Bertolotti syndrome

J.Sales-Llopis

Neurosurgery Department, General University Hospital Alicante, Spain

Latest PubMed-Related Articles

- How I do it: minimally invasive surgical decompression for lumbosacral extraforaminal stenosis (Far-Out Syndrome)
- Complete, Sustained Resolution of Pain With Pseudoarthrectomy for Bertolotti Syndrome: A Case Report
- Bertolotti syndrome: an often overlooked cause of specific back pain
- A case report of ineffective electroconvulsive therapy for chronic pain
- Bilateral Bertolotti Syndrome: 2-Dimensional Operative Video
- A Case of Unilateral Bertolotti's Syndrome
- A Case Study of Bertolotti's Syndrome in an Adolescent Patient
- Functional outcomes of microdiscectomy in Bertolotti syndrome: the relationship between lumbosacral transitional vertebrae and lumbar disc herniation: a prospective study in Greece

Bertolotti Syndrome is a congenital condition characterized by the presence of a lumbosacral transitional vertebra (LSTV)—typically an enlarged transverse process of the L5 vertebra—which forms a pseudoarticulation or fusion with the sacrum or iliac bone. This anomalous articulation can alter spinal biomechanics and is associated with chronic low back pain, especially in young adults.

History and etymology

The Italian radiologist Mario Bertolotti (1876-1957) was the first to describe the syndrome in 1917 [1].

Key Features

Structural anomaly: Lumbosacral transitional vertebra (LSTV)

Pain mechanism: Mechanical low back pain due to abnormal load transmission

Often associated with:

Degenerative disc disease at the adjacent segment (usually L4-L5)

Unilateral or bilateral pseudoarthrosis

Age group: Typically presents in adolescents or young adults (e.g., <30 years)

Can be underdiagnosed or misattributed to muscular or discogenic pain.

Although it may be a consideration in younger patients, the entity is considered controversial and has been both supported and disputed. Some studies suggest that lumbosacral transitional vertebrae types II and IV positively correlate with the prevalence and severity of lower back pain and buttock pain [2].

Epidemiology

Prevalence is estimated at ~5% (range 4-8%) of the population.

Castellvi Classification

The Castellvi classification is used for the lumbosacral transitional vertebra (LSTV).

LSTVs are classified into 4 types, with Type 2B being most commonly associated with symptomatic Bertolotti syndrome:

Type I: enlarged and dysplastic transverse process (at least 19 mm)

Ia: unilateral

Ib: bilateral

type II: pseudoarticulation of the transverse process and sacrum with incomplete lumbarization/sacralization; enlargement of the transverse process with pseudoarthrosis

