

Case report: 13-year-old boy with progressive calf pain

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Case: anamnesis

- 13-year-old boy
- Calf pain
 - numeric rating scale: 9/10
 - slight radiating pain to rest of posterior leg, and little backpain
- Acute onset after fall with bike 6 weeks earlier
- Continuous stabbing, burning, tingling sensation in the calf
- Pain at night that forces to get out of bed
 - No B-symptoms
- Normal miction and defaecation – no saddle anesthesia
- No significant personal medical history

Case: clinical examination

Inspection:

- Normal posture

Lumbosacral spine:

- Normal mobility
- Increased calf pain with anteflexion and ipsilateral lateroflexion
- Increased calf pain with ipsilateral straight leg raise test

Hips/pelvis/SIG:

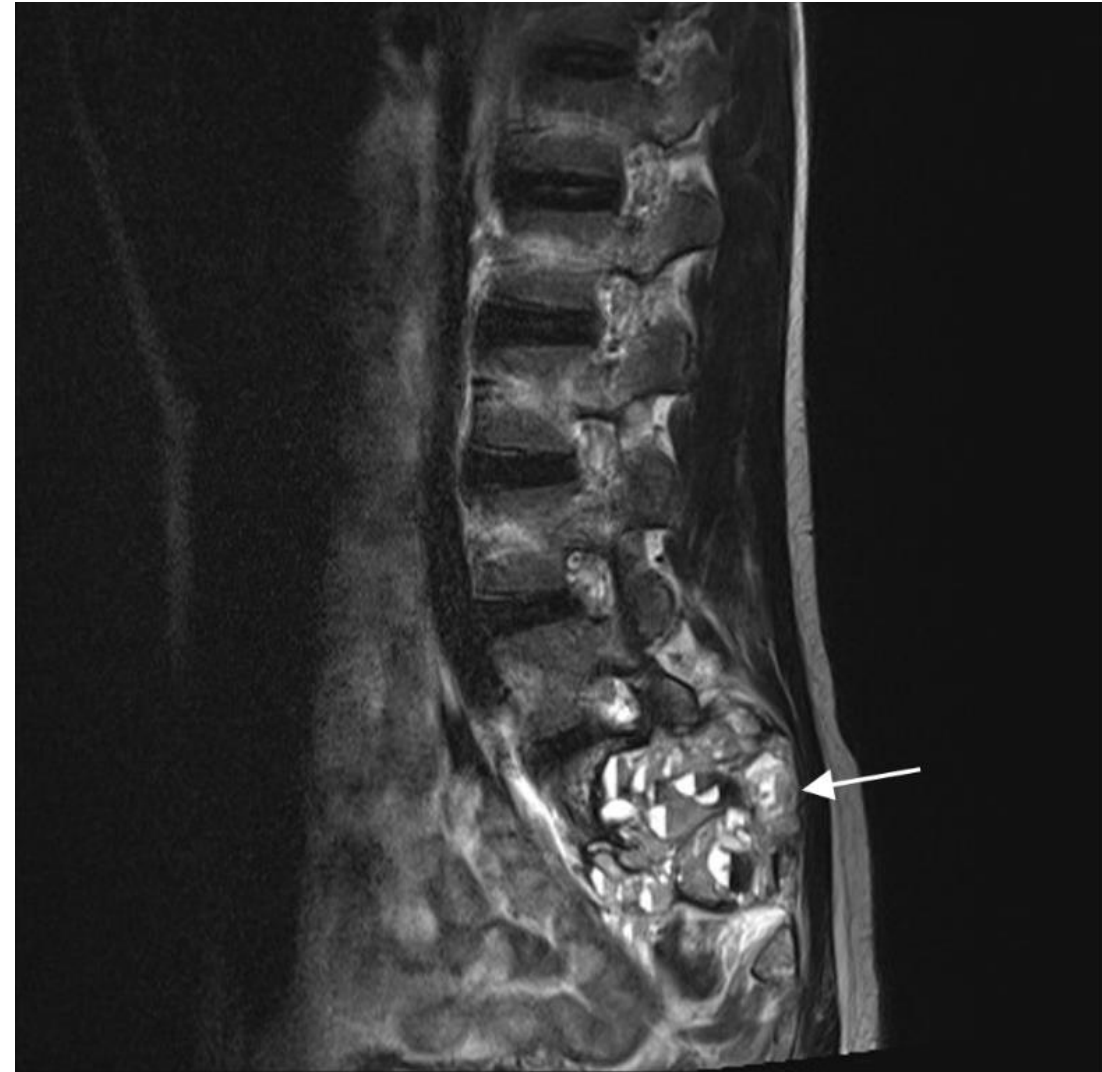
- Normal painless hip mobility
- Negative SIG-tests

