Gorham-Stout syndrome (Vanishing bone disease): A case of a rare entity

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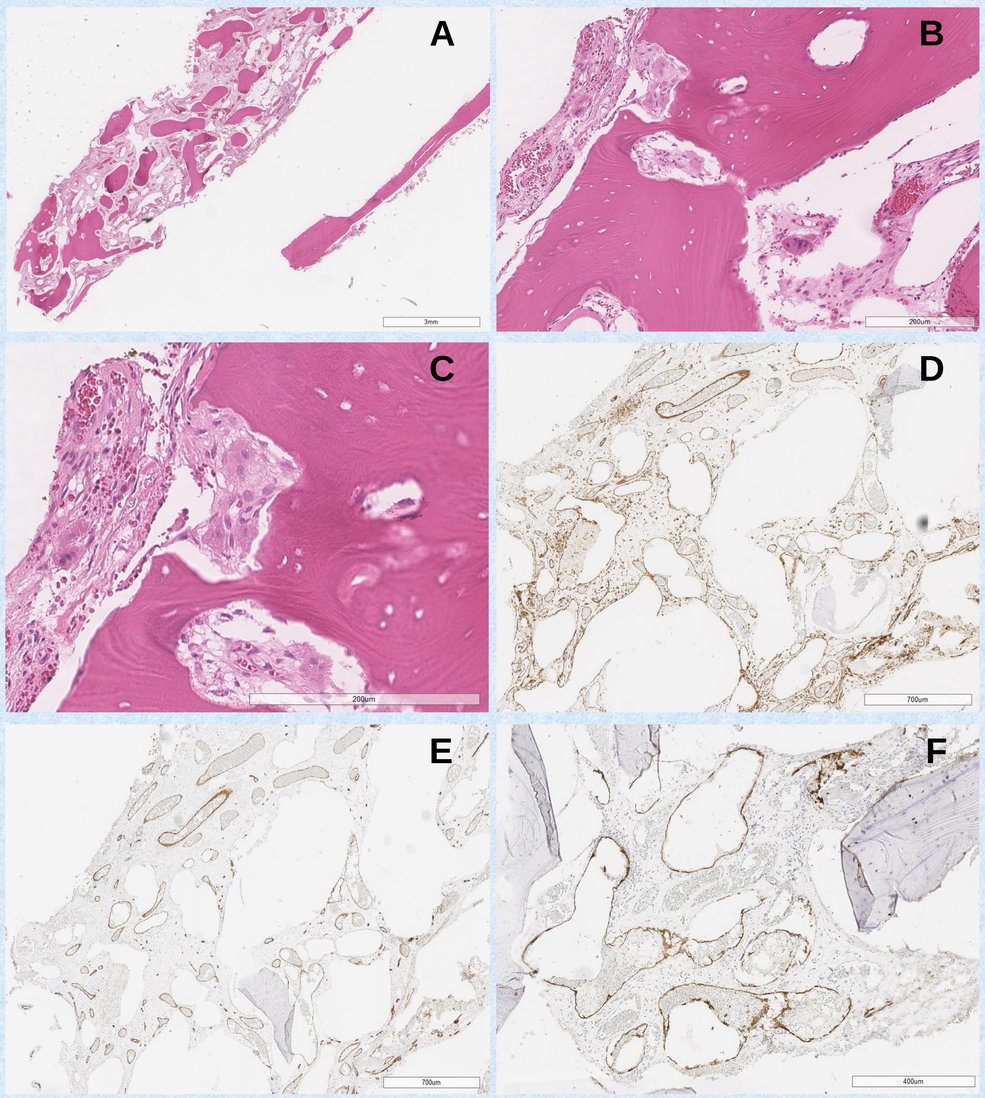
Clinical features:

A 17-year-old female presented with right femur pathological fracture and AVM in 7/2016, she underwent intermedullary nailing and open reduction internal fixation. She complained increasing right hip pain in 04/2017. X-rays demonstrated hardware failure. Biopsy showed vascular tissue, and she was started on systemic therapies (bisphosphonates, sirolimus) in 6/2017 with outpatient management to optimize bone tissue given chronic non-union of fractures despite fixation with pain. On 1/5/18 she underwent revision with hardware replacement with negative bone cultures. Due to persistent right hip pain, on X-ray showed right femoral neck fracture around the nail and she was admitted for a right femur removal of hardware and proximal femoral replacement. At baseline she has been modified independent with ambulation using axillary crutches for at least 2 years after the initial surgical interventions.

Results:

CT and X-ray shows intraosseous hemangioma, diffuse demineralization and osteolysis of the proximal femur, underlying severe osteoporosis and tapering of bone. Multiple pathology reviews showed fragments of viable bones with remodeling changes, negative for acute inflammation (A-C); Benign lymphovascular proliferation with network of thin-walled vessels highlighted by CD31 (D), CD34 (E), and D2-40 (F). Osteoclasts are noted within the adjacent bones, and demonstrate scalloped osteoclastic activity.



Conclusion and discussion:

Gorham-Stout syndrome is an aggressive form of skeletal angiomatoses, it usually affects children or young adults, characterizes by progressive destruction/absorption of osseous matrix, and overgrowth of vascular structures. It involves multiple bones including the skull, the maxillofacial region, the spine and pelvis, the proximal parts of the appendicular skeleton, like this case, the proximal femur. Due to the variable and unpredictable clinical course of massive osteolysis, and the reported serious complications in some cases, the treatment of choice is still evolving: surgical resection, radiation therapy, embolization, and systemic therapies i.e., bisphosphonates, sirolimus, steroids, and interferon-α have all been tried with various progression.