

## Case report

### **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.

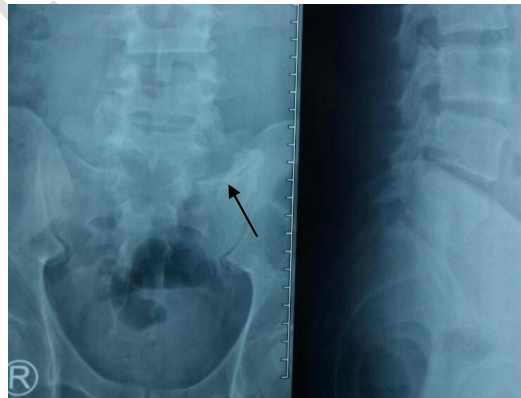
## 2. PRESENTATION OF CASE

**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).

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A 23 years old well and healthy Malay lady who works as a clerk attended to the .....Clinic for the treatment of lower back pain for 1 year.

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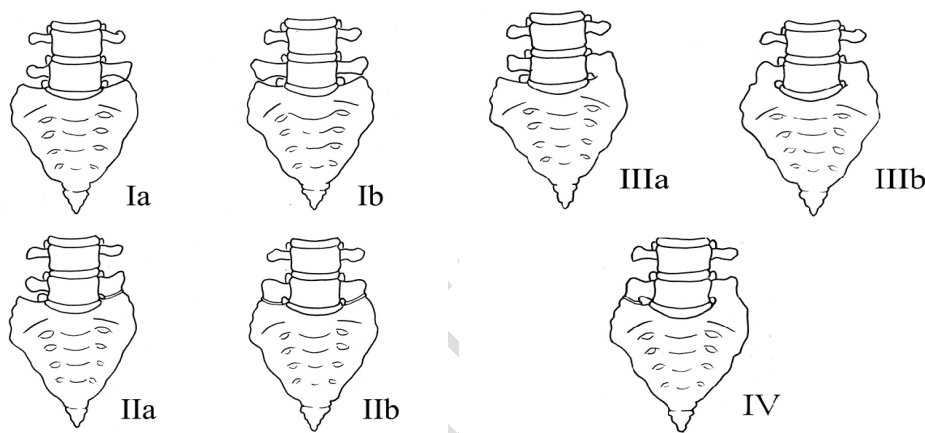
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75 **3. DISCUSSION**

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77 Low back pain in the presence of an LSTV was originally noted by Mario Bertolotti in 1917 and termed  
78 "Bertolotti's Syndrome". Low back pain is a prevalent problem with multiple causes. Approximately  
79 80% of adults seek a physician's help to deal with their low back pain at some point in their lives [5].  
80 Therefore, it is crucial that patients with Bertolotti's syndrome be accurately diagnosed and treated.  
81 According to Castellvi's classification as shown on Figure 3, there are four types of lumbosacral  
82 transitional vertebrae on the basis of morphologic characteristics. Type I includes unilateral (Ia) or  
83 bilateral (Ib) dysplastic transverse processes, measuring at least 19 mm in width (craniocaudal  
84 dimension). Type II exhibits incomplete unilateral (IIa) or bilateral (IIb) lumbarization/sacralization with  
85 an enlarged transverse process that has a diarthrodial joint between itself and the sacrum. Type III  
86 LSTV describes unilateral (IIIa) or bilateral (IIIb) lumbarization/sacralization with complete osseous  
87 fusion of the transverse process (es) to the sacrum. Type IV involves a unilateral type II transition with  
88 a type III on the contralateral side [6]. Literature indicates that Type II and IV have the most positive  
89 association with lower back pain [7].  
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91  
92 **Figure 3: Illustration demonstrating the Castellvi classification of LSTVs.**  
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97 Unilateral LSTV is more common than bilateral LSTV (9% vs. 3%) [8]. Men are more likely to be  
98 affected than women (28.1% vs. 11.1%) [9]. Patients with LSTV usually present to clinic with lower  
99 back pain after second decade of life [10]. According to Quinlan et al, the overall incidence of  
100 Bertolotti's syndrome has been reported to be between 4% to 8% in patients with low back pain and  
101 10% in patients presenting with back and leg pain under 30 years of age [3]. Throughout the  
102 literature, the prevalence of LSTV in patients seeking care for low back pain ranges from 4.6%-35.6%  
103 [11]. Due to its wide prevalence, Quinlan et al. encourages physicians to consider Bertolotti's  
104 syndrome in the differential diagnosis for low back pain, especially in younger patients [3].  
105