IIa: unilateral

see Castellvi Type 2B lumbosacral transitional vertebra

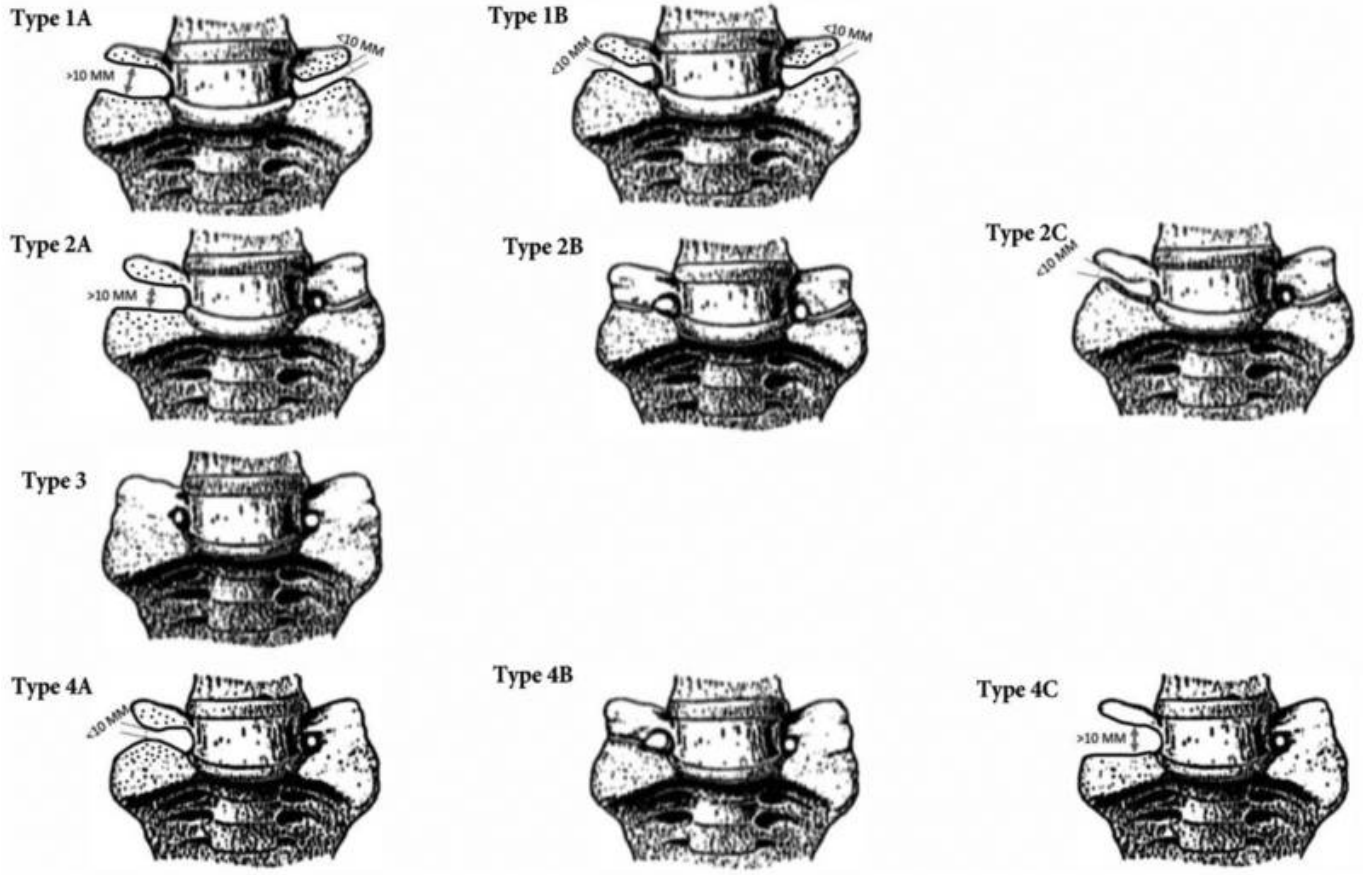
type III: transverse process fuses with the sacrum and there is complete lumbarization or sacralization, enlarged transverse process with complete fusion

IIIa: unilateral

IIIb: bilateral

type IV: type IIa on one side and type IIIa on the contralateral side [3].

Jenkins Classification



Clinical features

Bertolotti syndrome (BS) is characterized by chronic pain and functional impairment associated with lumbosacral transitional vertebrae (LSTVs).

Pathology

The study aimed to investigate the histologic characteristics of the pseudoarticulation between the enlarged transverse process and sacrum seen in Castellvi 2a LSTV and explore the involvement of nervous tissue in pain generation.

Methods: Immunohistochemical analysis using S100 protein staining was performed to assess the presence of nerve tissue.

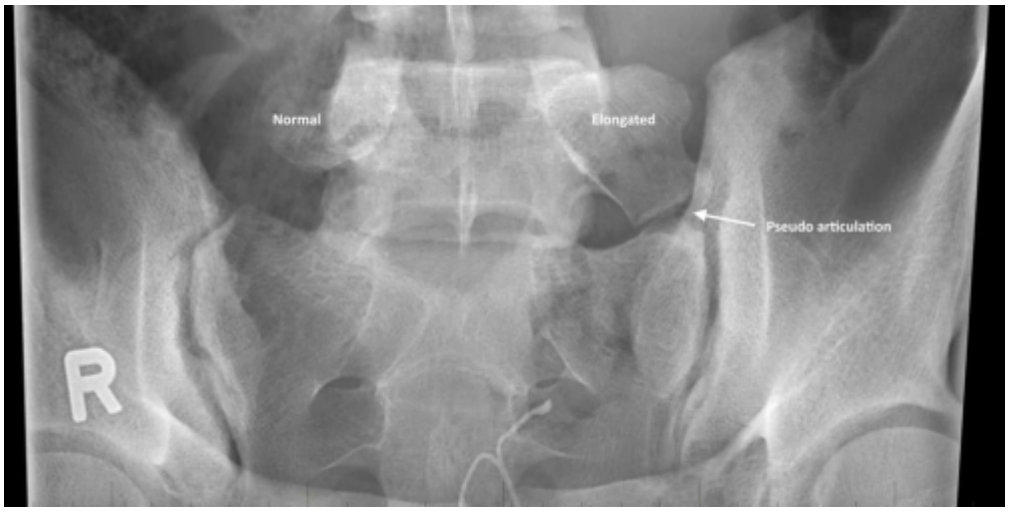
Results: These changes included fibrillation, chondrocyte cloning, alterations in the proteoglycan matrix, and focal chondrocyte necrosis. Notably, no nerve tissue was observed in any of the specimens, as confirmed by negative S100 protein staining.

