Case Reports and Reviews

Case Report

Bertolotti Syndrome Global Vision and Review

Gonçalo Januário

Head of Neurosurgical Department, Clínica Juaneda Miramar, Palma de Mallorca, Balearic Islands, Spain

Abstract

Bertolotti syndrome (BS) is a uncommon clinical condition, characterized by the presence of a lumbosacral transitional vertebra (LSTV) that may contribute to lower back pain.

Often underdiagnosed condition characterized by the presence of a lumbarization of the sacral vertebra or a pseudoarthrosis between the transverse process of the lower lumbar vertebra and the sacrum.

The condition is associated with altered biomechanics, result from abnormal development of the lumbosacral junction, leading to mechanical stress, nerve root compression, or musculoskeletal dysfunction. It typically presents as low back pain, often with associated sciatica or radicular symptoms.

While BS remains under-recognized, its diagnosis is essential in patients with chronic low back pain unresponsive to conventional treatments.

Diagnosis is primarily radiographic, with X-rays, CT scans, and MRIs are critical for identifying LSTV and ruling out other spinal pathologies.

Management strategies for Bertolotti syndrome include conservative treatments such as physical therapy, analgesics, and anti-inflammatory medications, as well as surgical options in severe cases or refractory cases.

Awareness of Bertolotti syndrome is essential for clinicians, as its presentation can overlap with more common lumbar pathologies, making accurate diagnosis and appropriate treatment critical for improving patient outcomes.

This syndrome is important to consider in the differential diagnosis of lower back pain to ensure appropriate management and improve patient outcomes.

Correspondence

Gonçalo Januário

Head of Neurosurgical Department, Clínica Juaneda Miramar, Palma de Mallorca, Balearic Islands, Spain

ORCID id: 0000-0003-2884-6422

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Introduction

The Bertolotti syndrome (BS) was first described by the Italian physician Giovanni Bertolotti in 1917. It is sometimes referred to as "lumbarization" or "sacralization" depending on the direction of the vertebral anomaly. Lumbarization refers to the presence of a 6th lumbar vertebra, while sacralization indicates that the 5th lumbar vertebra is fused or abnormally connected to the sacrum [1]. BS is a congenital condition characterized by lower back pain associated with an anatomical variation in the lumbosacral region. It involves the presence of an abnormal fusion or transitional vertebra at the lumbosacral junction, typically between the 5th lumbar vertebra (L5) and the sacrum. This anatomical variant can lead to biomechanical stress on the spine, resulting in pain and other symptoms [2].

Patients with BS often present with lower back pain, which can be exacerbated by certain movements, physical activities, or prolonged standing. The pain is often localized to the lumbosacral junction and can radiate into the buttocks or thighs. In some cases, additional symptoms such as radiculopathy or neurological deficits may be observed, though these are relatively rare [2].

The diagnosis of Bertolotti syndrome is primarily based on imaging techniques such as X-rays, CT (computed tomography) scans, and MRI (magnetic resonance imaging). These modalities allow for the identification of the transitional vertebra and any associated anomalies, such as disc degeneration or foraminal stenosis. Additionally, a careful clinical examination is necessary to rule out other causes of low back pain, such as herniated discs or spinal stenosis [3].

Recent research has further highlighted the relationship between Bertolotti syndrome and disc degeneration. Studies have found that the presence of a transitional vertebra may increase the risk of disc degeneration and facet joint arthritis at the affected segment, thereby contributing to chronic pain [4].

Management of Bertolotti syndrome typically involves conservative measures such as physical therapy, pain management, and lifestyle modifications. Non-steroidal anti-inflammatory drugs (NSAIDs) or other pain-relieving medications are often prescribed to alleviate symptoms. In cases where conservative treatment fails, surgical intervention may be considered. Surgical options can include decompression, fusion, or resection of the transitional vertebra, although these procedures are generally reserved for patients with severe or refractory symptoms [5].

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Furthermore, there is ongoing debate regarding whether Bertolotti syndrome is a causative factor in low back pain or merely a coincidental finding. Some studies suggest that a proportion of individuals with BS remain asymptomatic, while others argue that the anatomical variation can lead to altered biomechanics and eventually result in pain and dysfunction [6].

Mechanical Overview of Bertolotti Syndrome

As described previously BS is an anatomical variation in the lumbosacral junction that can lead to mechanical stress, contributing to lower back pain and other musculoskeletal issues.

The mechanical complications arise due to the altered biomechanics of the spine, caused by a transitional vertebra between the 5th lumbar vertebra (L5) and the sacrum. These anatomical changes can disrupt normal spinal movement, affect load distribution, and increase strain on adjacent structures such as the discs and facet joints.