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

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

127 Management of chronic low back pain associated with Bertolotti's syndrome should be initiated with a  
128 comprehensive conservative approach consisting of activity modification, physical therapy and home  
129 exercise, psychosocial support, and medications such as NSAIDs, muscle relaxants, anticonvulsants,  
130 and antidepressants. Conservative nonsurgical management includes local injection of anaesthetic  
131 and corticosteroids within the **pseudo-articulation** or contralateral facet joint, radiofrequency ablation  
132 and surgical management with partial transverse process resection, and/or posterior spinal fusion  
133 [17]. Direct local anaesthetic and steroid injection or surgical resection of the anomalous or  
134 contralateral facet joint has produced successful relief of pain and can yield valuable diagnostic  
135 information [18-21].  
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137 Interventional approaches can be beneficial in cases refractory to conventional conservative  
138 management. For those who presented with lumbosacral radiculopathy or radiculitis symptoms due to  
139 the direct compression or irritation of the nerve root by an enlarged transverse process or **pseudo-**  
140 **articulation**, a transforaminal or interlaminar epidural steroid injection maybe considered. In a study  
141 done by Zhang et al, a diagnostic lumbar facet medial branch block can be performed for those with  
142 clinical evidence of lumbar facet pain due to the reduced motion between the LSTV and the sacrum.  
143 Radiofrequency neurotomy of the target medial branches may provide longer-term pain relief if the  
144 patient has greater than 50% pain reduction from the diagnostic medial branch block. For those  
145 suspected of sacroiliac dysfunction due to increased load to the sacroiliac joint with restricted motion  
146 between the LSTV and the sacrum, a diagnostic sacroiliac joint injection maybe attempted. If the  
147 patient has greater than 50% pain reduction, radiofrequency neurotomy of the L5 dorsal ramus, S1  
148 and S2 lateral branches may provide longer-term pain relief [21]. In a case report, Burnham described  
149 a successful novel radiofrequency technique for treating symptomatic lumbosacral junction **pseudo-**  
150 **articulation** in which he fluoroscopically guided local **anaesthetic/** corticosteroid injection into the  
151 **pseudoarthrosis** circumferentially around the posterior pseudarthrosis articular margin. Accordingly,  
152 bipolar radiofrequency strip thermal lesions were created at the same locations. Complete pain relief  
153 and full restoration of function was achieved for 16 months post procedure [22]. Almeida et., also  
154 reported that radio-frequency denervation is another possible treatment option and provided  
155 temporary relief of pain due to an anomalous articulation [9].  
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157 Operative treatment is suggested in selected patients. For example, resection of the transverse  
158 process may be beneficial for those who demonstrate pain truly emanating from a transitional joint  
159 and failed conservative treatment. If the pain source is from a degenerated disc above a transitional  
160 level, posterior fusion is an option as well [1]. In a case report, Brault et al described successful  
161 treatment of contralateral facetogenic pain by resection of the ipsilateral anomalous articulation [18].  
162 Jonsson et al reported relief of pain in 9 of 11 patients following surgical resection of a unilateral LSTV  
163 **pseudo-articulation** [23]. Ugokwe et al and Almeida et al similarly describe successful treatment after  
164 surgical resection [9, 20]. In a case series of 8 patients who underwent surgical resection of the  
165 unilateral anomalous articulation and 8 patients who underwent posterolateral fusion of the LSTV,  
166 Santavirta et al reported improvement in pain in 10 of 16 patients at 9-year follow-up without a  
167 difference between the fusion or resection groups [1]. In a case report, Yousif et al performed a  
168 minimally invasive tubular resection of a transitional lumbosacral **pseudo joint** and the short-term  
169 outcomes were favourable with the patient discharged, pain free 36 hours postoperatively, with return  
170 to normal work duties in two weeks and remained pain free six weeks postoperatively. The use of a  
171 minimally invasive tubular approach was key in minimizing unnecessary tissue dissection via a small  
172 35 mm incision. It allowed mobilisation over an obstructive osseous ridge and meant that bony  
173 resection was limited only to the pathological joint, minimizing the risk of future back pain [24]. Li et al  
174 also performed minimally invasive tubular resection of the anomalous transverse process in 7 patients  
175 with Bertolotti's syndrome. Three (43%) of 7 patients reported complete resolution of low-back pain, 2  
176 (29%) of 7 patients had reduced low-back pain, and 2 patients (29%) experienced initial relief but  
177 return of low-back pain at 1 and 4 years postoperatively. Three (50%) of the 6 patients with radicular

178 pain had complete relief of this symptom. They suggest that minimally invasive resection of the LSTV  
179 be used in select patients in whom conservative therapy has failed and who have pain attributed to  
180 the LSTV [25].  
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#### 182 183 **4. CONCLUSION** 184

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

#### 194 195 **CONSENT AND ETHICAL APPROVAL**

196 Informed consent was taken from the patient and no ethical clearance is required.  
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#### 198 199 **COMPETING INTERESTS**