Conclusions: The study findings suggest that nerve tissue is not involved in BS's nociceptive mechanisms underlying pain. The histologic similarities between the pseudoarticulation and osteoarthritic joints indicate that pseudoarticulation may be a significant source of pain in BS. These insights contribute to our understanding of the pathophysiology of BS and support treatment paradigms prioritizing pain control with medications such as NSAIDs before considering surgical intervention. Future studies with larger sample sizes and in vivo models are needed to further

validate these findings and explore the changes in joint histology under biomechanical forces in LSTVs [4].

Diagnosis

Lumbosacral spine radiographs



X-rays (AP and lateral) → initial detection, are usually sufficient for identifying this skeletal abnormality, however, can not definitively identify the transitional vertebrae as the source of pain. MRI is useful when radicular features with a prolapsed disc co-occur.

CT scan

→ better bone anatomy

MRI

→ Evaluates disc pathology and excludes other causes.

Diagnostic injection

Diagnostic injection at the pseudoarticulation can confirm the source of pain.

Diagnostic Injection at the L5 Pseudoarticulation: Technique Guide

 Purpose: To determine whether the pseudoarticulation between an enlarged transverse process of L5 and the sacral ala is the source of a patient’s low back pain.

1. Pre-procedure Preparation: Patient position: Prone, with a cushion under the abdomen to reduce lumbar lordosis.

Monitoring: Standard vital signs.

Sterile field: Prep and drape the lower lumbar area using antiseptic solution.

Imaging guidance: Fluoroscopy or CT is mandatory for accurate needle placement.

2. Anatomical Target: The pseudoarticulation formed between the transverse process of L5 (usually

hypertrophic) and the sacral ala.

Often bilateral—each side must be assessed separately.

3. Needle Placement: Use a 22G spinal needle or equivalent.

Under fluoroscopic guidance (oblique or AP view), advance the needle toward the joint space between the L5 transverse process and the sacral ala.

Confirm bone contact at the pseudoarticulation.

Use contrast (e.g., iohexol) to verify extra-vascular and extra-thecal placement.

4. Injection Protocol: Inject:

1–2 mL of 1% lidocaine (or 0.25% bupivacaine) for diagnostic purposes.

Optional: + 1 mL of corticosteroid (e.g., triamcinolone 20 mg) if a therapeutic effect is also desired.

Inject slowly, monitor for reproduction or relief of symptoms.

5. Post-procedure Assessment: Monitor the patient for 30–60 minutes.

Ask for pain relief level (VAS scale) immediately and up to 24–48 hours.

> 75% pain relief is typically considered positive, confirming the pseudoarticulation as the pain generator.

6. Complications (rare): Bleeding or hematoma.

Infection (extremely rare under sterile technique).

Allergic reaction to local anesthetic or contrast.

Inadequate pain relief → negative test.

 Clinical Implication: A positive response may justify surgical resection of the enlarged transverse process in refractory cases of Bertolotti’s syndrome.

