female child presented with left proptosis since birth that was progressively increasing. On initial examination, she was a healthy child achieving normal milestones. Ophthalmic examination revealed left axial proptosis of 5 mm compared with the right eye. Extraocular movement, pupillary reactions, and fundus examination were normal. Examination of the right eye was unremarkable. No abnormality was found on her systemic examination. On magnetic resonance imaging there was a large retro-ocular mass that seemed to be arising from the optic nerve, pushing the globe forward (Fig. 1). Anterolateral transconjunctival orbitotomy was performed, and an unusual-looking dark violet-colored mass was seen, oozing with viscous fluid, and apparently surrounding the optic nerve (Fig. 2).

To the best of our knowledge, the mass was surrounding the optic nerve snugly, apparently not arising from it. The mass was removed in part. The biopsy was subjected to histopathologic examination (Fig. 3), and it showed sheets and scattered dyshesive ovoid to polygonal cells with eosinophilic cytoplasm and pleomorphic hyperchromatic nuclei in a background of myxoid stroma, suggesting soft tissue sarcoma. Immunohistochemistry showed tumour cells that were vimentin positive, the Ki-67 level was 70%, and epithelial membrane antigen staining was weakly focally positive. Immunoperoxidase stains (Fig. 4) were positive for the *BCOR* gene. However, molecular (cytogenetics) studies were not done. Clinical validation of BCL-6 corepressor (BCOR) was done via immunohistochemistry. The clones of antibodies used were BSB-128. Based on histopathologic examination and immunohistochemistry, a diagnosis of primitive myxoid mesenchymal tumours of infancy was made. After 8 months, patient presented with recurrence. The recurrence was more aggressive than the primary presentation. It was locally aggressive and recurred because of incomplete removal, lesser understanding of the disease, and limited treatment options, that is, local resection. It was a painful blind eye due to severe exposure keratopathy and impending perforation. Enucleation of her left globe with removal of the residual tumor was carried out. The mass removed was subjected to histopathologic examination, which was consistent with the primary diagnosis of primitive myxoid mesenchymal tumours of infancy.

The occurrence of soft tissue sarcomas in infancy is rare, but the most common ones are embryonal rhabdomyosarcoma, congenital fibrosarcoma, Ewing sarcoma, and primitive sarcoma such as undifferentiated sarcoma.[2] The first case of primitive myxoid mesenchymal tumour of infancy in orbit was reported by Hayes et al.[3] involving an otherwise healthy 8-month-old female with a 6-week history of progressive proptosis. The tumour was along the lateral wall of the orbit. Initially, the patient underwent debulking biopsy with the residual disease, which recurred 12 months after the first surgery.[3] In our case, the tumour was seen to be surrounding the optic nerve, which has not been reported before. The patient presented with regrowth 8 months after the initial surgery, which unfortunately ended up in enucleation. There was an early recurrence of the tumour in both reported cases. This indicates the aggressive behavior of the tumour and the dire need for complete excision to prevent the recurrence of the residual disease. The case presented here is unique because it is the first case of primitive myxoid mesenchymal tumours of infancy surrounding the optic nerve. To our knowledge, no other case has been reported so far. The only case of orbital tumour reported in 2020 was extraconal, arising in the lateral wall of the orbit and sphenoid trigone and extending into the lateral orbit.[3] Both patients were female. In 2006 Alaggio et al.[2] reported 6 cases of primitive myxoid mesenchymal tumours of infancy. Of these, 3 cases were congenital. These tumours occurred in the trunk, extremities, and head and neck. In 2010, Mulligan et al.[4] reported an 8-month-old child with a tumour occurring on thenar eminence. After conservative excision, the lesion recurred, necessitating partial amputation because it was nonresponsive to chemotherapy.

Primitive myxoid mesenchymal tumor of infancy is a rare entity. It is a tumour of early childhood characterized by local infiltration of surrounding structures that is aggressive and has a poor response to chemotherapy, but metastases are rare.[5] In 2019, the first case of primitive myxoid mesenchymal tumour of infancy with brain metastasis was reported by Saeed et al.[6] The metastasis occurred in the cerebellum, and the primary tumour was located in the extremity and had already been treated with surgery and chemotherapy. Immunohistochemistry has an important role in diagnosing different types of mesenchymal tumours. The biological insight into various tumour types is amplified by locating

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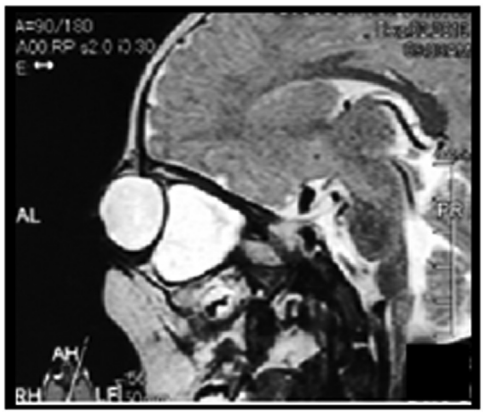

Fig. 1—Magnetic resonance imaging showing mass surrounding the optic nerve.

