Multiple hereditary exostoses: A pseudoaneurysm masquerading as tumor

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Radiology Case. 2016 Aug; 10(8):50-59 :: DOI: 10.3941/jrcr.v10i8.2849

ABSTRACT

Multiple hereditary exostoses is an autosomal dominant condition characterized by numerous benign osteochondromas. Complications are rare and can include deformity, growth abnormality, fracture, adventitial bursa formation, local mass effect on a nerve, malignant degeneration, and vascular complications including stenosis, occlusion, arteriovenous fistula, and pseudoaneurysm. We present a case of multiple hereditary exostoses leading to a deep femoral artery pseudoaneurysm in the proximal medial thigh with subsequent rupture and hematoma, masquerading as tumor.

CASE REPORT

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A 28 year-old man presented with excruciating groin pain following a run. The pain was described as electric and occurring in waves lasting 15-20 minutes, severe enough to require transport via ambulance. The patient denied any immediate preceding major trauma, but he described minor trauma in the region several months prior with soreness and bruising which was self-limited. He denied any back pain, claudication symptoms, and had no history of preexisting vascular disease.

Initial radiographs demonstrated multiple exostoses of the bilateral femurs and left iliac wing, including a large exostosis of the left lesser trochanter (Figure 1). The patient reported a prior diagnosis of multiple hereditary exostoses (MHE), but he had not been seen in follow up since childhood. His pain self-resolved and the remaining exam was normal, so he was discharged with presumed diagnosis of muscular cramps and an appointment for magnetic resonance imaging (MRI) and orthopedic surgery follow-up.

The patient was lost to follow-up for 6 months, at which point he underwent MRI demonstrating numerous exostoses of the pelvis and left femur, including the aforementioned large left lesser trochanter exostosis extending to the metaphysis. The exostosis was surrounded by a 11 x 10 x 6 cm heterogeneously enhancing lesion extending inferiorly, with mass effect on the hamstring musculature and surrounding soft tissues (Figure 2).

An ultrasound was performed for further characterization and potential biopsy of the soft tissue mass. The ultrasound demonstrated a 3.6 x 2.8 x 2.4 cm pseudoaneurysm arising from a branch of the deep femoral artery surrounded by a large hematoma (Figure 3), similar in size to that seen on MRI. There was no evidence of pseudoaneurysm rupture and no flow demonstrated within the hematoma. Biopsy was deferred. A computed tomography (CT) angiogram obtained two weeks later (Figure 4) demonstrated a non-opacified pseudoaneurysm without evidence of active extravasation. It is favored that the non-opacification was due to delayed filling rather than

thrombosis, as both the prior ultrasound and subsequent conventional angiogram demonstrated patency.

The patient underwent definitive treatment consisting of coil embolization of a left medial femoral circumflex artery pseudoaneurysm (Figure 5) and resection of the left lesser trochanteric exostosis, with excellent results. There was contrast extravasation from the pseudoaneurysm on angiography at the time of resection, indicating likely ongoing intermittent hemorrhage.

The lesion was removed piecemeal and consisted grossly of innumerable fragments of hard white-tan bone and blood clot. Pathology demonstrated focal cartilage with endochondral ossification as well as a thin capsule of synovial metaplasia with fibrin, compatible with hematoma capsule (Figure 6).

Six months following surgery, the patient continues to do well; he is ambulating freely and reports no further pain or swelling in the region.

DISCUSSION

Etiology & Demographics:

Osteochondromas are the most frequent benign bone tumor, with low rates of malignant degeneration and complication. When seen in multiplicity, they can be associated with an autosomal dominant condition called multiple hereditary exostoses[1-3]. The prevalence of MHE in the population is reported to be 0.9 – 2 per 100,000, and the median age at the time of diagnosis is 3, with nearly all cases diagnosed by age 12[1-3]. There is an approximate 1.5:1 male to female predominance[3].

Malignant transformation of osteochondromas is rare, occurring in approximately 5% of patients with MHE compared to 1% in sporadic cases [3, 4]. Non-malignant complications are more common and include deformity, growth disturbance, and bursa formation; fractures and localized mass effect on vessels and nerves[5-7] are less frequent. Vascular complications may include stenosis, occlusion, arteriovenous fistula formation, or, as in this case, pseudoaneurysm[7-12]. Pseudoaneurysm is the most frequent vascular complication and occurs secondary to repeated abrasion by a large or growing osteochondroma, which eventually erodes the arterial surface[12-14].

Clinical & Imaging Findings:

Osteochondromas are benign, cartilage-capped osseous outgrowths whose characteristic feature is continuity of the cortical surface and medullary space with the underlying bone. Multiple osteochondromas on bone survey is essentially diagnostic of MHE.

Again, pseudoaneurysm is the most common vascular complication. The most commonly affected vessel is the popliteal artery, largely due to the high frequency of osteochondromas in the distal femur combined with the lack of mobility of the vessel as it is tethered in Hunter's canal [11]. However, as demonstrated in this case, pseudoaneurysm can occur anywhere in the setting of a sufficiently large osteochondroma adjacent to a vessel.

