Bertolotti syndrome: a not-to-miss cause of chronic low back pain in young adults

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ABSTRACT

Low back pain (LBP) in young adults is a common condition that needs to be appropriately examined in cases being refractory to classic treatment strategies. We present two cases of chronic LBP with challenging diagnosis and treatment refractoriness. The first case corresponds to a young lady that had been treated mistakenly with an anti-tumor necrosis factor because her treating doctors diagnosed unilateral sacroiliitis which turned out to be a magnetic resonance imaging (MRI) artifact (partial volume artifact). The second case is about another young lady with chronic LBP that did not respond to the classic treatment with non-steroidal anti-inflammatory drugs. Both cases have been diagnosed as having Bertolotti syndrome. Bertolotti syndrome is an anatomical abnormality consisting of partial unilateral or bilateral fusion of the transverse process of the lowest lumbar vertebrae to the sacrum. The presentation of both cases highlights the importance of a minute history taking and clinical examination especially in young patients with chronic LBP.

Keywords: Transverse process; L5 sacralisation; Spine anatomical abnormality; chronic low back pain; Bertolotti syndrome.

INTRODUCTION

Low back pain (LBP) in young adults is a common condition1. A global review of the prevalence of LBP in the adult general population showed a point prevalence of 11.9%, a one-month prevalence of 23.2%, and one-year prevalence of 38%. The lifetime prevalence was 39.9%2. It may be acute or chronic. Chronic LBP is not always related to an inflammatory process and other possible causes must be excluded. Among those, are anatomical abnormalities of the lumbar spine which are often discovered through radiographs or computed tomography (CT) scans3. In refractory cases of LBP that do not respond to common treatments, a rigorous clinical examination is an imperative.

CASE 1

A 23-year-old female was referred to the Rheumatology department with a history of mechanical LBP lasting for about 8 months after seeing an orthopedic doctor and a rheumatologist. She had a CT scan of the lumbar spine showing a mild stenosis at the L5-S1 level as well as mild degenerative changes. She was treated with naproxen 500 mg twice daily, as well as other non-steroidal anti-inflammatory drugs (NSAIDs) with no improvement. Due to the persistence of the symptomatology, she had a magnetic resonance imaging (MRI) of the pelvis. The findings were described as having unilateral sacroiliitis and she was treated with infliximab (INF) 5 mg/kg at weeks 0-2 - 6 and 8 weeks thereafter. She received 6 infusions of INF in total, but without any obvious benefit.

Past medical and family history were unremarkable. She denied psoriasis, oral ulcers, diarrhea, urethritis and conjunctivitis. She had no features of inflammatory back pain such as morning stiffness, pain at rest or during the night. On physical examination, she showed no signs of distress or discomfort except on walking. The lumbar pain was reproduced while extending the left thigh and was aggravated on leaning her body forward. Chest expansion was normal, while Schober test was difficult to measure due to the eliciting pain on leaning forward. Gaenslen’s test was negative while the Faber test was positive on the left. She had no signs of enthesitis or peripheral arthritis nor extra-articular manifestations of spondyloarthritis (SpA). Routine laboratory tests including acute phase reactants were within normal limits, while the human leucocyte antigen (HLA)-B27 was negative. Reviewing the MRI scan, we observed a high
T2-weighted signal with fat suppression only in one coronal plane image, which was not found on the transverse plane (Figure 1). This is a partial volume artifact related to a high signal of the projected iliac vessels. For this reason and in order to avoid this artifact and a misinterpretation of the acquired data, a perpendicular positive plane is always required.

After this, we also reviewed the CT of the lumbar spine. We observed a significant stenosis at L5-S1 level and a large transverse process on the left side. Mul-

**Figure 1.** MRI of the pelvis. Coronal view. Partial volume artifact. A high T2-weighted with fat suppression signal is visible mimicking bone oedema (white arrow - not visible in other planes).

**Figure 2.** Significant stenosis at L5-S1 level and a large transverse process on the left side (Fig. 2a). CT scan and multiple projection reconstruction revealed sacralisation of the L5, especially of the left transverse process which was bridged with the sacrum (Fig. 2b).
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Multiple projection reconstruction (MPR) revealed sacralization of the L5 especially of the left transverse process which was bridged with the sacrum (Figure 2). Thus, the diagnosis was stenosis and partial left sacralization of the L5 vertebrae, a condition known as Bertolotti syndrome. INF was discontinued and she had 6 courses of physiotherapy and received analgesics with significant improvement.

CASE 2

A 37-year-old female presented to the Rheumatology outpatients’ clinic due to chronic LBP. The patient has been treated by several orthopedists with several courses of different NSAIDs with no significant improvement on most of the times. Due to the treatment failure, she has been advised to be examined by a Rheumatologist and exclude any inflammatory causes. As in the previous case, she had no inflammatory features of LBP and there was no family history nor other clinical or laboratory features suggestive of SpA. In addition, the character of the pain was mechanical rather than inflammatory. The x-ray of the lumbar spine revealed sacralization of the L5 vertebral body and the left transverse process, compatible with the diagnosis of Bertolotti syndrome (Figure 3).

DISCUSSION

Bertolotti syndrome is an anatomical abnormality consisting of partial unilateral or bilateral fusion of the transverse process of the lowest lumbar vertebrae to the sacrum. It is a frequent cause of LBP in young patients affecting 4-8% of the general population but it can easily be missed if the clinician is not able to recognize this pathology. Diagnosis is based on appropriate patient history, a careful clinical examination and the application of the correct imaging modality. Patients with LBP must be examined and investigated carefully to avoid wrong diagnosis and treatment.

The presentation of both cases highlights the importance of a thorough history taking and clinical examination especially in young patients with chronic LBP. In addition, the appropriate imaging modality in the appropriate clinical context must be ordered and interpreted by an experienced radiologist.

REFERENCES