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A CASE REPORT OF BERTOLOTTI SYNDROME INCIDENTALLY COEXISTING IN A 12 YEAR MALE WITH HISTORY OF PERINEAL BONE TRAUMA.

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ABSTRACT

Introduction: **Bertolotti's Syndrome (BS) is a rare cause of low back pain in adolescents, often associated with lumbosacral transitional vertebrae (LSTV). This condition results in structural changes at the lumbosacral junction, leading to pain through altered biomechanics and pseudoarthrosis. Due to its rarity and overlapping symptoms with other lumbar pathologies, BS is frequently undiagnosed.**

Case Presentation: **This case report discusses a 12-year-old male patient that developed BS following trauma to the perineal region, presenting with low back pain and difficulty walking. Imaging studies revealed an enlarged left transverse process of the L5 vertebra articulating with the sacrum, consistent with Type II LSTV according to the Castellvi classification.**

Conclusion: **This case highlights the importance of considering BS in the differential diagnosis of low back pain in young patients, particularly in the context of trauma. While conservative management can offer significant relief, surgical intervention may be necessary in refractory cases. Early diagnosis and tailored treatment are crucial for optimal outcomes, and further research is needed to establish standardized treatment guidelines and improve patient care.**

Keywords: Bertolotti Syndrome, Lumbosacral transitional vertebra, Lower back pain, Conservative management, Corticosteroid injection.

INTRODUCTION

Bertolotti's Syndrome (BS) is a rare cause of lower back pain, especially in younger patients[1,2], and is defined by the presence of lumbosacral transitional vertebrae (LSTV) that lead to structural and biomechanical changes at the lumbosacral junction[1,3]. The syndrome was first described by Mario Bertolotti in 1917, and it remains a topic of debate in terms of its clinical significance and management. The classification of LSTV includes four types based on morphologic characteristics, with BS often associated with Type II to IV configurations, while Type I is generally considered asymptomatic[4]. In this report, we present a case of BS in a 12-year-old patient, detailing the clinical presentation, imaging findings, and the outcomes following conservative management.

Bertolotti Syndrome should be considered in young patients with unexplained back pain. Conservative management can offer significant symptomatic relief, but surgical intervention may be necessary in refractory cases. Early diagnosis and tailored treatment are essential for optimal outcomes. Further research is necessary to establish standardized treatment guidelines and optimize patient outcomes.

Case Profile

The patient is a 12-year-old male who presented with a history of trauma to the perineal region sustained during martial arts training. He reported abdominal swelling and haematuria (bloody urine) post-injury. He also had a prior history of intermittent low back pain, which was initially mild and did not require medical attention.This was however aggravated after the injury to the perineum. On evaluation, the patient was found to have stable vital signs, including a heart rate of 80 beats per minute and blood pressure of 110/80 mmHg. Physical examination revealed redness and swelling in the perineal region, with difficulty walking, though lower limb reflexes, sensations, and power were intact.

CT urography was performed (axial images with coronal reformatted images and volume rendering) which revealed markedly distended urinary bladder injury (with hyperdense collection noted within the bladder which is suggestive of hematoma collection). There was associated aggravation of the lower back pain (which preceded the trauma and was made worse by the trauma) after the perineal injury and thus

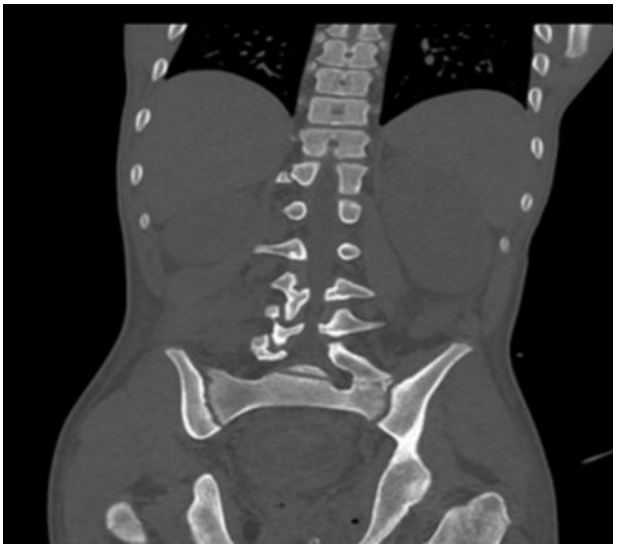

Figure 1: Coronal CT scan of the Lumbosacral Region (*displays an enlarged left transverse process of the L5 vertebral body, which articulates with the sacral ala, indicating a characteristic finding of Bertolotti Syndrome*).

necessitated further evaluation of the bone window images for the CT urography.

This revealed an enlarged left transverse process of the L5 vertebral body articulating with the sacral vertebra (Figures 1 and 2). This is consistent with a Type II LSTV according to the Castellvi classification, where a pseudoarthrosis is formed between the transverse process of the L5 and sacrum. The urinary bladder was catheterised and the hematoma resolved within 3 weeks.