Localized pain in a patient with known MHE should raise the suspicion of complication, which can be further evaluated with MRI to both confirm the diagnosis of osteochondroma and allow evaluation for complications or malignant degeneration to chondrosarcoma[15]. In this case, the MRI findings of heterogeneous T1 and T2 signal with irregular pattern of enhancement were suspicious for malignant transformation with an associated large soft tissue component. Malignant degeneration to chondrosarcoma should also be suspected if the osteochondroma's cartilaginous cap exceeds 2 cm in thickness, an example of which is shown in Figure 7.

Although a hematoma secondary to pseudoaneurysm was included in the differential, the diagnosis was only confirmed after obtaining a diagnostic ultrasound. This highlights the value of ultrasound imaging in identifying potential vascular complications in patients with osteochondromas, particularly before pursuing biopsy which could have led to further morbidity [12]. In this particular case, CT angiography did not identify an opacified pseudoaneurysm or active extravasation, although this was identified at the time of surgery, highlighting the possibility of intermittent bleeding of the pseudoaneurysm as the vascular wall is repeated abraded.

Treatment & Prognosis:

Treatment of pseudoaneurysms is surgical, with either vascular coil embolization or bypass grafting of the lesion. Resection or debulking of the osteochondroma to decrease localized mass effect and deformity is also critical at the time of surgery [14]. The overall prognosis is excellent in these cases.

Differential Diagnoses:

In a patient with known MHE with a new soft tissue mass on CT or MRI, the primary differential diagnosis would be malignant degeneration of an osteochondroma to chondrosarcoma.

When considering a classic isolated osteochondroma, the main differential consideration would be bizarre parosteal osteochondromatous proliferation (BPOP) which is typically also seen in the hands and feet of younger patients. BPOP appears as a pedunculated or sessile mineralized mass on a tubular bone surface, but without continuity of the medullary cavity. 70% occur in the hands and feet, with the remainder occurring in the long bones.

TEACHING POINT

A heterogeneous, enhancing mass abutting an osteochondroma should raise the suspicion for malignant degeneration versus neurovascular complication, including ruptured pseudoaneurysm. If vascular complication is suspected, follow up interrogation with Doppler ultrasound is critical in identifying the vascular nature of the lesion and to preclude the disastrous complication which may result from biopsy of these lesions.

