A 53-year-old female with Stickler Syndrome and sacrococcygeal chordoma: a related entity?

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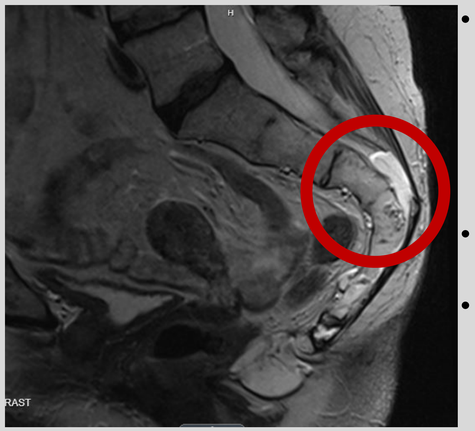
Background

- Stickler syndrome is a rare chondroplastic disorder which may present with hearing loss, retinal detachment, midfacial underdevelopment and cleft palate and is attributed to pathogenic collagen genes.
- While likely underreported, the incidence is postulated to be 1-3 cases per 10,000 births.[1]
- Chordomas are notochord remnants tumors with a documented incidence of 0.08 per 100,000 people.[2]
- These rare disorders are not known to share a common pathogenesis.

Case Description

- A 53-year-old female with Stickler syndrome and Pierre Robin features presented to a musculoskeletal physiatry clinic with left hip pain that limited ambulation.
- Family history is notable for a sister with Stickler Syndrome and history of chordoma.
- On exam, bilateral lower extremity strength 5/5, sensation grossly intact.
- She has scoliosis and severely restricted hip range of motion, mainly with flexion and internal rotation. She has pain before getting to 90° of hip flexion.
- Palpation revealed moderate tenderness over left gluteal muscles, negative over lumbrosacral area.
- Left hip logroll, FADIR, and FABER/Patrick's testing are also very painful.
- Negative thigh thrust bilaterally.
- FADIR on the right also reproduces some left groin pain.

Radiological Findings – T2 MRI



- Mass involving the sacrum and tissues anterior to the sacrum posterior to the rectum.
- Measures approx. 42 x 37 mm
- Extends superior as S3, to posterior aspects of S4 and S5 segments of the sacrum.

Clinical Course

- She was referred for MRI, which revealed severe left hip osteoarthritis and a sacral mass from the S3-S5 level.
- Biopsy revealed pathology consistent with chordoma. She underwent wide resection with removal of half of S3, and S4 and S5 spinal nerves.
- Her post-operative course was complicated by urinary retention and neuropathic and incisional pain with associated pelvic floor dysfunction.
- She was eventually referred for total hip arthroplasty.
- Pelvic floor physical therapy helped with her neurogenic bowel and bladder.
- Prior to THA patient with 48 total PT visits. Gradual and consistent improvement in transfers, gait, and right gluteal strength. Left hip pain and mobility limited due to osteoarthritis.
- Post THA patient completing physical therapy, pursuing return to work, and Women's Health PT

Discussion

- To our knowledge, these are the first two reported cases of chordoma in patients with Stickler syndrome.
- However, both the notochord and cartilage tissues express similar genes, which implies the possibility of overlapping pathophysiology. In embryologic development, the notochord expresses collagen genes to form a precursor matrix for the elements of the spinal cord.
- Notably, type X collagen will be replaced by bone during ossification.
- Aberrant gene expression in the notochord due to collagen gene mutations may lead to inappropriate notochord development and predispose patients to chordoma development.

Conclusion

- Stickler syndrome patients presenting with musculoskeletal complaints should be thoroughly evaluated for dysfunction related to abnormal collagen deposition.
- In patients with Stickler Syndrome, physiatrists should consider the possibility of development of a chordoma and screen for neurologic signs and symptoms.

1. Nowak CB. Genetics and hearing loss: a review of Stickler syndrome. *J Commun Disord.* 1998;31(5):437-454
2. Walcott BP, Nahed BV, Mohyeldin A, Coumans JV, Kahle KT, Ferreira MJ. Chordoma: current concepts, management, and future directions. *Lancet Oncol.* 2012;13(2):e69-e76