Popliteal pseudoaneurysm in a young patient with multiple hereditary exostosis

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ABSTRACT

Multiple hereditary exostosis is an osteogenic disorder that causes outgrowths of cartilaginous bone tumors that are associated with adjacent neurovascular compressive injuries. We present the case of an adolescent male with multiple hereditary exostosis complicated by popliteal pseudoaneurysm formation who underwent excision of the osteochondroma and vein patch angioplasty repair of the artery. We highlight the rare association between this genetic disease and subsequent vascular complications and review the available literature of arterial complications of this disease. (J Vasc Surg Cases Innov Tech 2023;9:101291.)

Keywords: Multiple hereditary exostosis; popliteal pseudoaneurysm

Multiple hereditary exostosis (MHE) is an autosomal-dominant disorder characterized by outgrowths of cartilaginous bone tumors located at the metaphysis of bones that undergo endochondral ossification. Complications include osteoarticular deformities, fractures, and chondrosarcomatous malignant transformation.[1,2] Rarely, these osteochondromas can compress adjacent neurovascular structures, resulting in paresthesia, occlusions, dissections, and aneurysms. Vascular complications of MHE are rare, with a minority of cases affecting the lower extremity arteries in young patients.[3] We describe a rare case of popliteal artery pseudoaneurysm in an adolescent patient with MHE. A PubMed literature review using search terms "multiple hereditary exostosis" as well as "hereditary multiple osteochondromas" was performed. Search results were then reviewed, adding further cited sources if applicable, yielding 18 cases of vascular complications as part of a literature review of this rare entity. Written informed consent was obtained for his case details and imaging studies.

CASE REPORT

A 17-year-old Caucasian male with known MHE presented to the orthopedic oncology clinic with atraumatic, worsening leg pain for 1 month with associated thigh swelling. He was diagnosed with MHE at 12 years of age and has a positive family history for MHE in his mother and maternal grandmother. Physical examination revealed a palpable, tender, firm mass over the left distal posteromedial knee and a nontender mass over the proximal medial tibia. Close examination of the thigh showed pulsatile movement of the skin. Magnetic resonance imaging was concerning for a popliteal artery pseudoaneurysm adjacent to an osteochondroma, and follow-up computerized tomography angiography revealed a 4 × 7 × 8 cm pseudoaneurysm of the above-knee popliteal artery immediately adjacent to a large distal femoral exostosis (Figs 1 and 2). He was admitted and taken to the operating room with orthopedic oncology and vascular surgery on the day of presentation.

Procedure. A longitudinal incision was made over the midsuperficial femoral artery, extending to the above-knee popliteal artery, obtaining proximal and distal control. Upon entering the pseudoaneurysm, a femoral osteochondroma with serrations just medial to the artery was visualized. A clear 5-mm longitudinal defect of the popliteal artery was identified as the source of the pseudoaneurysm (Fig 3). The artery appeared friable and unsuitable for primary repair, and the decision was made to resect to healthy margins and perform a vein patch angioplasty using the ipsilateral great saphenous vein. The orthopedic oncology team then marginally excised the broad-based serrated osteochondroma measuring 3 cm.

He was placed on aspirin 81 mg/day and discharged home on post-operative day 3. At the 1-month follow-up, duplex ultrasound examination revealed a widely patent repair, and radiographs revealed expected postoperative changes. Four months postoperatively, he was asymptomatic without significant changes on repeat radiographs and was cleared for full weight-bearing and physical activity.

DISCUSSION

MHE is an autosomal-dominant condition that ranges from asymptomatic, cosmetic deformities to painful osteochondromas beginning in early childhood and progressing throughout puberty. This rare disorder is associated with mutations in the *EXT1* and *EXT2* genes, which are responsible for encoding for proteins involved in heparan sulfate maturation/glycosylation, with a predicted incidence of 1 in 50,000 individuals, although it has been reported to vary geographically.[4] Defects within this process result in poor chondrocyte differentiation

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Author conflict of interest: none.

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The editors and reviewers of this article have no relevant financial relationships to disclose per the Journal policy that requires reviewers to decline review of any manuscript for which they may have a conflict of interest.

2468-4287

https://doi.org/10.1016/j.jvscit.2023.101291

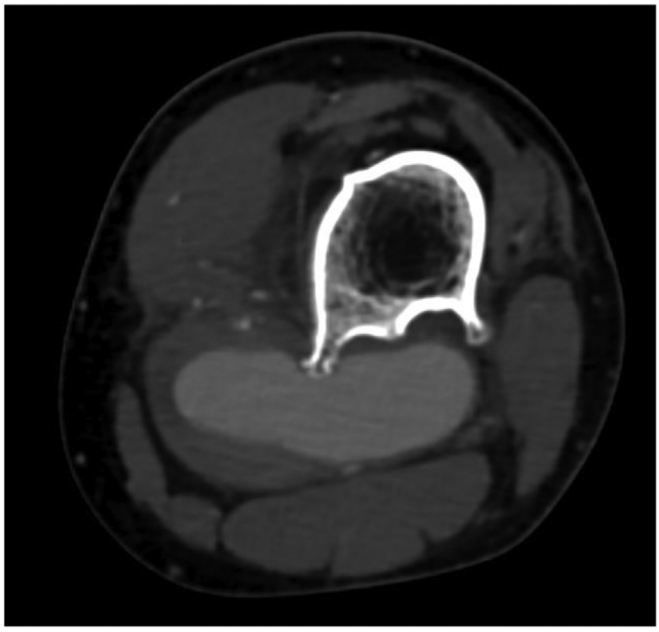

Fig 1. Axial computed tomography angiography revealed popliteal artery pseudoaneurysm secondary to erosion of an adjacent femoral exostosis.