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FIGURES

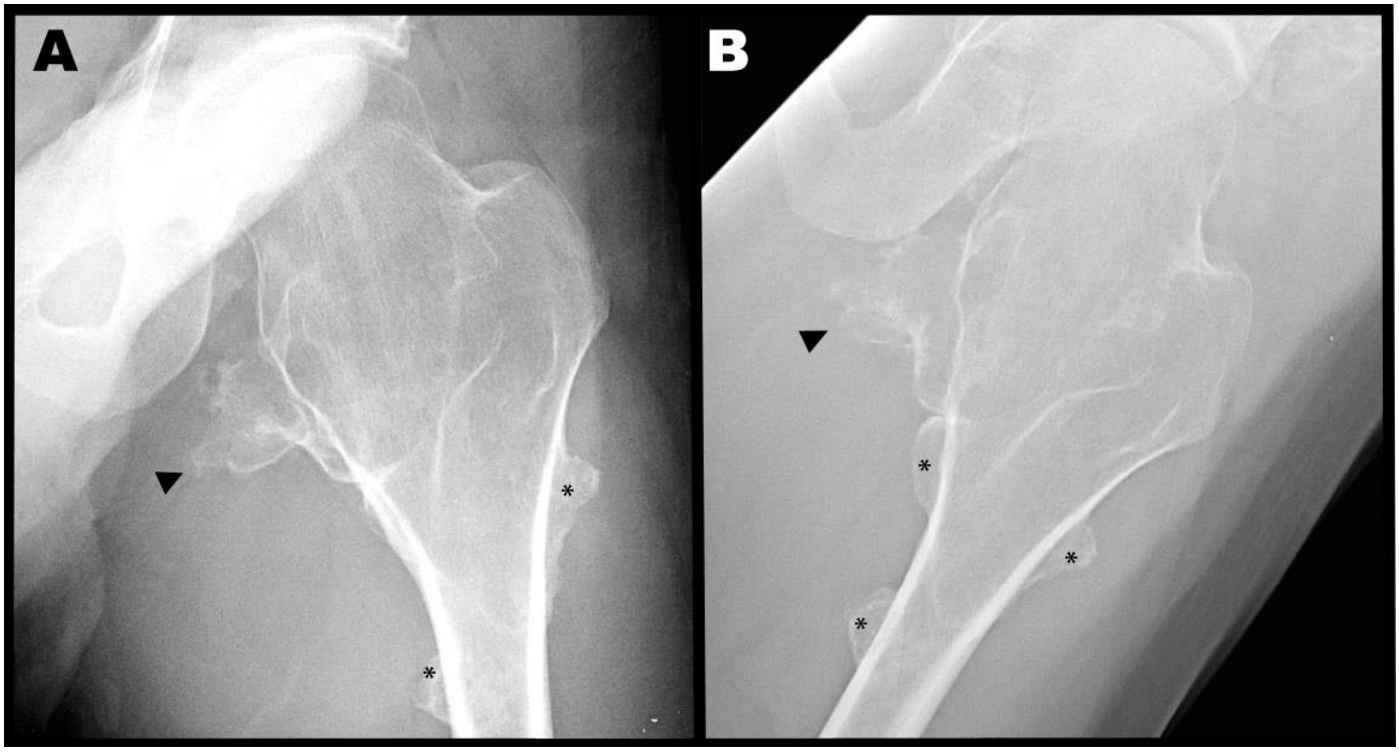

Figure 1: 28 year-old man with MHE with ruptured pseudoaneurysm secondary to large exostosis.
Findings: Anteroposterior (A) and oblique (B) radiographs of the left proximal femur demonstrate numerous exostoses involving the lesser trochanter, femoral neck, and proximal femoral diaphysis (*). There is cortical irregularity of the exostosis arising from the lesser trochanter (arrowhead)