Treatment

Bertolotti syndrome treatment

Prognosis

- Harness risk stratification of diabetic patients with dengue in a cohort study
- Redefining the Treatment of Lumbosacral Transitional Vertebrae for Bertolotti Syndrome: Long-Term Outcomes Utilizing the Jenkins Classification to Determine Treatment
- Operative Treatment of Bertolotti Syndrome: Resection Versus Fusion
- Lumbar radiculopathy due to Bertolotti's syndrome: Alternative method to reveal the "hidden

zone" - A report of two cases and review of literature
- Surgical interventions for Bertolotti's syndrome: case report and review of unsatisfactory cases in the literature
- Paraneoplastic neurological syndromes associated with non-Hodgkin lymphoma: a case series
- The effectiveness of preoperative assessment using a patient-specific three-dimensional pseudoarticulation model for minimally invasive posterior resection in a patient with Bertolotti's syndrome: a case report
- Bertolotti syndrome: a not-to-miss cause of chronic low back pain in young adults

Systematic reviews

A systematic review was conducted by the PRISMA guidelines (Preferred Reporting Items for Systematic Reviews and Meta-Analyses). The SCOPE criteria (Structuring Comparative Outcome Reporting in Epidemiology) were considered to ensure comprehensive and transparent reporting. A systematic search was performed in PubMed using the search terms 'Bertolotti', 'Bertolotti syndrome', and 'Lumbosacral transitional vertebrae' by a single investigator, resulting in the inclusion of 112 studies. Articles that were not available in English or German were excluded.

Results: Patients with symptomatic Bertolotti syndrome often suffer from chronic low back pain and radiculopathies. Imaging techniques, particularly X-rays and MRI, play a key role in diagnosis. Conservative treatments show limited success. Surgical resection of the transverse process (processectomy), especially after positive test infiltrations, leads to significant pain relief, particularly in younger patients without degenerative changes.

Discussion: Treatment choice depends on individual anatomy and the presence of degenerative changes. While conservative measures are initially recommended, processectomy shows promising results in carefully selected patients. Fusion surgeries should only be considered in cases of instability. Further studies are needed to confirm the effectiveness of invasive procedures [5].

Retrospective Observational Cohort Studies

In a Retrospective Observational Cohort Study reviewed 103 patients from 2012 through 2021 who had surgically treated Bertolotti syndrome. We identified 56 patients with Bertolotti syndrome and at least 6 months of follow-up. Patients with iliac contact preoperatively were presumed to be more likely to have hip pain that could respond to surgical treatment, and those patients were tracked for those outcomes as well.

Type 1 patients (n = 13) underwent resection. Eleven (85%) had improvement, 7 (54%) had good outcome, 1 (7%) had subsequent surgery, 1 (7%) was suggested additional surgery, and 2 (14%) were lost to follow-up. In Type 2 patients (n = 36), 18 underwent decompressions and 18 underwent fusions as a first line. Of the 18 patients treated with resection an interim analysis saw 10 (55%) with failure and needing subsequent procedures. With the subsequent procedure, 14 (78%) saw improvement. For fusion surgical patients, 16 (88%) saw some improvement and 13 (72%) had a good outcome. In Type 4 patients (n = 7), 6 (86%) did well with unilateral fusion, with durable benefit at 2 years. In patients who had hip pain preoperatively (n = 27), 21 (78%) had improvement of hip pain postoperatively.

The Jenkins classification system provides a strategy for patients with Bertolotti syndrome who fail

conservative therapy. Patients with Type 1 anatomy respond well to resection procedures. Patients with Type 2 and Type 4 anatomy respond well to fusion procedures. These patients respond well regarding hip pain [6)]

Retrospective Case-Control Studies

The purpose of a Retrospective Case-Control study was to evaluate the interreader reliability of detection and classification of LSTV with standard AP radiographs and report its accuracy by use of intermodality statistics compared with MRI as the gold standard.

Study design/setting: Retrospective case-control study.

Patient sample: A total of 155 subjects (93 cases: LSTV type 2 or higher; 62 controls).

Outcome measures: Interreader reliability in detection and classification of LSTV using standard AP radiographs and coronal MRI as well as accuracy of radiographs compared with MRI.

Methods: After institutional review board approval, coronal MRI scans and conventional AP radiographs of 155 subjects (93 LSTV type 2 or higher and 62 controls) were retrospectively reviewed by two independent, blinded readers and classified according to the Castellvi classification. Interreader reliability was assessed using kappa statistics for detection of an LSTV and identification of all subtypes (six variants; 1: no LSTV or type I, 2: LSTV type 2a, 3: LSTV type 2b, 4: LSTV type 3a, 5: LSTV type 3b, 6: LSTV type 4) for MRI scans and standard AP radiographs. Further, accuracy and positive and negative predictive values were calculated for standard AP radiographs to detect and classify LSTV using MRI as the gold standard.