Anatomical Variations in Bertolotti Syndrome, (Figure 1)

BS is a condition characterized by an anatomical variation in the lumbosacral junction with abnormal articulation or fusion between the last lumbar vertebra (L5) and the sacrum, creating a "transitional" vertebra. This pathology may cause mechanical issues and contribute to lower back pain. This anatomical variation can be classified into several types based on the nature of the abnormality in the vertebral structures at the L5-S1 segment.

Below are the four primary types of BS, with references from recent studies to support the mechanical implications and clinical findings.

Sacralization of the Fifth Lumbar Vertebra (L5-Sacralization)

The sacralization occurs when the 5th lumbar vertebra (L5) becomes fused or abnormally connected to the sacrum. This fusion can be partial or complete, significantly reducing movement at the L5-S1 junction.

Mechanical Impact

- The fusion leads to reduced motion at the L5-S1 joint, causing abnormal stress on adjacent lumbar segments (L4-L5), leading to disc degeneration, facet joint arthritis, and instability at other spinal levels [4].
- Altered biomechanics of the lumbosacral region increase load on the facet joints and intervertebral discs, potentially leading to degenerative changes and chronic low back pain.

Symptoms

- Chronic low back pain, especially localized in the lower lumbar and sacral regions.
- Pain may worsen with prolonged standing or certain movements, such as bending and lifting.

Lumbarization of the Sacrum (S1-Lumbarization)

Lumbarization refers to a condition in which the first sacral segment (S1) does not fuse with the sacrum, effectively giving rise to a 6th lumbar vertebra. This additional lumbar-like segment results in increased mobility at the lumbosacral junction.

Mechanical Impact

- The increased mobility at the L5-S1 junction can lead to instability in the lower back. The abnormal motion between S1 and L5 may increase the risk of degenerative disc disease and facet joint arthritis at these levels.
- Excessive motion at L5-S1 may lead to the development of abnormal stress patterns, further promoting degenerative changes in adjacent vertebrae [5].

Symptoms

- Localized pain in the lower back, particularly around the lumbosacral junction.
- Pain may increase with flexion, extension, or rotational movements of the spine.

Mixed Sacralization and Lumbarization (Partial Sacralization or Partial Lumbarization)

In cases of mixed sacralization and lumbarization, there is partial fusion between L5 and the sacrum (sacralization), but the fusion is not complete. Alternatively, there may be partial lumbar- like characteristics in the sacrum (lumbarization). This mixed form results in abnormal biomechanics that combine features of both sacralization and lumbarization.

Mechanical Impact

- The partially fused segments contribute to instability in the lumbosacral junction, leading to altered motion patterns.
- The abnormal joint dynamics between L5 and S1 increase the risk of facet joint degeneration and disc degeneration [6].
- The biomechanical stresses may result in asymmetrical load distribution, contributing to chronic pain or discomfort.

Symptoms

- Asymmetrical low back pain, often exacerbated by bending or twisting motions.
- Pain on one side of the lower back, depending on the degree of fusion or separation.

Transitional Vertebra with Unilateral or Bilateral Joint Formation

In this type, the transitional vertebra between L5 and the sacrum has a synovial or fibrocartilaginous joint on one side (unilateral) or both sides (bilateral). These abnormal joints can contribute to instability in the lumbosacral region and lead to mechanical dysfunction.

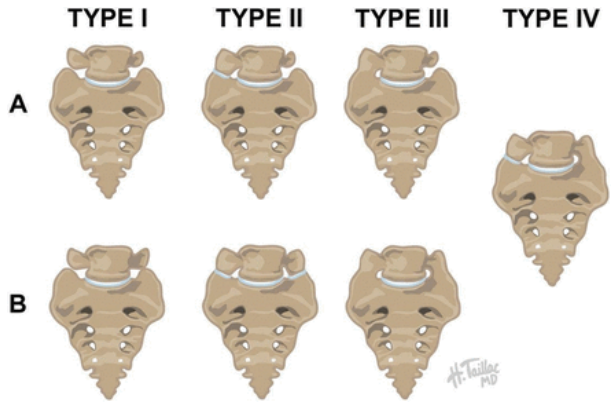
Mechanical Impact

- Facet joint formation between L5 and the sacrum can lead to increased strain on the adjacent structures, promoting degenerative arthritis and joint instability [4].
- The abnormal joint dynamics between L5 and S1 may alter the normal load distribution, leading to increased stress on the intervertebral discs and potentially causing disc bulging or herniation.