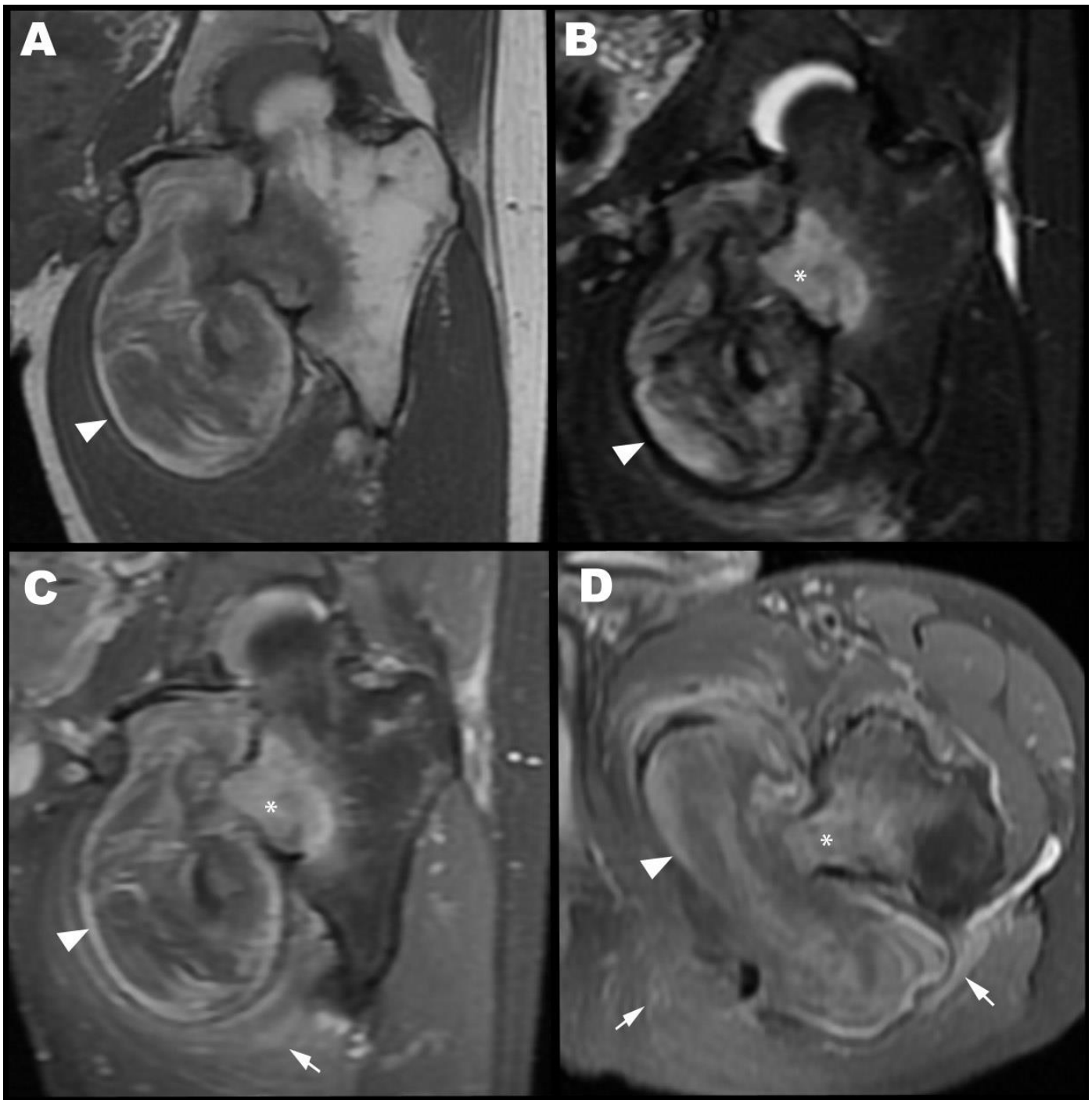

Figure 2: 28 year-old man with MHE with ruptured pseudoaneurysm secondary to large exostosis.
Findings: Coronal T1 (A) and STIR (B) MRI images through the proximal femur demonstrate a large, heterogeneous lesion (arrowheads) adjacent to an osteochondroma of the lesser trochanter. The osteochondroma demonstrates increased T2 signal extending into the subjacent femur (*), compatible with edema. T1 fat-saturated post-gadolinium coronal (C) and axial (D) images demonstrate irregular enhancement of the osteochondroma (*), adjacent mass (arrowheads), and hamstring musculature (arrows).
Technique: MRI: 1.5T Coronal T1 (TR: 668, TE: 12.59); 1.5 T Coronal STIR (TR: 5900, TE: 72.75); 1.5T Coronal T1 fat-saturated post-gadolinium (TR: 672 TE: 12.41), Agent: gadobutrol (10 mL), venous phase; 1.5T Axial T1 fat-saturated post-gadolinium (TR: 927, TE: 12.53), Agent: gadobutrol (10 mL), venous phase.

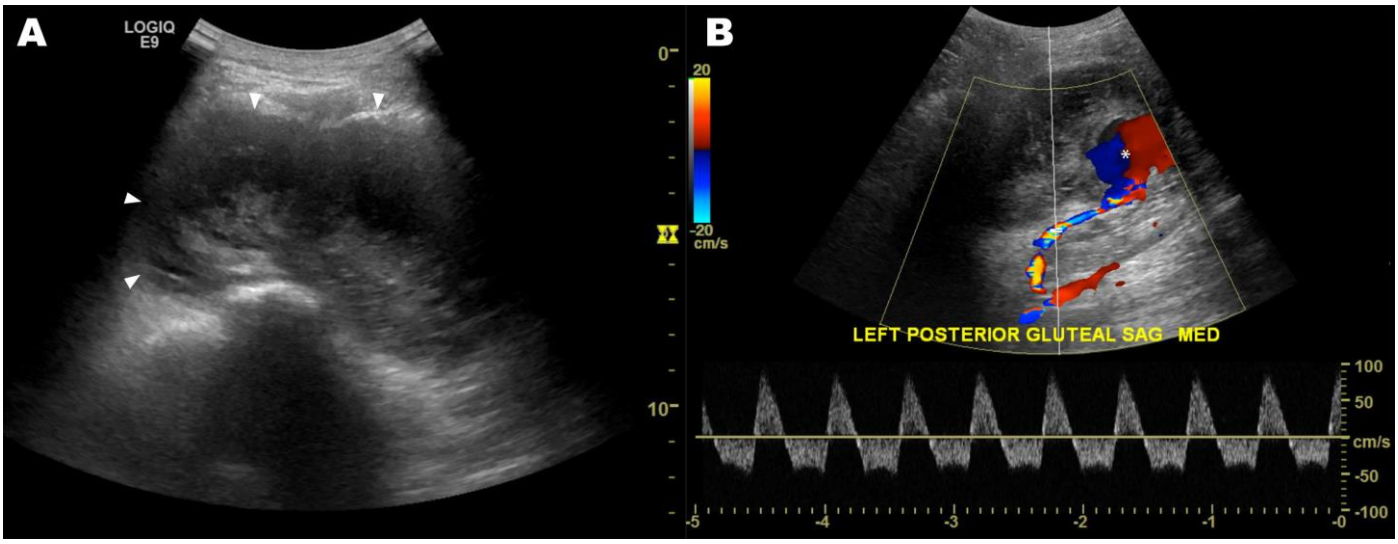

Figure 3: 28 year-old man with MHE with ruptured pseudoaneurysm secondary to large exostosis.
Findings: Grayscale images (A) through the proximal, medial thigh and inferior gluteal region demonstrate a large, heterogeneous, hypoechoic collection with multiple septations compatible with a hematoma (arrowheads). Color Doppler interrogation (B) shows a large pseudoaneurysm arising from a branch of the deep femoral artery with to-and-fro flow of blood - the classic 'yin-yang' sign (*).
Technique: Ultrasound: Grayscale - Curved, 4MHz; Color - Linear, 9MHz.