Results: The interreader reliability was at most moderate for the detection (k=0.53) and fair for classification (wk=0.39) of LSTV in standard AP radiograph. However, the interreader reliability was very good for detection (k=0.93) and classification (wk=0.83) of LSTV in MRI. The accuracy and positive and negative predictive values of standard AP radiograph were 76% to 84%, 72% to 86%, and 79% to 81% for the detection and 53% to 58%, 51% to 76%, and 49% to 55% for the classification of LSTV, respectively.

Conclusion: Standard AP radiographs are insufficient to detect or classify LSTV. Coronal MRI scans, however, are highly reliable for classification of LSTV [7)].

Cross-Sectional Observational Studies

Nardo et al. assessed the prevalence and degree of lumbosacral transitional vertebrae (LSTV) in the Osteoarthritis Initiative (OAI) cohort to assess whether LSTV correlates with low back pain (LBP) and buttock pain and to assess the reproducibility of grading LSTV.

Materials & methods: Institutional review board approval was obtained, and informed consent documentation was approved for the study protocol. Standard standing pelvic radiographs that included the transverse processes of L5 were graded according to Castellvi classification of LSTV in 4636 participants (1992 men and 2804 women; aged 45-80 years) from the OAI cohort. These data were correlated with prevalence and severity of LBP and buttock pain.

Results: Prevalence of LSTV was 18.1% (841 of 4636), with a higher rate in men than in women (28.1% vs 11.1%, respectively; P<.001). Of the 841 individuals with LSTV, 41.72% were type I (dysplastic enlarged transverse process), 41.4% were type II (pseudoarticulation), 11.5% were type III (fusion), and 5.2% were type IV (one transverse process fused and one with pseudoarticulation). Of the participants without LSTV, 53.9% reported LBP, while the prevalence of LBP for types I, II, III, and IV was 46%, 73%, 40%, and 66%, respectively (P<.05, χ2 test). Types II and IV had higher prevalence and severity of LBP and buttock pain (P<.001) [8].

Case report from the HGUA

Bertolotti's Syndrome Associated with L4-L5 Disc Protrusion in a Young Adult with Occupational Low Back Pain

Abstract: We report the case of a 24-year-old male presenting with chronic low back pain radiating to both legs, exacerbated by trunk flexion and occupational heavy lifting. Imaging revealed bilateral enlarged transverse processes of L5 articulating with the sacral ala (Castellvi type 2B), consistent with Bertolotti's syndrome. A concomitant L4-L5 posterior disc protrusion with annular tear was also identified. No nerve root compression or spinal canal stenosis was observed. This case highlights the importance of recognizing lumbosacral transitional anomalies as a potential cause of mechanical low back pain in young adults with physically demanding occupations.

—

Keywords: Bertolotti's syndrome, transitional vertebra, enlarged transverse process, low back pain, disc protrusion, Castellvi classification

—

Case Presentation: A 24-year-old male warehouse worker presented with a several-year history of chronic low back pain, resistant to physiotherapy. The pain had gradually become more disabling, with recent episodes of radiation to both lower limbs. He reported worsening of symptoms with trunk flexion and heavy lifting. Neurological examination was unremarkable.

Plain radiographs revealed bilateral hypertrophic transverse processes of the L5 vertebra with suspected articulation with the sacral ala. A lumbar spine MRI was performed for further evaluation.

—

Imaging Findings:

* **Transitional vertebra at L5** with **bilateral enlarged transverse processes** forming **pseudoarticulations with the sacrum**, consistent with **Castellvi type 2B** anomaly (Bertolotti's syndrome). * At **L4-L5**, a **posterocentral disc protrusion with annular tear** was observed. * **No nerve root compression**, **no foraminal stenosis**, and **no spinal canal narrowing**. * **Conus medullaris** was normally positioned, and **cauda equina roots** appeared normal.

—

Diagnosis:

* **Bertolotti's syndrome** (Castellvi type 2B) * **L4-L5 disc protrusion with annular tear**, non-

compressive

A diagnostic injection at the L5 pseudoarticulation is a minimally invasive procedure used to confirm whether the pseudoarticulation between an enlarged transverse process of L5 and the sacrum (as seen in Bertolotti’s syndrome) is the source of a patient's low back pain.

 What is it? A local anesthetic, sometimes combined with a corticosteroid, is injected under fluoroscopic or CT guidance directly into the pseudoarticulation. If the patient experiences significant pain relief, the pseudoarticulation is likely the pain generator.