Symptoms

- Localized low back pain, often aggravated by activities such as bending, twisting, or lifting.
- Pain may radiate to the buttocks or thighs if there is nerve root compression due to abnormal joint formation.

These variations are considered congenital, but the mechanical effects are more pronounced in adulthood, particularly under physical stress.



Transitional vertebrae as classified by Castellvi et al. [7]
Type I: large transverse process.
Type II: diarthrodial joint between the transverse process and sacrum/ilium.
Type III: bony union between the transverse process and sacrum/ilium.
Type IV: unilateral type II and contralateral type III defects. For types I to III, subset A signifies a unilateral defect and subset B signifies bilateral defects.
(Original art by Heather Taillac, MD, Ochsner Clinic Foundation, New Orleans, LA.)

Figure 1. Diagram showing a transitional vertebra in Bertolotti syndrome.

Recently was present the Jenkins classification for Bertolotti syndrome and compared with the Castellvi classification presented above for patients with symptomatic lumbosacral transitional vertebra (LSTV) presenting for treatment. According to the defender of the Jenkins classification the original Castellvi classification does not organize the lumbosacral pathology into a sequence of structural variations that intuitively explain the complex anatomical and biomechanical range of all LSTV variants with regard to Bertolotti syndrome [8].

During the time the authors found the measurement of the gap between the transverse process and sacral ala to better predict the presence of Bertolotti syndrome and identify it a better target for diagnostic injections and subsequent surgical treatment for LSTV of the symptomatic patients [9].

The Jenkins classification use the transverse process proximity to better capture the clinical presentation of the symptoms of those with Bertolotti syndrome.

For the diagnosis are recommended the use of MRI, CT, and X-Ray imaging. The patients with radiographic finding of LSTV, appropriate clinical presentation, and identification of LSTV as the primary pain generator via diagnostic injections were diagnosed with Bertolotti's syndrome.

Finally the Jenkins classification improves on the prior Castellvi classification to more comprehensively describe the functional anatomy, identify uncaptured anatomy, and

Table 1*. Anatomic Definitions for Proposed Jenkins Classifications*

Classification	Laterality (More Abnormal Side)	Anatomic Description
Primary		
1A	(L)/(R)	Unilateral dysplastic transverse process (<10 mm between TP and ala [e.g., type 1A(L) gap is >2 mm, <10 mm only on left, >10 mm on right])
1B	(L)/(R)	Bilateral dysplastic transverse process (both sides <10 mm gap [e.g., type 1B(R) closer on right than left])
2A	(L)/(R)	Incomplete unilateral lumbarization/sacralization with enlarged transverse process that has a diarthrodial joint between itself and sacrum (<2 mm separation, with planar surface on transverse process parallel to opposite surface of ala but >10-mm gap on opposite side [e.g., type 2A(L): type 2 on left, with >10 mm on right])
2B	(L)/(R)	Incomplete bilateral lumbarization/sacralization with enlarged transverse process that has a pseudojoint between itself and sacrum (<2 mm of separation on both sides, with planar surfaces on transverse process parallel to opposite side of ala [e.g., type 2B is symmetrical but type 2B(R) has a larger interface on right])
2C	(L)/(R)	Dysplastic transverse process on 1 side and incomplete lumbarization/sacralization on other side (<10 mm but >2 mm on 1 side and <2 mm on other side [e.g., type 2C(L): type 1 on right and type 2 on left])
3	(L)/(R)	Bilateral lumbarization/sacralization with complete osseous fusion of transverse process to sacrum (no radiographic fusion plane viable on MRI, CT, or Ferguson radiographic views on either side)
4A	(L)/(R)	Lumbarization/sacralization with complete osseous fusion on 1 side with dysplastic transverse process on other side (type 1 on 1 side and type 3 on other side [e.g., type 4A(L): type 1 is open on right; type 3 on left])
4B	(L)/(R)	Lumbarization/sacralization with complete osseous fusion on 1 side and incomplete lumbarization/ sacralization on other side (type 3 on 1 side and type 2 on other side [e.g., type 4B(R): type 2 is open on right; type 3 on left])
4C	(L)/(R)	Lumbarization/sacralization with complete osseous fusion on one side, >10-mm gap on other side (type 3 on 1 side, type 1 on other side [e.g., type 4C(L): open on left; type 3 on right])
Secondary		
+L/R/B	+L/R/B	Denotes which side has contact between TP and iliac crest [e.g., type 2C(L)+L]

L, left side more prominent; R, right side more prominent; +, transverse process touches (within 2 mm) iliac bone; TP, transverse process; C, type 1 lumbosacral transitional vertebra on 1 side and type 2 lumbosacral transitional vertebra on other side; MRI, magnetic resonance imaging; CT, computed tomography.

better predict optimal surgical procedures to treat those with Bertolotti's Syndrome (Table 1).