Lower legs:

- Normal strength
- Burning and tingling with touch of posterior ipsilateral leg – further normal sensation for touch
- Absent ipsilateral achilles tendon reflex – further normal tendon reflexes

Case: further evaluation

- Suspicion of neurogenic problem, most probable radicular compression of radix S1 and/or S2
 - Several/clusters of red flags:
 - Young age of 13 years
 - Severe nocturnal pain
 - Continuous progressive pain with a non-mechanical component
 - Significant/high energetic trauma
 - Young age in combination with neurological deficits
- ➔MRI lumbosacral spine:
Lobulated bone lesion (6,2 cm x 5,7 cm x 5,4 cm) with fluid-fluid levels, expanding presacral and in the spinal canal with radix S1 and S2 not being clearly observably anymore
Preferred diagnosis: **aneurysmal bone cyst (ABC)**



Case: treatment and outcome

Treatment: surgical resection

- Preoperative: CT lumbosacral spine to assess bony anatomy
- Immediate preoperative: embolization of tumor to reduce bleeding
- Surgery: macroscopic complete piecemeal resection
+ osteosynthesis L5 to os ilium
- Peroperative cryosection:
result: difficult differential diagnosis between ABC and telangiectatic osteosarcoma
→ molecular investigation: FISH for translocation of USP6 gene was
positive, which confirms (primary) ABC

Outcome:

- 6 weeks after surgery: no pain
- Follow-up MR-scan 3 months after surgery: recidive ABC, albeit a lot smaller and no symptoms
- Follow-up MR-scan 6 and 12 months after surgery: unchanged
- 12 months after surgery: no pain or other complaints

Aneurysmal bone cyst: general

WHO definition:

*“destructive, expansile, benign neoplasm of bone composed of multiloculated blood-filled cystic spaces”*¹

Incidence: 0.14/100,000 persons/year²

Median age: 13 years; 53.3% in second decade²

Location: most frequent in long bones, but possible in all bones (rare: extraskelletal)^{3,4}

Etiology: Most cases primary; mainly because of upregulation of USP6 gene⁵⁻⁷
30% secondary to another tumor, benign or malign^{1,8,9}

Symptoms/consequences:^{1,3,9-11}

- Local pain and swelling
- Deformities limb and spine
- Nerve impingement → neurological symptoms
- Pathological fracture (in spine: vertebral compression fracture)
- Vertebral instability

Aneurysmal bone cyst: investigations

Imaging:

- X-ray: shows lesion, but often fails to make diagnosis ⁹
- MRI: best technique to complement X-ray ⁹
- CT: lower sensitivity than MRI ⁹
sometimes used preoperative to better define osseous borders ^{3,12}

Biopsy:

= necessary(!) as differential diagnosis contains benign and malign tumors ^{3,9}
especially important to distinguish from telangiectatic osteosarcoma ^{3,9}

Molecular pathology:

fluorescence in-situ hybridization (FISH): translocation in USP6 gene? ^{6,13-15}
↳ present in primary ABC

Aneurysmal bone cyst: treatment

- Treatment: ¹⁶
 - Stable asymptomatic lesions with low chance on destruction or impingement of surrounding tissue
 - possibility of conservative approach with observation
 - In other cases
 - treatment indicated
- No consensus in literature about optimal treatment
 - Curettage, with bone graft as needed, seen by many as standard treatment ^{3, 17}
 - Also dependent on location of ABC

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