Review Article

Bertolotti's syndrome: Prevalence, classification and current concepts of management: A review

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Abstract

Bertolotti's Syndrome (BS) or Lumbosacral Transitional Vertebra (LSTV) is the most common congenital malformation of the lumbosacral junction. It encompasses sacralization of L5 to lumbarization of S1. The prevalence ranges from 5% to 15% in different population base. Adolescents and middle-aged people are commonly affected. Clinical spectrum can range from asymptomatic individual to a constant low Backache (LBA) with or without radicular pain in buttocks or legs. Currently, there is no consensus about its association with low back pain and its subsequent management. We thoroughly evaluated all prominent literature regarding its diagnostic methods, classification, and subsequent management.
Key Word: bertolotti's syndrome; lumbosacral transitional vertebra; sacralisation; lumbrization.

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INTRODUCTION

Bertolotti's syndrome or lumbosacral transitional vertebra syndrome is a common cause of lower back pain, especially in younger population. Since it's identification by Bertolotti in 1917, it has been a constant topic of debate regarding its association with low back ache (LBA) and subsequent management. Being a most common congenital abnormality of the LS junction, symptomatology extends from asymptomatic incidental finding to constant source of low backache (LBA) in the middle aged population. Morphological changes in the L5 and/or S1 vertebrae lead to abnormal biomechanics and kinematic at LS junction that results in decrease motion at the involved part with consequently increased mobility and stress at adjacent segment of the spine. The problem ranges from enlarged transverse process of 5th lumbar vertebra, pseudo-arthrosis at enlarged transverse process and sacral ala and iliac bone, and complete fusion, to lumbarization of first sacral vertebral body. The disorder typically affects younger individuals of middle age group. As it involves younger population, constant LBA can cause psychosocial frustration, work inefficiency and loss of productive and economic activity. Till date, there is no consensus regarding its diagnostic method and subsequent treatment. We thoroughly reviewed all the prominent literatures on lumbosacral malformation and highlighted it's prevalence and potential pathological association with LBA, various diagnostic methods and different management techniques being used.

Anatomical Changes and Pathology: In lumbosacral transitional vertebra syndrome (LSTV) either the last lumbar vertebra (L5) acquires varying degrees of articulation to the sacrum and/or to the ilium, or the first sacral segment (S1) gets separated from the sacrum with transition to lumbar configuration. LSTV is a congenital anomalous enlargement of transverse processes of most caudal lumbar vertebra with subsequent articulation or fusion. Mario Bertolotti in 1917 first identified and termed it as bertolotti's syndrome. He stated that LSTV may cause low back pain due to arthritic changes at the pseudo arthrosis site. Further, Castellvi *et al*[1] classified it into four types (Fig 2). Type first comprises unilateral (IA,) or bilateral (IB) dysplastic transverse processes, measuring at least 19 mm height in vertical dimension. Type 2 involves