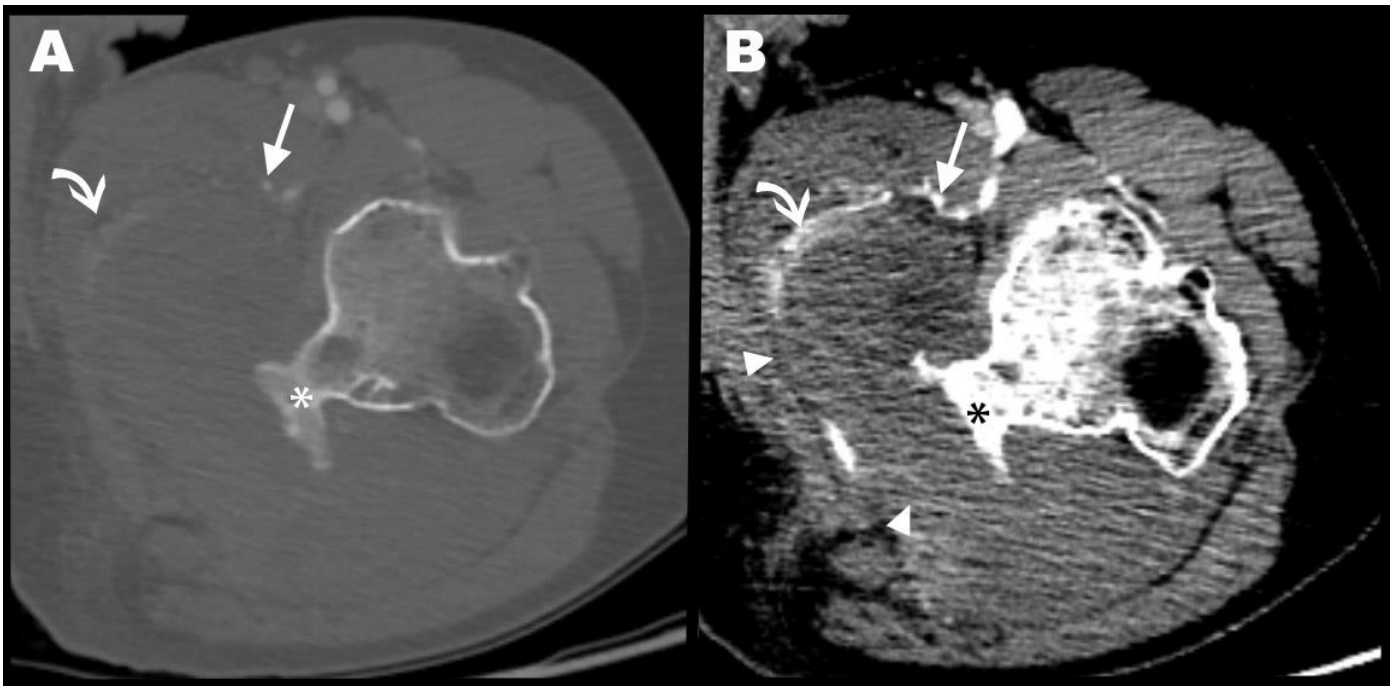

Figure 4: 28 year-old man with MHE with ruptured pseudoaneurysm secondary to large exostosis.
Findings: Contrast enhanced axial CT shown on bone (A) and soft tissues (B) windows demonstrates an exostosis of the lesser trochanter (*) with surrounding large, non-enhancing collection (arrowhead). The medial femoral circumflex artery (straight arrow) is displaced by the hematoma and was confirmed at time to surgery to be the source of pseudoaneurysm identified on ultrasound. No contrast-opacified pseudoaneurysm is identified which may be secondary to delayed filling. Note that the curvilinear density at the anteromedial margin of the hematoma represents calcifications, not extravasation (curved arrow).
Technique: Axial CT, 148 mAs, 120 kV, 1.25 mm slice thickness, 150 mL Omnipaque 350 intravenous contrast, angiographic phase.