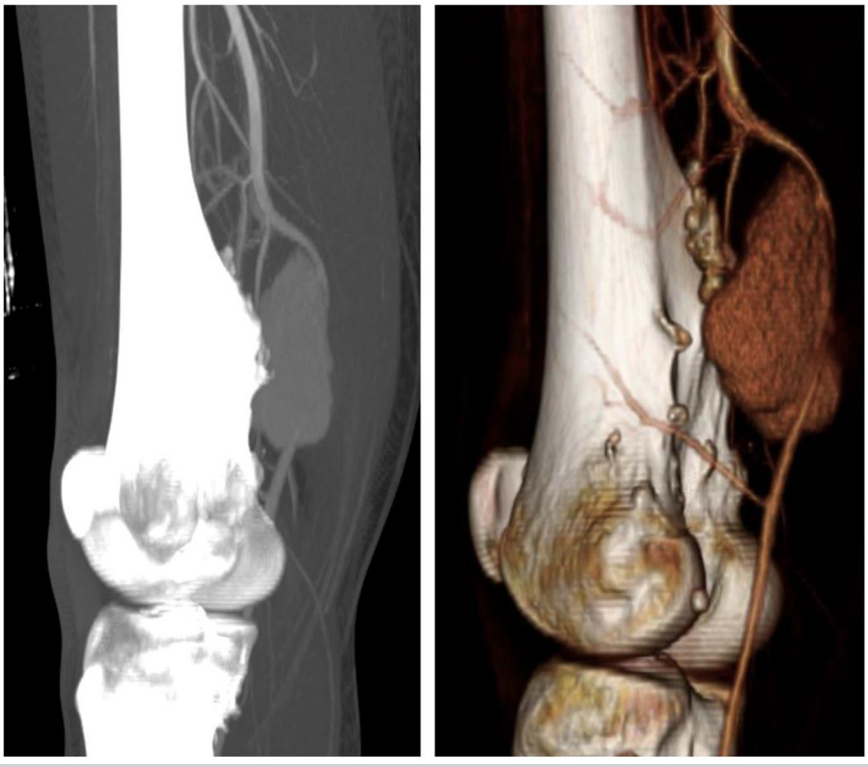

Fig 2. Sagittal computed tomography angiography (left) and three-dimensional reconstructed figure (right), demonstrating the extension of the exostosis into the adjacent popliteal artery, with normal vessel proximally and distally.

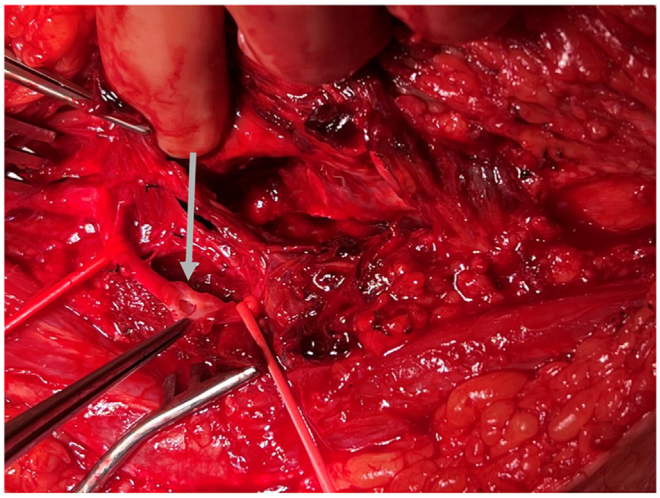

Fig 3. Exposure of the above-knee popliteal artery with short-segment erosive defect amenable to patch angioplasty.

and impair the endochondral ossification process, resulting in these outgrowths of cartilaginous tumors.[5] Growths can be identified as early as 5 years of age and, as suggested by the nomenclature, have a cartilaginous cap that is initially less likely to be traumatic to nearby structures. As endochondral ossification occurs, the cartilage cap ossifies and is more likely to cause compressive and traumatic effects on nearby structures, including vessels. This compression is worsened through continued and repetitive motions, and can result in a multitude of vascular or neurovascular complications. We report one such case with a popliteal artery pseudoaneurysm.

A minority of individuals with MHE experience significant morbid events, which include fractures, osseous deformity, compression of nerves, and compression of vasculature; <5% of cases are expected to involve malignant transformation.[6] Recurrence rates are reported to be nearly zero with complete excision.[7] Complications involving the surrounding vasculature are even more rare, with the most common being a popliteal arterial aneurysm, as in our case. Other vascular complications seen in the current published literature, primarily in small series, are aneurysms, dissections, claudication, phlebitis, acute limb ischemia, and thoracic outlet syndrome, involving several arterial beds (including the femoropopliteal, subclavian, vertebral, and basilar arteries).

