

Bertolotti's syndrome: An Underdiagnosed Cause of Lower Back Pain; A Case Report of Two Cases and Current Management Options

ABSTRACT

Bertolotti's syndrome is a spinal disorder characterized by lumbosacral transitional vertebra (LSTV) in which an abnormal enlargement of the transverse process of the fifth lumbar vertebra and is associated with lower back pain. It is a syndrome that is not an uncommon cause of back pain in young adults and may be a source of chronic pain in 10% of lower back pain patients younger than 30 years. In Bertolotti's syndrome, the fifth lumbar vertebra transverse process may be enlarged either unilaterally or bilaterally and may articulate or fuse with the sacrum or ilium. It is an anatomical variant of sacralization of fifth lumbar vertebra. This causes alterations in the biomechanical characteristics of the lumbar spine resulting from asymmetrical motion between the LSTV and the sacrum. It is often a factor that is not addressed in the evaluation and treatment of lower back pain. The syndrome affects 4% to 8% of the population. It can cause pain due to involvement of various structures: lumbosacral neo-articulation, contralateral facet arthrosis, sciatica, discogenic, or sacroiliac pain. It is characterized by low back pain, with a normal physical examination. Bertolotti's syndrome is frequently underdiagnosed and should be considered as a differential diagnosis for lower back pain. Therefore, its pathophysiology, epidemiology and treatment must be a topic of general knowledge to physicians that often treat this condition.

Key Words: Bertolotti syndrome, lumbar transitional vertebra, lower back pain, sacralization

1. INTRODUCTION

Bertolotti's syndrome is characterised by lumbosacral transitional vertebra (LSTV) in which the fifth lumbar vertebra transverse process may be enlarged either unilaterally or bilaterally and may articulate or fuse with the sacrum or ilium and producing a chronic, persistent low back pain [1]. It is an anatomical variant of sacralization of fifth lumbar vertebra. In 1917, Mario Bertolotti stated that these abnormal vertebrae may produce low back pain due to arthritic changes occurring at the site of pseudarthrosis [2]. The overall incidence of Bertolotti's syndrome has been reported to be between 4% to 8% in patients with low back pain and 10% in patients presenting with back and leg pain under 30 years of age [3]. It is an important cause of low back pain in young patient. The biomechanics of LSTV is attributed to an alteration or reduction of movement between the transitional vertebra and the sacrum that can ultimately lead to pain from stress in the facet joint and/or is exacerbated by disc degeneration [4]. Currently there is no agreement as to the best method of treatment for Bertolotti's syndrome patients. Low back pain in Bertolotti's syndrome has been addressed by various methodologies but there is no consensus regarding definitive management.

2. PRESENTATION OF CASE

Case 1: A 23 years old otherwise well and healthy Malay lady who works as a clerk presented to us with lower back pain for 1 year. It was a dull aching on and off localised pain which was aggravated by prolonged standing and relieved upon rest. There was no weakness or numbness of bilateral lower limbs. No history of antecedent trauma or fall and no bowel or bladder dysfunction. There was no history of prolonged cough; loss of appetite; loss of weight or tuberculosis contact. On examination, there was no midline spinal tenderness. There was mild tenderness over right lower paraspinal muscles. Neurology of bilateral lower limbs was normal. Straight leg raising test over bilateral lower limbs was normal. Inflammatory markers were within normal limits. Lumbosacral x ray showed abnormal articulation between the L5 transverse process and the medial aspect of the right ilium consistent with Bertolotti's syndrome as shown in Figure 1. We admitted and started patient on analgesics like celecoxib and methylcobalt and referred physiotherapist for back strengthening exercises. She was discharged on second day of admission and upon review in clinic after 1 month, 3 month and 6 months and currently patient is otherwise well and pain control is adequate and she is able to do daily activities and work without much difficulties.

Case 2: A 43 years old otherwise well and healthy Malay gentleman who works as a gardener presented to us at our outpatient clinic with lower back pain for 5 months which worsened in the past 2 weeks. It was a dull aching on and off localised pain which was aggravated by strenuous activities and relieved upon rest. There was no weakness or numbness of bilateral lower limbs. No history of antecedent trauma or fall and no bowel or bladder dysfunction. There was no history of prolonged cough; loss of appetite; loss of weight or tuberculosis contact. On examination, there was no midline spinal tenderness. There was mild tenderness over left lower paraspinal muscles. Neurology of bilateral lower limbs was normal. Straight leg raising test over bilateral lower limbs was normal. Inflammatory markers were within normal limits. Lumbosacral x ray showed abnormal articulation between the L5 transverse process and the medial aspect of the left ilium consistent with Bertolotti's syndrome as shown in Figure 2a and 2b. We started patient on analgesics like tramadol and paracetamol and referred physiotherapist for back strengthening exercises. He was reviewed in clinic after 3 month and 6 months and currently patient is otherwise well and pain control is adequate and he is able to do daily activities and work without much difficulties.