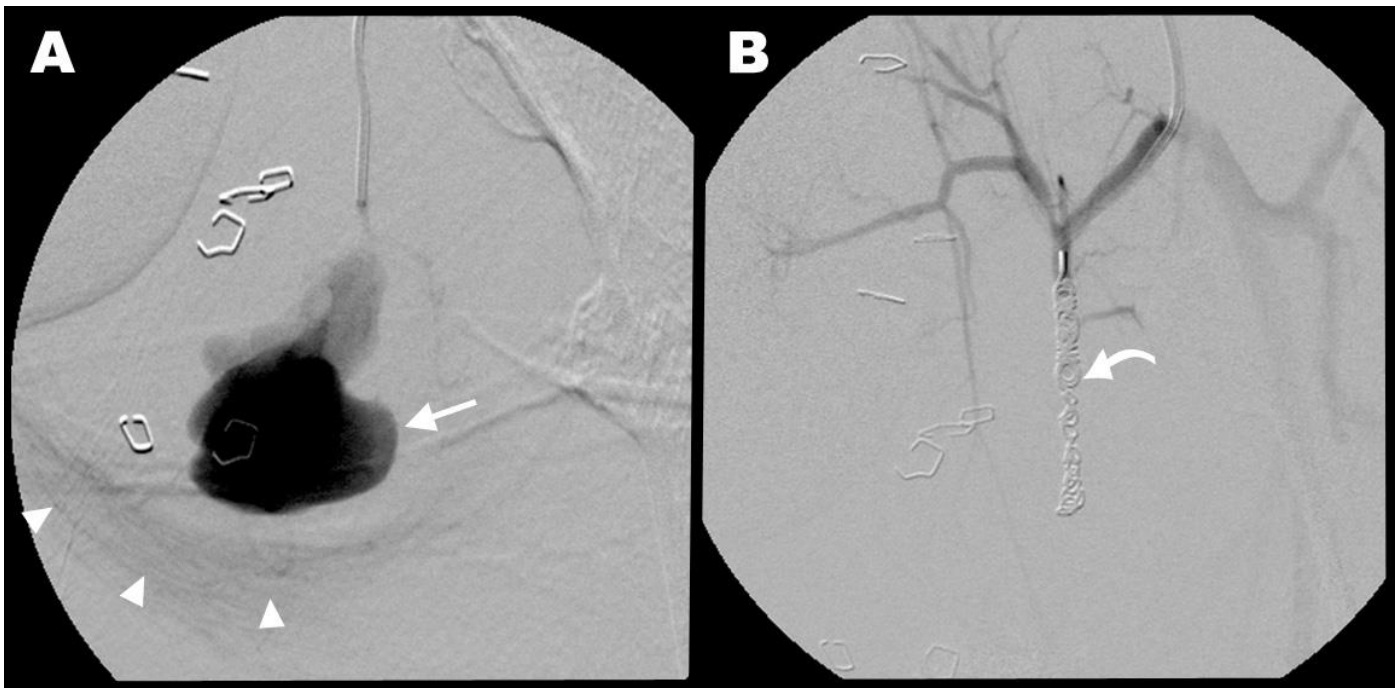

Figure 5: 28 year-old man with MHE with ruptured pseudoaneurysm secondary to large exostosis.
Findings: (A) Digital subtraction angiography demonstrates a pseudoaneurysm arising from the descending branch of the left medial circumflex artery (straight arrows). Subtle contrast blush is seen in the soft tissues inferiorly consistent with active extravasation (arrowheads). (B) Post-coil embolization (curved arrow) image shows no further opacification of the pseudoaneurysm and no evidence of extravasation.
Technique: Digital subtraction angiography of the proximal left lower extremity, Omnipaque 350 contrast.

