BACKGROUND

- EWSR1::NFATC2-rearranged tumors constitute an emerging spectrum of disease entities.
- Solitary bone cysts in the young and vascular malformations/hemangiomas of the bone in elderly patients have been reported to harbor EWSR1::NFATC2 rearrangements and display benign behavior.
- EWSR1::NFATC2 fusion-positive round cell sarcomas are a recently described subtype of “Ewing family” tumors with highly aggressive behavior and poor response to chemotherapy.
- A recent cohort of five adult patients with epithelioid vascular lesions harboring EWSR1::NFATC2 rearrangements was described with cytologic atypia, including a distinct morphology comprised of alternating vasoformative and solid growth and mild to moderate nuclear pleomorphism.

CASE SUMMARY

- An 11-year-old female presented an extensive history of vertebral bony pathology.
- At 2 years of age, the patient presented with an L4 vertebral fracture with pathology showing a fibrovascular lesion with hemosiderin deposition.
- At 6, 8, and 10 years of age, sequential biopsies of a progressive L4 vertebral mass resulted in the diagnosis of a benign vascular malformation. (Fig. 1)
- At the most recent presentation, imaging revealed a 4.4 x 4.8 x 7.4 cm mass expansile into the retroperitoneum. (Fig. 1)
- Grossly, the mass was red-brown, lobulated, and firm, with cut surfaces demonstrating bony tissue with marked hemorrhage. (Fig. 2)
- Histologically, the tumor contained large thin- and thick-walled vascular structures with foci of intraluminal endothelial cell proliferation and epithelioid endothelial cells with atypical mitoses (Fig. 3a-c).

RESULTS

- Immunohistochemically, intraluminal spindle cells were CD31+ (Fig. 3d), CD34- (Fig. 3e), and Prox1+ (Fig. 3f). Chromogranin, CD56, panCK, EMA, BCL-2, CD99, NKK2.2, P53, myogenin, Phox2b, Glut-1, HMB-45, HHV-8, S-100, CAMTA1, and TFE3 were negative. BAF47/INI1 was retained. Ki-67 was focally increased with associated epithelioid cells.

NGS RNA sequencing revealed an EWSR1::NFATC2 fusion, later confirmed by breakapart EWSR1 FISH (Fig. 3g).

- The patient is currently disease free 6 months after bulk resection and is maintained on sirolimus therapy.

DISCUSSION

- The findings are consistent with a vascular anomaly with EWSR1::NFATC2 rearrangement, scattered proliferative epithelioid endothelial cell clusters, and rare atypical mitoses.
- The patient’s very early age at initial presentation and epithelioid and intravascular endothelial proliferations represent a unique pediatric presentation of an EWSR1::NFATC2-rearranged epithelioid vascular tumor with apparent aggressive behavior.

REFERENCES