Figure 1 (Case 1): Lumbosacral X ray shows abnormal articulation between the L5 transverse process and the medial aspect of the right ilium consistent with Bertolotti's syndrome (labelled by arrow).

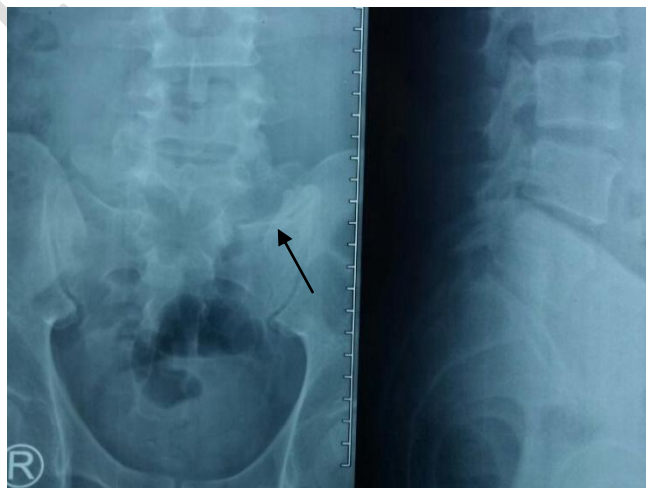


Figure 2a and 2b (Case 2): Lumbosacral X ray shows abnormal articulation between the L5 transverse process and the medial aspect of the left ilium consistent with Bertolotti's syndrome (labelled by arrow).

3. DISCUSSION

Low back pain in the presence of an LSTV was originally noted by Mario Bertolotti in 1917 and termed "Bertolotti's Syndrome". Low back pain is a prevalent problem with multiple causes. Approximately 80% of adults seek a physician's help to deal with their low back pain at some point in their lives [5]. Therefore, it is crucial that patients with Bertolotti's syndrome be accurately diagnosed and treated. According to Castellvi's classification as shown on Figure 3, there are four types of lumbosacral transitional vertebrae on the basis of morphologic characteristics. Type I includes unilateral (Ia) or bilateral (Ib) dysplastic transverse processes, measuring at least 19 mm in width (craniocaudal dimension). Type II exhibits incomplete unilateral (IIa) or bilateral (IIb) lumbarization/sacralization with an enlarged transverse process that has a diarthrodial joint between itself and the sacrum. Type III LSTV describes unilateral (IIIa) or bilateral (IIIb) lumbarization/sacralization with complete osseous fusion of the transverse process(es) to the sacrum. Type IV involves a unilateral type II transition with a type III on the contralateral side [6]. Literature indicates that Type II and IV have the most positive association with lower back pain [7].