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unilateral (IIA, fig 1.) or bilateral (IIB) lumbarization/sacralization with formation of a di-arthrodial joint between the overgrown transverse process and sacral ala. Type 3 exhibits unilateral (IIIA) or bilateral (IIIB, fig. 3 & 4) fusion of enlarged transverse processes to sacral ala. Type IV involves a unilateral type II transition with a type III on the contralateral side. Transitional vertebra in lumbosacral region disturbs normal biomechanical anatomy. The sacrum, a large triangular bone dissipates and transmits body weight to the limb through SI joints. This capability of it surely depends on surface area of the articulation. A cadaver study theorized that congenitally small sacral surface area tries to add more surface by body's compensatory mechanism and functional requirement and consequently it involves L5 transverse processes in the neo-articulation. Conversely, in case of large sacral surface area, the articulation leaves S1 from sacroiliac articulation and S1 lumbarization occurs[2]. Apart from transverse process changes and neo-articulation (sacralization), morphological changes also affect other parts of the vertebra affected. In sacralization, pedical height, its transverse and sagittal dimension and sagittal angulation are decreased and caudal angulation increased. Width of laminae and height of pars interarticularis (PI) decrease. Smaller PI renders the spine vulnerable to spondylolysis and spondylolisthesis[3]. Lumbarization of the S1 conversely results in obtused pedicles, short distance between facets and sacral promontory[4]. Transitional vertebra also shows facets joint changes. In lumbarization, facet linear dimension and surface area are decreased and with maximal coronal orientation. Sacralization does not show much changes in morphology[5]. Nicholson *et al* observed a radiographically decreased intervertebral disc height at lumbar transitional segment and the sacrum in comparison to a normal disc between L5 and S1. Similarly, in case of lumbarization of S1, the S1-S2 disc assumes larger height, contrary to the normal disc which is rudimentary and vestigial[6]. Pathophysiology of pain generation in presence of LSTV has always been unclear. Many studies found increased prevalence of degenerative process at superjacent disc level to LSTV [1,7]. Causes of low back pain in association with transitional vertebra is multifactorial in origin. Presence of extra-foraminal stenosis, spinal stenosis; nerve root canal stenosis and facet joint degeneration were proposed mechanism for low back pain with or without radicular pain[8,9]. Connolly *et al* studied skeletal scintigraphy in young patients with LSTV and demonstrated that mechanical stress at pseudo articulation at L5 transverse process and sacral la and/or ilium could cause pain[10]. There is always ambiguity regarding which nerve is which in the scenario of LSTV because of altered anatomy. McCulloch and waddle stated that functional L5 root always originates from the last mobile segment of the spine. The last mobile segment is defined as the vertebra which has fully formed disc with bilaterally well-developed faced joints and separate transverse processes. So, in sacralized L5, the functional L5 correspond to the anatomical L4 nerve root. In lumbarized S1, L5 nerve root corresponds to S1 root[11]. Even after identification of transitional vertebra, there are many controversies regarding its association and mechanism of LBP. An increase in prevalence of disc degeneration and protrusion or extrusion is found in the disc above the transition L5 vertebra[1,12,13]. It has been found and theorized that reduced mobility at L5-S1 leads to hypo-mobility at that level and consequently hypermobility and stress at L4-L5 cause degeneration, disc protrusion/extrusion and early facets joint degeneration. Intervertebral disc at LSTV is protected (fig 5.)[12,13,14]. Asymmetric transitional vertebra has been considered as a potential source of back pain[15]. Presence of sclerotic changes and bony osteophytes signifies that a slight amount of motion may occur at pseudo articulation site, resulting in LBA[16]. Nardo *et al* proposed that there is positive relationship between LSTV type & type IV and LBA.[17] Quinlan *et al*[18] also concluded that LSTV should be considered a possible cause of LBA. Wiltse, in 1984, pointed out that the bony enlargement of L5 transverse process can lead to nerve entrapment between the anomaly and sacral ala and named it "far out syndrome" [19]. Conversely, several studies also found contradictory result[20] Luoma *et al*[21] in an MRI study of asymptomatic patients found that though there was increased prevalence of degeneration of the disc above the LSTV, an association between the findings and LBA was lacking. Tiny *et al*[22] included 4000 patients and did not reported that LSTV was associated with LBA.

Epidemiology: Prevalence of LSTV is varied in the literature due to differences in definition and diagnostic modalities employed. It ranges from 4% to 35.9% with a mean of 12.3%[5,23,24] Apazidis *et al* reported Castellvi type Ia as most common with a prevalence rate of 14.7%, though type Ia is considered of low clinical significance[24] Nardo *et al* reported that 40% of his cases of LSTV were of type I and II. Type III and type IV accounted for 11.5% and 5.25% respevtively[17] LSTV was reported more in man than woman. Sacralization is more common in man, while lumbarization of S1 is more prevalent in woman [17,25]. Common finding in some families suggested some genetic association responsible for the segmental development of the lumbosacral spine [23]. Embryonic development and segmentation of spine is probably controlled by families of genes, termed homeobox genes. These are abbreviated to *Hox* in mice and *HOX* in humans and are found on 4 clusters known as A, B, C, and D. Individual genes are numbered from 1 to 13. 1 is a cephalic gene and 13 is a more caudally placed gene[26].