Figure 2: 3D Reconstruction of the Lumbosacral Region (*shows the anatomical relationships of the lumbosacral spine and pelvis, highlighting the prominent transverse process of the L5 vertebra and its articulation with the sacrum. It helps visualize the extent of abnormal articulation and any related degenerative changes).*

Following the review of the CT scan and diagnosis of Bertolotti syndrome was made, conservative management was initiated. A local anaesthetic and corticosteroid injection was administered into the pseudoarthrosis at the articulation site between the L5 transverse process and the sacrum. This was done 6 times at 3 months interval in other to help with reduction in the pain. After an 18-month follow-up, the patient reported improvement of his symptoms, though complete resolution was not achieved. Thus further follow up and physiotherapy were recommended and there was significant improvement noted on further visits after about 6 months.

DISCUSSION

Lumbosacral transitional vertebrae are present in approximately 25% of the general population, with Bertolotti's Syndrome accounting for 4.6% of all cases, and having a higher incidence in the under-30 age group[2], where the prevalence can reach up to 11.4%[2,3,5,6]. LSTV can manifest as either a lumbarized S1 segment or a sacralized L5 segment. Castellvi et al. (1984)[1,4,7] classified LSTVs into four typees which include type I with dysplastic transverse process which can be unilateral or bilateral. The type II LSTV has incomolete lumbarisation or sacralisation with a diarthrodial joint between the transverse process and sacrum. Complete lumbarisation or sacralisation with complete osseous fusion of the transverese process and sacrum is seen in type III. Type IV LSTV has mixed configuration with unilateral type II and a contralateral type III TRANSITION.

Type I LSTVs are generally asymptomatic, while Types II to IV have been implicated in BS due to mechanical stress and altered biomechanics, leading to lower back pain, disc degeneration, and even nerve root compression[4,5].

In BS, the sources of pain are multifactorial and include pseudoarthrosis, facet joint arthrosis, asymmetric biomechanics, and discogenic pain. Degenerative changes are often noted at the disc level above the LSTV, rather than at the transitional segment itself. The transitional joint is not usually the source of nerve root symptoms but may contribute to foraminal narrowing and increased stress on adjacent structures. There is associated degenerative changes also noted with exit foramina narrowing (with compression on the exiting nerve roots), disc herniations and strain of the psoas and quadrate lumborum muscle[6,8].

Our patient presented with Type II LSTV, as evidenced by the articulation of the enlarged left transverse process of L5 with the sacrum, forming a pseudoarthrosis. After conservative treatment using

local anaesthetic and corticosteroid injections into the pseudoarthrosis, the patient reported symptomatic improvement at the 18-month follow-up, although there was no complete resolution. This aligns with findings from Sumarriva *et al.* (2022)[9] and Santavirta *et al.*[7], who noted that resection of the transverse process was beneficial in cases where conservative treatment failed. However, in the presence of disc pathology, resection is preferred, while posterolateral fusion is indicated if discogenic pain is identified[2,6,9].

Management and treatment options

The patient has conservative management which included an initial anaesthetic with corticosteroid in the first 18months in which there was improvement. There was near resolution 6 months after in which treatment was coupled with physiotherapy.

Conservative treatment remains the first-line approach for BS, including physical therapy, activity modification, and pharmacologic measures like non-steroidal anti-inflammatory drugs (NSAIDs), corticosteroids, and muscle relaxants.Consrevative Injection therapies using corticosteroids and local anaesthetics directly into the pseudoarthrosis have shown efficacy in both diagnosis and management, as noted in our case[5,7,10].

If conservative measures fail, surgical intervention is considered. Surgical options include resection of the transverse process or posterolateral fusion, depending on the presence and severity of disc pathology. Santavirta et al[7] reported a 60% success rate with surgical resection of the transverse process. Newer interventions, including radiofrequency ablation (RFA) of the pseudoarthrosis, have shown promise, offering sustained pain relief in some patients. From recent studies, bipolar RFA and pulsed RFA have been successfully applied in cases of persistent pain, showing promises as a minimally invasive alternative [2,3,9,11].

CONCLUSION

Bertolotti's Syndrome is an uncommon but significant cause of lower back pain in young patients. Though Bertolotti syndrome is commoner in the age group 30 to 50 years, it should be considered as a cause of low back pain in the younger age group. Thus a high index of suspicion is required for diagnosis, especially in the presence of characteristic imaging findings of LSTV. While conservative treatment is effective in many cases, surgical intervention may be necessary for refractory pain. Continued research into advanced imaging and minimally invasive techniques is necessary to optimize management and improve patient outcomes.

Further research is needed to establish standardized treatment guidelines and to better understand the pathophysiology of Bertolotti Syndrome, as there is currently no consensus on the optimal management approach.

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