In treating individuals with lower extremity arterial complications secondary to MHE, there is no standard of care, owing to its uncommon occurrence. Nonetheless, previously published reports[8-24] have described nearly identical modes of management; namely, excision of the culprit osteochondroma and open vascular repair (Table). Only one case was managed medically, an early report of superficial femoral artery occlusion[8] that yielded recanalization and improvement in ischemic rest pain with only anticoagulation and lumbar sympathectomy. The remaining reports all involved excision and arterial repair, with either vein patch,[10] prosthetic patch,[18,24] saphenous vein bypass,[12-15,17] primary repair,[11,16,19,21] or end-to-end reanastomosis.[3,20] In the only case involving the profunda femoris artery,[22] the pseudoaneurysm was coil embolized before excision. Endovascular repair without managing the compressive mass is likely less favorable, but could be used a temporizing measure in an unstable or high-risk patient. Because the majority of reported individuals are young, the long-term durability of open repair, as opposed to that of stent graft placement, should be an important consideration. With respect to the treatment of popliteal aneurysms not secondary to MHE, although endovascular repairs are increasing in frequency, they offer little to no benefit in limb salvage or mortality. Additionally, they were associated with equivocal or higher reintervention rates compared with open surgical treatment.[25-28] Overall, given the younger patient population presenting with MHE-related arterial complications, the need for excising the exostoses, and the improved durability/decreased reintervention rate, open repair is likely superior in treating these complications. More research is needed to evaluate these treatment modalities in this uncommon scenario.

CONCLUSIONS

Here, we describe a rare case of popliteal artery pseudoaneurysm in an adolescent patient with MHE treated with an open surgical approach. We recommend open

Table. Literature review of cases of MHE affecting the femoropopliteal artery

Authors	Age, years	Complication	Presentation	Treatment(s)
De Matos et al (1983)[6]	34	Superficial femoral artery occlusion	Acute onset short-distance claudication, rest pain	Anticoagulation and lumbar sympathectomy
Recht et al (1993)[7]	13	Popliteal artery pseudoaneurysm	Ecchymosis, pain, swelling	Excision, no details regarding vascular repair
Qattan et al (1994)[8]	23	Popliteal artery pseudoaneurysm	Swelling, claudication	Excision and saphenous vein patch
Chamlou et al (2002)[9]	21	Popliteal artery pseudoaneurysm	Pulsatile mass, swelling	Excision and primary repair of defect
Burstzyn et al (2005)[10]	12	Superficial femoral artery pseudoaneurysm	Swelling, pain	Excision and saphenous vein bypass
Al-Hadidy et al (2007)[11]	16	Popliteal artery pseudoaneurysm	Swelling, pain with activity	Excision and saphenous vein bypass
Toumi et al (2010)[12]	38	Thrombosed popliteal artery, popliteal venous thrombosis	Swelling, cyanosis	Excision and saphenous vein bypass
Chaouch et al (2011)[13]	20	Popliteal artery aneurysm	Swelling, intermittent claudication	Excision and saphenous vein bypass
Pavic et al (2011)[14]	14	Popliteal artery pseudoaneurysm	Pain, pulsatile mass	Excision and primary repair
Vanhegan et al (2012)[15]	21	Popliteal artery pseudoaneurysm & rupture	Prominent ecchymosis, swelling, pain	Excision saphenous vein bypass
Rangdal et al (2013)[16]	8	Popliteal artery pseudoaneurysm	Swelling	Excision and PTFE patch
Jamieson et al (2014)[17]	20	Popliteal artery pseudoaneurysm	Swelling	Excision and primary repair
Onan et al (2014)[18]	12	Popliteal artery pseudoaneurysm, nerve compression	Knee swelling, motor deficits	Excision and end-to-end reanastomosis
Aouini et al (2015)[19]	52	Superficial femoral artery pseudoaneurysm, deep vein thrombosis, nerve compression	Chronic numbness and weakness, swelling	Excision and primary repair
Nasr et al (2015)[3]	17	Thrombosed superficial femoral artery pseudoaneurysm	Acute limb ischemia	Excision and end-to-end reanastomosis
Trivedi et al (2016)[20]	28	Profunda femoris artery pseudoaneurysm	Pain	Coil embolization followed by excision
Ferrari et al (2017)[21]	81 (deceased, autopsy study)	Popliteal artery aneurysms	Bilateral popliteal pseudoaneurysms on autopsy	n/a
Syed et al (2019)[22]	35	Popliteal artery pseudoaneurysm	Pain, swelling, pulsatile mass	Excision and PTFE patch

n/a, Not applicable; *PTFE*, polytetrafluoroethylene.

surgical procedure to allow for treatment of the osteochondroma while providing a durable vascular repair.

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Submitted Apr 14, 2023; accepted Jul 24, 2023.