Diagnosis and imaging: Due to dysplastic changes in vertebrae and intervertebral disc space, it is not always certain to identify and number the vertebrae. A Full x-ray of whole spine or a CT scan is not always done. A standard anteroposterior and lateral radiograph of lumbosacral spine can help reveal the transition vertebra. Normally, most caudal rectangular vertebra is considered as L5 in a lateral radiograph. In LSTV, the last rectangular shape can coincide with either L4 or L6/S1. Wigh and Anthony[9] described the "squaring" of the S1 as its ratio of AP distance of superior end plate and inferior end plate in lateral radiograph as ≤ 1.37. Sacralized L5 may depict the picture similar to S1 and transforms into a rhombus and wedge shaped. Typically, in lumbarization, involved transitional vertebral body of S1 appears squared up and infrajacent disc space appears decreased in height and a vestigial one[27,28]. The normal acute angulation between L5 and S1 is reduced and lumbar lordosis is found to be exaggerated. Anteroposterior radiograph with beam directed 30^0 cephalad (Ferguson view) and coronal sections of CT scan reveal from a broadened and enlarged transverse processes (TP) of L5, a pseudo-articulation at TP and sacral ala and ilium to complete fusion. Axial cuts of CT scan highlight a neo-articulation, facet joint orientation and their spatial relationship. 3D NC-CT also allows to see morphological changes in the transitional vertebra. A standard radiograph can fairly diagnose all types of LTV with accuracy ranging from 53% to 58%. Numbering of vertebra in association with BS is also important as it has a great bearing on localization of the level in planning of surgery. Without definite identification and numbering, a wrong level surgery may be ensued. For that purpose, a preoperative identification and numbering with correlation with intraoperative images are desired. Many a times only a radiograph or MRI of lumbosacral region is available. What appears as L1 with transverse process in AP radiograph, in essence may be a T12 vertebra with hypo-plastic rib. Similarly, Hughes RJ *et al*[29] described use of iliolumbar ligament for the numbering purpose as it reliably arises from L5 transverse processes (TP). It originates from L5 TP and extends to posteromedial iliac crest. It is visible as low signal images in both T1 and T2 weighted axial images of MRI. Hughes *et al* labeled an LSTV as L5 when no iliolumbar ligament was identified at the level above. When an iliolumbar ligament was seen to arise above the LSTV, then the vertebral body with iliolumbar ligament was labelled L5 and the LSTV as S1. However, this technique assumes that there is always 7 cervical, 12 thoracic and 5 lumbar vertebrae. It also does not respect the possibility of various segmentation anomaly at thoracolumbar junction. Whole spine radiograph with use of MR localizer helps in counting the vertebrae inferiorly from C2 and may help in true numbering[30]. However, MR sagittal images sometime may not pick up the changes. Tokgoz *et al*[31] did a large study in 1049 patients using MRI of lumbosacral area with a whole spine localizer and reported that about 1.3% patients were wrongly diagnosed as having LSTV, while 35.1% patients of abnormal segmentation were wrongly labelled as having normal segmentation. Even after correctly identifying LSTV, they reported incorrect vertebral level numbering in 60% of cases with total diagnostic error in 14.1% cases. Lumbarization of S1 demonstrated a well formed IVD at S1-S2 level with squared up S1 body. Sacralization revealed a rhombus shaped L5 body. Some anatomic landmarks such as right renal artery, aortic bifurcation and level of connus medullaris have also been studied. Lee *et al*[27] stated that aortic bifurcation and right renal artery are reliable landmarks in MRI or CT for identification of lumbosacral segments. Most the times, right renal artery present at L1-L2 disc level, but it is not always imaged or present in other locations in approximately 25% of cases[29]. Farshad *et al.* described a novel method of identifying an LSTV. The difference in the vertical mid-vertebral angles (Diff-VMVA) and the difference in the vertical anterior vertebral angles (Diff- VAVA) of the last three caudal segment of spine were calculated in sagittal MRI and lateral radiograph. A Diff- VMVA of $<10^0$ identified type III and type IV LSTV with a sensitivity of 100% and a specificity of 89% on MRI and a sensitivity of 94% and a specificity of 74% on lateral radiograph. Also, a Diff- VAVA, a sensitivity of 100% and a specificity of 76%% were obtained[32]. O' Driscoll *et al*[33] classified lumbosacral junction into four types [Table I]. Type 4 correlates with fused LSTV Castelvi type III or type IV. Paik *et al*[23] suggested inclusion of cervical spine in MR Imaging for numerically counting of the vertebrae to identify LSTV. In their study, 89.2% had 5 lumbar vertebrae (L5), 2.6% had 4 lumbar vertebrae (L4) and 8.2% had 6 lumbar vertebrae (L6). Types II, III, or IV LSTV were present in 10.6% of the patients, including 5.3% of sacralized L5 and 5.3% of lumbarized S1. Only 83.9% patients were modal type with 5 lumbar vertebrae without transitional vertebra. The last lumbar vertebra with no transition, looking like a modal L5 type, can be an L4 or an L6, as seen in 2.6% and 2.9% of the patients, respectively. Spinal nerves can be pinched because of bony growth. Weber *et al* reported two cases of LSTV resulting in entrapment of L5 nerve root between enlarge transverse process and sacral ala with osteophytes and bone spur formation (fig 6 &7). An excision of the abnormal growth and bone osteophytes resulted in successful relieving of radicular pain and dysesthesia in the L5 nerve distribution[16]. Unilateral or bilateral dysplastic facet joints were also found just below the transitional vertebra. Radiographical presence of degeneration at the new articulation and intervertebral joints in LSTV, in itself, may not be a source of pain generation. Metabolic activity of the region was also studied widely by using planar and SPECT bone scintigraphy. Jonsson *et al*[34] reported normal planar bone scan findings in eight patients with unilateral LSTV articulation. Pekindil G. et. al demonstrated non-focal mild uptake in eight asymptomatic LSTV cases. Symptomatic LSTV without degeneration patients showed nonfocal mild uptake; whereas symptomatic LSTV patients with radiographically noted degenerative spine revealed focal, marked uptake on SPECT bone scintigraphy. It is the degenerative spine at the pseudo-articulation site and facet joints which cast an increased uptake[35].