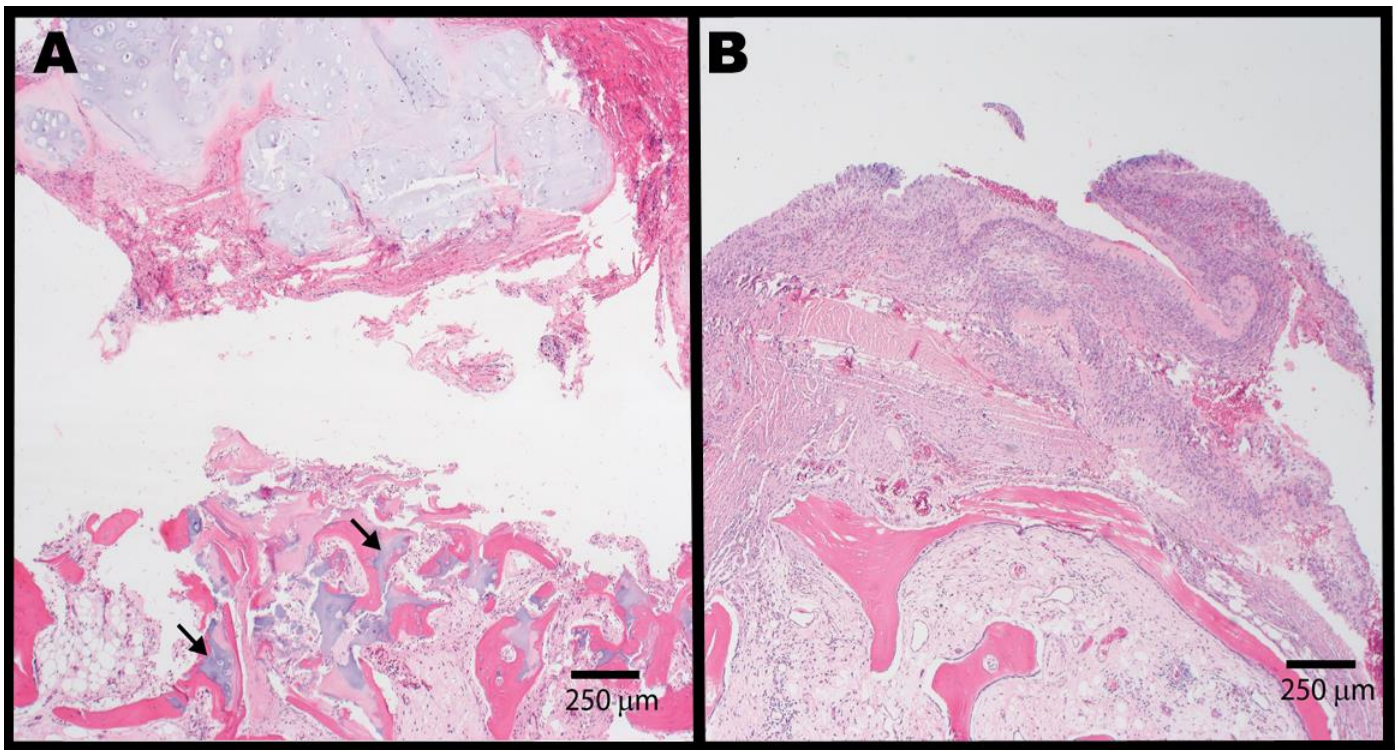

Figure 6: 28 year-old man with MHE with ruptured pseudoaneurysm secondary to large exostosis.
(A) The fragmented pathology specimen demonstrated focal hyaline cartilage with endochondral ossification (arrows) into lamellar bone. The findings were compatible with a fragmented osteochondroma. (B) A thin capsule with synovial metaplasia and fibrin likely represented the capsule of the hematoma.