Mechanical Impact on Spinal Function

The mechanical implications of Bertolotti syndrome primarily arise from the disruption of normal lumbar movement patterns and load distribution [10]:

1. **Altered Biomechanics:** The transitional vertebra changes the mechanical alignment of the lumbosacral junction. This can increase shear forces at the affected segment, causing abnormal stress on the intervertebral discs and facet joints. The altered alignment can result in pain and reduced mobility.
2. **Increased Stress on Discs and Facet Joints:** A transitional vertebra can lead to early degeneration of the intervertebral discs, as the altered biomechanics increase load on the discs. Facet joints can also undergo osteoarthritic changes due to the abnormal motion at the lumbosacral junction, which may cause joint instability and pain.
3. **Possible Nerve Root Compression:** In some cases, Bertolotti syndrome may lead to foraminal stenosis or nerve root compression. This can occur if the anomalous vertebra creates additional bony structures that encroach upon the neural foramen, leading to symptoms of radiculopathy, including pain, numbness, and tingling in the lower extremities.

Recent Mechanical Studies and Findings

Recent studies have investigated the mechanical outcomes of Bertolotti syndrome, particularly focusing on the impact on spine function, disc degeneration, and facet joint arthritis [11]:

- Kwan et al. (2023) found that patients with sacralization of L5 show a higher incidence of facet joint arthritis and disc degeneration in the affected segment due to altered weight-bearing and movement patterns.
- Zhang et al. (2022) studied the relationship between Bertolotti syndrome and lumbar spine instability. Their research indicated that the altered mechanical stress from a transitional vertebra may accelerate degenerative changes in the adjacent vertebral segments, leading to pain and instability.

Biomechanical Consequences

- **Increased Shear Forces:** A transitional vertebra can cause an abnormal mechanical load distribution, leading to increased shear forces on the discs, particularly at the L4-L5 and L5-S1 levels. This can cause early disc degeneration, disc herniation, or bulging, which may be painful and result in reduced range of motion [6].
- **Facet Joint Arthritis:** The abnormal motion between the transitional vertebra and the sacrum may lead to abnormal wear on the facet joints. Over time, this can contribute to osteoarthritis and cause significant back pain [4].
- **Compensatory Changes:** Because of the altered structure of the lumbosacral region, the spine may compensate by increasing the mobility of adjacent segments, leading to hypermobility or instability in other parts of the spine. This compensatory mechanism can further contribute to pain and dysfunction.

Clinical Implications and Management

Understanding the mechanical implications of Bertolotti syndrome is crucial for management:

- **Conservative Management:** This typically includes physical therapy to strengthen the muscles around the lumbosacral region, improve stability, and reduce pain. Bracing may also be used in some cases to reduce stress on the affected segments.
- **Surgical Intervention:** In severe cases where conservative management fails, surgery may be considered. The most common surgical approaches include:
  - Decompression of nerve roots in the presence of foraminal stenosis or nerve root impingement.
  - Fusion of the L5-S1 segment to stabilize the lumbosacral junction.
  - Resection of the transitional vertebra (particularly in cases of sacralization) to reduce mechanical stress and pain.

Conclusions

Bertolotti syndrome is a clinically significant condition that can contribute to lower back pain. This syndrome significantly alters the biomechanics of the lumbosacral junction, creating a predisposition for degenerative changes such as disc degeneration and facet joint arthritis. These mechanical effects often lead to chronic lower back pain, and understanding the biomechanical implications is crucial for proper diagnosis and management.

BS encompasses a spectrum of anatomical variations that affect the lumbosacral junction, each with unique mechanical and clinical implications.

Whether it involves sacralization, lumbarization, mixed forms, or the presence of abnormal joint formations, these variations disrupt normal spinal biomechanics, leading to conditions such as facet joint arthritis, disc degeneration, and lumbar instability.

However, due to the variety of anatomical presentations and the complexity of its symptoms, careful diagnostic assessment is crucial in determining whether the syndrome is the primary cause of the patient's discomfort.

Accurate diagnosis and tailored treatment plans are essential for managing the pain and mechanical dysfunction associated with Bertolotti syndrome.

In BS the option of conservative treatments are often effective, but surgery may be necessary in more severe cases.

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