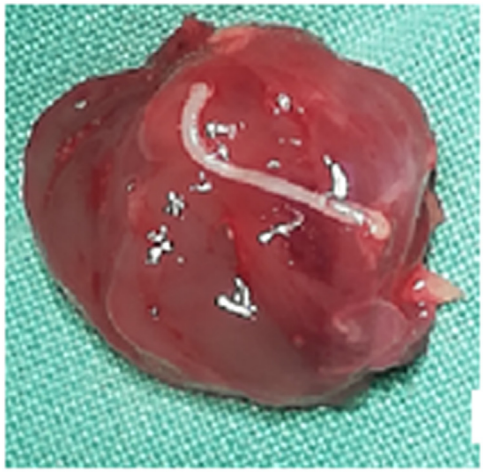

Fig. 2—Gross picture of the resected mass.

the genomic alteration in various mesenchymal neoplasms.[7] In primitive myxoid mesenchymal tumour of infancy, on immunohistochemical analysis, diffuse reactivity is seen for vimentin, whereas there is no reactivity for muscle-specific actin, S-100, desmin, or myogenin.[2]

In the case reported here, vimentin was positive, the Ki-67 level was 70%, and epithelial membrane antigen staining

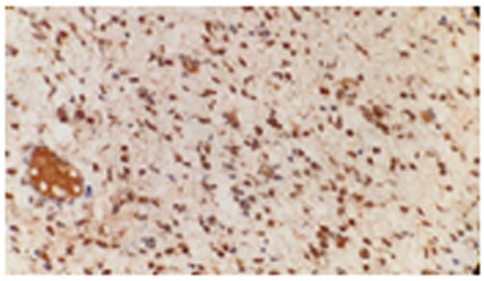

Fig. 4—Immunoperoxidase staining positive for BCOR.

was weakly focally positive. All skeletal muscle markers were negative, whereas immunoperoxidase stain was positive for BCOR. The immunoperoxidase technique is one of the methods used in immunohistochemistry, and in our case, BCOR was tested using the same technique. *BCOR* is a gene encoding for an epigenetic regulator involved in body structure development and cell differentiation. In almost all cases of primitive myxoid mesenchymal tumours of infancy, overexpression of BCOR is seen.[8] Primitive myxoid mesenchymal tumour of infancy is a myofibroblastic tumour with a low potential for metastasis but a high local recurrence rate.

The occurrence of this rare entity in the orbit is unique; on a literature search, we found that only one other case has been reported previously. This is the second case we are reporting here. The location of the tumour in orbit is different in each case. Both patients were female, and both presented with recurrence within 1 year of the initial surgery, emphasizing the fact that complete and meticulous surgical excision is required because the response of the tumour to chemotherapy is poor and the chances of the early recurrence from the residual disease are very high.

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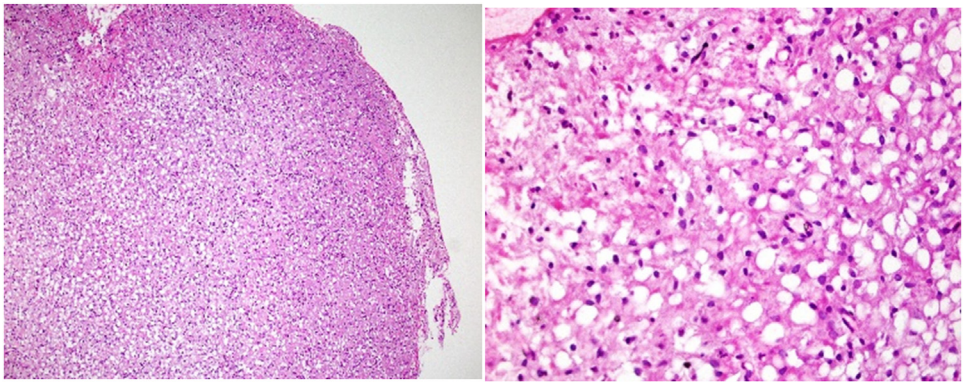

Fig. 3—Histopathologic image (low and high power).

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Footnotes and Disclosure

The authors have no proprietary or commercial interest in any materials discussed in this article.