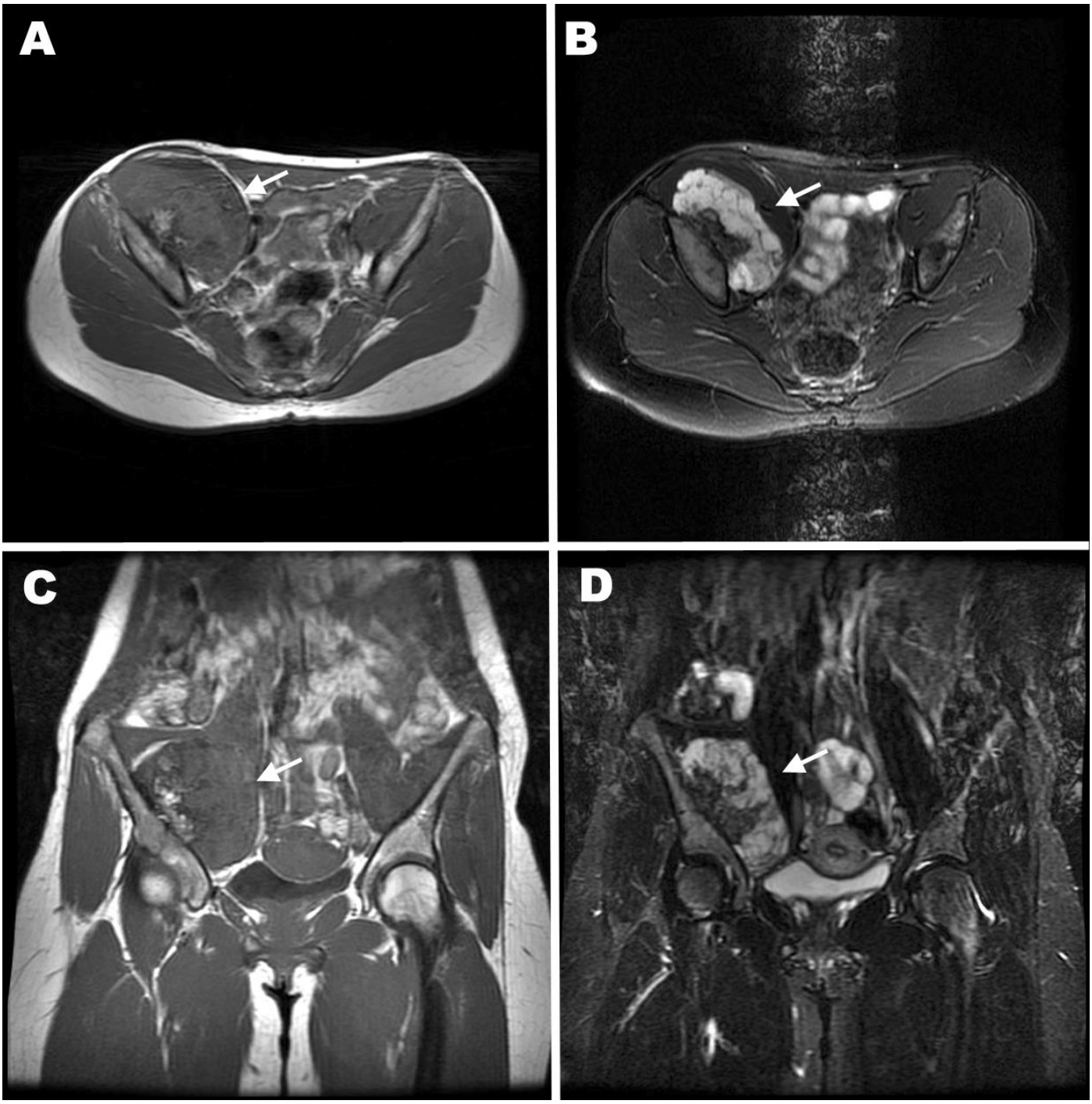

Figure 7: 26 year-old woman with chondrosarcoma, provided for comparison.
Findings: Axial T1 (A) , axial STIR (B), coronal T1 (C), and coronal STIR (D) MRI images through pelvis demonstrate a large mass (arrow) arising from the right ilium with local mass effect. A large, T2 hyperintense cartilaginous 'cap' is identified and measures greater than 2cm in thickness. This lesion was proven to be chondrosarcoma on biopsy.
Technique: MRI: 1.5T Axial T1 (TR: 783, TE: 10.2); 1.5 T Axial STIR (TR: 5154, TE: 83.2); 1.5T Coronal T1 (TR: 647, TE: 14.3); 1.5T Coronal STIR (TR: 5250, TE: 67.2)

	Osteochondroma	**Malignant degeneration to chondrosarcoma**	**Osteochondroma resulting in ruptured pseudoaneurysm**	**Bizarre parosteal osteochondromatous proliferation**
X-Ray	• Classically metaphyseal, projecting away from epiphysis. • Can be sessile or pedunculated. • Cartilage cap may be radiographically occult or thick with chondroid 'rings and arcs' calcification.	Destructive mass with cortical irregularity of underlying bone.	Large osteochondroma near a region of soft tissue mass, which may or may not be visible radiographically	• Pedunculated or sessile mineralized mass on a tubular bone surface, but without continuity of the marrow cavity. 70% in hands and feet, remainder in long bones
CT	• Similar to radiograph, with better demonstration of continuity of the medullary space. • Slight peripheral enhancement of the cartilage cap can be normal.	• Similar to radiography, better demonstrating cortical destruction. • Associated soft tissue mass typically hypodense to muscle and may have chondroid calcification.	• Contrast filling within a pseudoaneurysm arising off its parent vessel, although a thrombosed pseudoaneurysm may not be readily seen. • If the pseudoaneurysm has ruptured, surrounding mixed density hematoma with possible heterogeneous enhancement if subacute or chronic. • Adjacent osteochondroma may display cortical irregularity or may appear intact.	Discontinuity of the cortex and medullary space from the underlying bone.
MRI	• Hyperintense marrow on T1 surrounded by hypointense cortex on T1 and T2. • Cartilage cap is hypo- or iso-intense on T1 and hyperintense on T2.	• Chondroid matrix will be low signal on all sequences • Endosteal scalloping of the cortex • Thickening of cartilage cap > 1.5cm • Adjacent soft tissue mass	• T1 and T2 heterogeneous mass representing blood products of varying age, adjacent to an osteochondroma • Pseudoaneurysm may or may not be visible, possible manifesting as flow void on T2 or hyperintense on T1 if thrombosed	Variable appearance on T1, high signal on T2, with mild, heterogeneous enhancement and surrounding soft tissue edema
US	Cartilage cap will be well defined, hypoechoic, and seen between underlying bone and overlying fat and muscle	Thickened cartilaginous cap > 1.5cm	• Pseudoaneurysm will display the classic 'yin-yang' sign if patent, although this may be completely or partially absent if thrombosed. • Rupture will be demonstrated by a heterogeneous mass surrounding the pseudoaneurysm with displacement of nearby fat and muscle.	May not be well seen due to mineralization.

Table 1: Differential diagnosis table for ruptured pseudoaneurysm secondary to large exostosis.

Etiology	Repeated vessel abrasion by large or growing osteochondroma
Incidence	Pseudoaneurysm formation is the most frequent vascular complication of an osteochondroma, although the overall complication rate from osteochondroma is low
Gender Ratio	Male to female ratio of 1.5:1 for MHE. No published gender predilection for complications.
Age Predilection	Median age of diagnosis for MHE is 3, with nearly all cases diagnosed by age 12. No published age predilection for complications.
Risk Factors	Large or growing osteochondroma near a vessel
Treatment	Surgical bypass or endovascular coil embolization plus resection or debulking of causative osteochondroma
Prognosis	Excellent
Findings on Imaging	Large heterogeneous mass on CT and MRI representing hematoma adjacent to an osteochondroma, with or without active contrast extravasation. Doppler ultrasound may reveal classic 'yin-yang sign' of pseudoaneurysm as source of the hematoma.

Table 2: Summary table of ruptured pseudoaneurysm secondary to large exostosis.

ABBREVIATIONS

BPOP = bizarre parosteal osteochondromatous proliferation
CT = computed tomography
MHE = multiple hereditary exostoses
MRI = magnetic resonance imaging

KEYWORDS

multiple hereditary exostoses; multiple hereditary osteochondromas; complications; pseudoaneurysm; hematoma; metaphyseal tumor; CT; MRI; Ultrasound