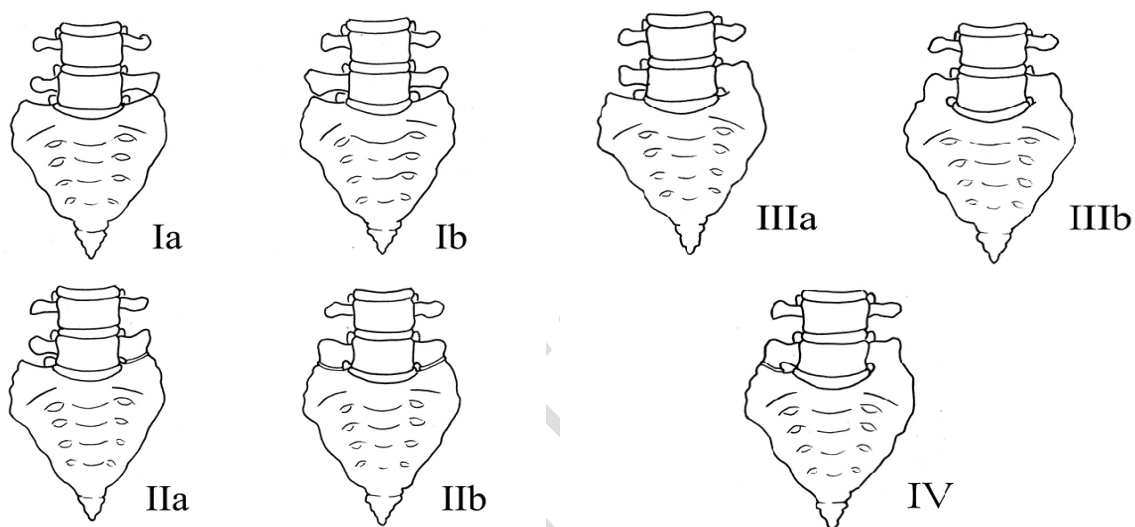


Figure 3: Illustration demonstrating the Castellvi classification of LSTVs.

Unilateral LSTV is more common than bilateral LSTV (9% vs. 3%) [8]. Men are more likely to be affected than women (28.1% vs. 11.1%) [9]. Patients with LSTV usually present to clinic with lower back pain after second decade of life [10]. According to Quinlan et al, the overall incidence of Bertolotti's syndrome has been reported to be between 4% to 8% in patients with low back pain and 10% in patients presenting with back and leg pain under 30 years of age [3]. Throughout the literature, the prevalence of LSTV in patients seeking care for low back pain ranges from 4.6%-35.6% [11]. Due to its wide prevalence, Quinlan et al. encourages physicians to consider Bertolotti's syndrome in the differential diagnosis for low back pain, especially in younger patients [3].

Various aetiologies for pain in symptomatic cases of Bertolotti's syndrome have been postulated. Aihara et al. [12], Luoma et al. [13], and Brown et al. [14] have all published MRI-based studies demonstrating that the disc above a transitional vertebra appears to be at increased risk of undergoing degenerative changes, whereas the disc below appears to be protected. The transverse process may be completely fused to the sacrum or ilium, or a pseudoarticulation may exist. This pseudoarticulation may undergoes degenerative changes and be the source of pain in these patients. Alternatively, the fused transitional vertebrae may result in instability above the level of the fusion. Elster et al. found that the incidence of degenerative disc disease and spinal stenosis was nearly nine times higher in the level adjacent to the transitional vertebrae. Cases are often associated with transitional vertebrae and there may be six lumbar vertebrae. These changes are associated with disc degeneration or instability, suggesting that there may be abnormal biomechanical stress above the fusion [15].

118
119 The diagnosis of Bertolotti's syndrome is based on lumbar spine radiographs showing a transverse
120 mega-apophysis. Cross-sectional imaging (bone scan) can support diagnostic suspicion by specifying
121 the morphological anomaly and highlighting the neoarticulation impingement (functional imaging: MRI
122 and bone scintigraphy) [16]. A diagnostic test that infiltrates the neoarticulation with anaesthetic and
123 anti-inflammatories is used to assess the possible relationship between the painful symptomatology
124 and the morphological anomaly (neo-articulation). If the test is positive, the pathology is almost
125 certainly Bertolotti's syndrome: i.e., pain related to a neo-articulation. The test is both diagnostic and
126 therapeutic because it also serves as the medical treatment for this syndrome [7].

127
128 Management of chronic low back pain associated with Bertolotti's syndrome should be initiated with a
129 comprehensive conservative approach consisting of activity modification, physical therapy and home
130 exercise, psychosocial support, and medications such as NSAIDs, muscle relaxants, anticonvulsants,
131 and antidepressants. Conservative nonsurgical management includes local injection of anaesthetic
132 and corticosteroids within the pseudoarticulation or contralateral facet joint, radiofrequency ablation
133 and surgical management with partial transverse process resection, and/or posterior spinal fusion
134 [17]. Direct local anaesthetic and steroid injection or surgical resection of the anomalous or
135 contralateral facet joint has produced successful relief of pain and can yield valuable diagnostic
136 information [18-21].