200 Authors have declared that no competing interests exist.  
201

#### 202 203 **REFERENCES** 204

- 205 1. Santavirta S, Tallroth K, Ylinen P, Suoranta H: Surgical treatment of Bertolotti's syndrome.  
206 Follow up of 16 patients. *Arch Orthop Trauma Surg* 1993, 112:82-87, 1993.
- 207 2. Bertolotti M: Contributo alla conoscenza dei vizi differenziazione regionale del rachide con  
208 speciale riguardo all'assimilazione sacrale della V. lombare. *Radiol Med.* 4:113-144, 1917.
- 209 3. Quinlan JF, Duke D, Eustace S. Bertolotti's syndrome. A cause of back pain in young people.  
210 *J Bone Joint Surg.* 88:1183-1186, 2006.
- 211 4. Ju CI, Kim SW, Kim JG, Lee SM, Shin H, Lee HY, et al. Decompressive L5 transverse  
212 processotomy for Bertolotti's syndrome: A preliminary study. *Pain Physician.* 20: E923-E932,  
213 2017.
- 214 5. Anderson GBJ: The epidemiology of spinal disorders, in Frymoyer JW (ed): *The Adult Spine:*  
215 *Principles and Practice.* Philadelphia, Lippincott-Raven. 4: 93-141, 1997.
- 216 6. Castellvi AE, Goldstein LA, Chan DPK: Lumbosacral transitional vertebra and their  
217 relationship with lumbar extradural defects. *Spine* 1983, 9:493-495, 1983.
- 218 7. Jancuska JM, Spivak JM, Bendo JA. A review of symptomatic lumbosacral transitional  
219 vertebrae: bertolotti's syndrome. *Int J Spine Surg.* 9:42, 2015.
- 220 8. Mahato NK. Morphometric analysis and identification of characteristic features in sacra  
221 bearing accessory articulations with L5 vertebra. *Spine J.* 10 (7): 616-621, 2010.
- 222 9. Nardo L, Alizai H, Virayavanich W, Liu F, Hernandez A, Lynch JA, et al. Lumbosacral  
223 transitional vertebrae: association with low back pain. *Radiology.* 265 (2): 497-503, 2012.
- 224 10. Almeida DB, Mattei TA, Soria MG, et al. Transitional lumbosacral vertebrae and low back  
225 pain: diagnostic pitfalls and management of Bertolotti's syndrome. *Arq Neuropsiquiatr.*  
226 67:268-72, 2009.
- 227 11. Apazidis A, Ricart PA, Diefenbach CM, Spivak JM. The prevalence of transitional vertebrae in  
228 the lumbar spine. *The spine journal: official journal of the North American Spine Society.* 11  
229 (9): 858-862, 2011.
- 230 12. Aihara T, Takahashi K, Ogasawara A, Itadera E, Ono Y, Moriya H: Intervertebral disc  
231 degeneration associated with lumbosacral transitional vertebra: A clinical and anatomical  
232 study. *J Bone Joint Surg Br.* 87:687-691, 2005.
- 233 13. Luoma K, Vehmas T, Raininko R, Luukkonen R, Riihimäki H: Lumbosacral transitional  
234 vertebra: Relation to disc degeneration and low back pain. *Spine.* 29:200-205, 2004.

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14. Brown MF, Rockall AG, Hallam P, Hall-Craggs MA, Edgar MA: Transitional lumbosacral vertebra: Incidence of disc degeneration above and below. *J Bone Joint Surg Br.* 82-B (Suppl II):180, 2000.
15. Elster AD. Bertolotti's syndrome revisited: transitional vertebrae of the lumbar spine. *Spine.* 14:1373-7, 1989.
16. Cuenca C, et al. Bertolotti's syndrome in children: from low-back pain to surgery. A case report. *Neurochirurgie* 2009.
17. Konin GP, Walz DM. Lumbosacral transitional vertebrae: Classification, imaging findings, and clinical relevance. *Am J Neuroradiol.* 31 (10): 1778-1786, Nov 2010.
18. Brault JS, Smith J, Currier BL. Partial lumbosacral transitional vertebra resection for contralateral facetogenic pain. *Spine (Phila Pa 1976).* 26:226-29, 2001.
19. Marks RC, Thulbourne T. Infiltration of anomalous lumbosacral articulations: steroid and anesthetic injections in 10 back pain patients. *Acta Orthop Scand.* 62:139-41, 1991.
20. Ugokwe KT, Chen TL, Klineberg E. Minimally invasive surgical treatment of Bertolotti's syndrome: case report. *Neurosurgery.* 62:454-55, 2008.
21. Zhang R, Cheng J. Interventional management of chronic low back pain associated with Bertolotti's syndrome: report of case series. *Ann Clin Case Rep.* 2: 1348, 2017.
22. Burnham R. Radiofrequency sensory ablation as a treatment for symptomatic unilateral lumbosacral junction pseudarticulation (Bertolotti's syndrome): a case report. *Pain Med.* 11(6): 853-855, 2010.
23. Jonsson B, Stromqvist B, Egund N. Anomalous lumbosacral articulations and low back pain: evaluation and treatment. *Spine (Phila Pa 1976).* 14:831-34, 1989.
24. Yousif S, Wood M. Minimally invasive resection of lumbosacral pseudojoint resulting in complete resolution of a lower back pain – A case report and review of Bertolotti syndrome. *J Clin Neurosci,* 2018.
25. Li Y., Lubelski D., Abdullah, K.G., Mroz, T.E., & Steinmetz, M.P. Minimally invasive tubular resection of the anomalous transverse process in patients with Bertolotti's syndrome. *Journal of Neurosurgery: Spine SPI.* 20 (3), 283-290, 2014.

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