 Purpose: Diagnostic: To determine if the transitional segment is the true cause of the pain.

Prognostic: A positive response may predict good results from surgical resection of the transverse process.

Therapeutic (temporarily): Some patients may benefit from longer-lasting steroidal relief.

 Clinical Relevance in This Case: In the case of Christian Requena:

He presents with Bertolotti's syndrome (Castellvi 2B).

There is no significant nerve compression, so his pain is likely mechanical and related to the transitional segment.

A diagnostic injection would help confirm the pseudoarticulation as the pain generator before considering surgical intervention.

Management and Follow-up: Conservative treatment was advised, including postural re-education and ergonomic adaptation. Diagnostic injection at the L5 pseudoarticulation is under consideration. Surgical resection of the transverse process may be evaluated if symptoms persist.

—

Discussion: Bertolotti’s syndrome is a frequently underdiagnosed cause of low back pain in young patients. It results from a congenital lumbosacral transitional vertebra, often leading to abnormal biomechanical stress and adjacent disc degeneration. Differentiating symptomatic cases from incidental findings is essential. MRI aids in evaluating coexisting degenerative changes, while treatment may range from conservative approaches to surgical intervention in refractory cases.

—

Conclusion: This case underscores the relevance of considering Bertolotti’s syndrome in the differential diagnosis of chronic low back pain, particularly in young adults with mechanical stressors and atypical radiating symptoms, even in the absence of overt neural compression.

1)
LUPO M. Prof. Mario Bertolotti [Prof. Mario Bertolotti]. Minerva Med. 1958 May 9;49(37):Varia, 840-6. Italian. PMID: 13565569.

2) , 8)
Nardo L, Alizai H, Virayavanich W, Liu F, Hernandez A, Lynch JA, Nevitt MC, McCulloch CE, Lane NE, Link TM. Lumbosacral transitional vertebrae: association with low back pain. Radiology. 2012 Nov;265(2):497-503. doi: 10.1148/radiol.12112747. Epub 2012 Sep 5. PMID: 22952380; PMCID: PMC3480814.

3)
Castellvi AE, Goldstein LA, Chan DP. Lumbosacral transitional vertebrae and their relationship with lumbar extradural defects. Spine (Phila Pa 1976). 1984 Jul-Aug;9(5):493-5. doi: 10.1097/00007632-198407000-00014. PMID: 6495013.
4)
Desai A, Obiri-Yeboah D, McGrath K, Sheehan J, Loss J, Reith JD, Steinmetz MP. Histologic Assessment of Lumbosacral Transitional Vertebrae Pseudoarticulation as a Source of Pain in Bertolotti Syndrome. World Neurosurg. 2024 Jun 13:S1878-8750(24)00990-2. doi: 10.1016/j.wneu.2024.06.032. Epub ahead of print. PMID: 38878893.
5)
Hauser AL, Von Glinski A, Noriega Urena JF, Lange T, Murad S, Lewik G, Schulte T. Das Bertolotti-Syndrom: eine unterdiagnostizierte Ursache für spezifischen Rückenschmerz [Bertolotti syndrome: an often overlooked cause of specific back pain]. Orthopadie (Heidelb). 2025 Apr 29. German. doi: 10.1007/s00132-025-04656-1. Epub ahead of print. PMID: 40301172.
6)
Jenkins AL 3rd, Chung RJ, O'Donnell J, Hawks C, Jenkins S, Lazarus D, McCaffrey T, Terai H, Harvie C, Matsoukas S. Redefining the Treatment of Lumbosacral Transitional Vertebrae for Bertolotti Syndrome: Long-Term Outcomes Utilizing the Jenkins Classification to Determine Treatment. World Neurosurg. 2023 Jul;175:e21-e29. doi: 10.1016/j.wneu.2023.03.012. Epub 2023 Mar 8. PMID: 36898630.
7)
Farshad-Amacker NA, Lurie B, Herzog RJ, Farshad M. Interreader and intermodality reliability of standard anteroposterior radiograph and magnetic resonance imaging in detection and classification of lumbosacral transitional vertebra. Spine J. 2014 Aug 1;14(8):1470-5. doi: 10.1016/j.spinee.2013.08.048. Epub 2013 Oct 3. PMID: 24210581.