137
138 Interventional approaches can be beneficial in cases refractory to conventional conservative
139 management. For those who presented with lumbosacral radiculopathy or radiculitis symptoms due to
140 the direct compression or irritation of the nerve root by an enlarged transverse process or
141 pseudoarticulation, a transforaminal or interlaminar epidural steroid injection maybe considered. In a
142 study done by Zhang et al, a diagnostic lumbar facet medial branch block can be performed for those
143 with clinical evidence of lumbar facet pain due to the reduced motion between the LSTV and the
144 sacrum. Radiofrequency neurotomy of the target medial branches may provide longer-term pain relief
145 if the patient has greater than 50% pain reduction from the diagnostic medial branch block. For those
146 suspected of sacroiliac dysfunction due to increased load to the sacroiliac joint with restricted motion
147 between the LSTV and the sacrum, a diagnostic sacroiliac joint injection maybe attempted. If the
148 patient has greater than 50% pain reduction, radiofrequency neurotomy of the L5 dorsal ramus, S1
149 and S2 lateral branches may provide longer-term pain relief [21]. In a case report, Burnham described
150 a successful novel radiofrequency technique for treating symptomatic lumbosacral junction
151 pseudarthrosis in which he fluoroscopically guided local anesthetic/ corticosteroid injection into the
152 pseudarthrosis circumferentially around the posterior pseudarthrosis articular margin. Accordingly,
153 bipolar radiofrequency strip thermal lesions were created at the same locations. Complete pain relief
154 and full restoration of function was achieved for 16 months post procedure [22]. Almeida et also
155 reported that radio-frequency denervation is another possible treatment option and provided
156 temporary relief of pain due to an anomalous articulation [9].

157
158 Operative treatment is suggested in selected patients. For example, resection of the transverse
159 process may be beneficial for those who demonstrate pain truly emanating from a transitional joint
160 and failed conservative treatment. If the pain source is from a degenerated disc above a transitional
161 level, posterior fusion is an option as well [1]. In a case report, Brault et al described successful
162 treatment of contralateral facetogenic pain by resection of the ipsilateral anomalous articulation [18].
163 Jonsson et al reported relief of pain in 9 of 11 patients following surgical resection of a unilateral LSTV
164 pseudoarticulation [23]. Ugokwe et al and Almeida et al similarly describe successful treatment after
165 surgical resection [9,20]. In a case series of 8 patients who underwent surgical resection of the
166 unilateral anomalous articulation and 8 patients who underwent posterolateral fusion of the LSTV,
167 Santavirta et al reported improvement in pain in 10 of 16 patients at 9-year follow-up without a
168 difference between the fusion or resection groups [1]. In a case report, Yousif et al performed a
169 minimally invasive tubular resection of a transitional lumbosacral pseudojoint and the short-term
170 outcomes were favourable with the patient discharged, pain free 36 hours postoperatively, with return
171 to normal work duties in two weeks and remained pain free six weeks postoperatively. The use of a
172 minimally invasive tubular approach was key in minimizing unnecessary tissue dissection via a small
173 35 mm incision. It allowed mobilisation over an obstructive osseous ridge and meant that bony
174 resection was limited only to the pathological joint, minimizing the risk of future back pain [24]. Li et al
175 also performed minimally invasive tubular resection of the anomalous transverse process in 7 patients
176 with Bertolotti's syndrome. Three (43%) of 7 patients reported complete resolution of low-back pain, 2
177 (29%) of 7 patients had reduced low-back pain, and 2 patients (29%) experienced initial relief but

178 return of low-back pain at 1 and 4 years postoperatively. Three (50%) of the 6 patients with radicular
179 pain had complete relief of this symptom. They suggest that minimally invasive resection of the LSTV
180 be used in select patients in whom conservative therapy has failed and who have pain attributed to
181 the LSTV [25].
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183

184 **4. CONCLUSION**

185
186 Bertolotti's syndrome is frequently underdiagnosed and should be considered as a differential
187 diagnosis for lower back pain in young patients after ruling out all other causes of back pain. It is often
188 a factor that is not addressed in the evaluation and treatment of lower back pain. In light of the
189 diversity of clinical presentations, management of patients with low back pain associated with
190 Bertolotti's syndrome has to be individualized. Multimodal care is often required for optimal patient
191 outcome. The treatment, whether conservative or operative, is still debated. Understanding the
192 biomechanical and pathophysiological mechanisms underlying the chronic low back pain is the key to
193 choosing the appropriate procedures.
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195 196 **CONSENT AND ETHICAL APPROVAL**

197 Informed consent was taken from the patient and no ethical clearance is required.
198

199 200 **COMPETING INTERESTS**

201 Authors have declared that no competing interests exist.
202

203 204 **REFERENCES**

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