Management: Treatment of lumbosacral transitional vertebra has been controversial as some studies refuted its association with low back ache[21,22]. In contrast, some authors also indicated its relationship with increased lumbar degenerative disc disease at supradjacent level and pain generation at false joint between L5 transverse process and sacral ala[14,18] Presence of transition vertebra and concurrent back pain pause a great diagnostic and treatment decision making problem to the clinicians. Several authors indicated that LSTV, particularly castellvi type II and type IV potentially could be a source of low back pain[17] and should always be considered in differential diagnoses of low back pain in younger population. Reduced mobility at the L5S1 level and consequently, increased abnormal and asymmetric stress can result in early degenerative changes within the "neo-articulation" or in the normal contralateral facet joint. As far as management is concerned, it begins with conservative management as is the case with patients of LBA without LSTV. There are several small studies and case reports about successful initial management of the patients using therapeutic mobilization and physical therapy, [36] although, literature evidenced BS patients do benefit poorly if physiotherapy is used alone or in combination with anti-inflammatory and analgesic agents. Considering the multifactorial origin site of pain, it is quite difficult to pinpoint the pain generators using a radiograph and/or an MRI alone. Injection of local anesthetic agents in the potential site of pain is fairly good approach to pin point all the areas which are problematic. Also bone scan or SPECT/CT scintigraphy may potentially suggest the culprit areas. Anuj *et al* used block method to locate the pain generators in BS caltelvi type IA. In their study, after confirmation of pain site using blocks, radio-frequency ablation of the rami communicants, SI joint RF, DRG pRF and nucleoplasy were used as a definitive treatment with relief extended beyond 6 months. Out of them, neo-articulation was the worst in terms of pain relief[37] sacroilitis was also fared poorly with a radiofrequency denervation. Relief from local anesthetic and steroid injection at neo-articulation site has been variable with no consistent results. Robert c mark *et al* prospectively followed a cohort of 10 patients with severe LBP diagnosed with an LSTV on X-ray. All patients received X-ray guided injection of steroids and local anesthetic agent. Out them, 8 patients had immediate pain relief and 1 patient had total pain relief after one week of injection. 5 of them had recurrence of pain ranging from 1 day to 12 weeks. Three patients reported partial pain relief lasting from 7 to 41 months and 1 patient was pain free till 2 years after the injection[38]. Avimadje *et al* retrospectively studied 12 patients with LSTV with same side LBP and buttock pain. 11 patients received steroid injection at pseudo articulation site. Out of them 9 patient reported 50% pain relief at 1 month follow up. One patient lost to follow up. 7 out of 8 patients had improved or had no pain at 6 to 24 months later. 2 patients received 2nd injection at one and two months respectively, after the first injection. They emphasized that results of local steroid injection are also unpredictable, but still it should be considered in patients with LSTV before considering for surgical means of treatment[15]. There is paucity of literature regarding surgical treatment of LBA in association with bertolotti's syndrome. Santavirta *et al.* treated 16 patients of bertolotti's syndrome aged 27 to 58 years (mean-34) with operative treatment. Surgical methods included posterolateral fusion in a half of the patients and surgical resection of enlarged transverse process in another half. He observed that though, pain intensity in postoperative group was improved, it was slightly better than conservatively treated groups. He emphasized that operative treatment should be considered in those patients where conservative treatment has exhausted. Prior to surgical execution, disc pathology just above and below the transitional vertebra should be considered. Surgical resection of the transverse process should be considered when the pain truly arises from the neo-articulation. Conversely, posterolateral fusion may be offered to those who have disc pathology in term of degeneration, protrusion etc. at infradjacent disc level[39]. Jonsson *et al.* anaesthetized the neo-articulation first and observed considerable improvement in majority of his patients, even-though bone scintimetry using 99^{m} Tc MDP had not shown an increased uptake in most of the patients. Out of 11 patients (mean follow up of 17 months) 9 patients reported long lasting significant to complete alleviation of symptoms after surgical resection of the enlarged TP. It can be inferred that not all the neo-articulation site and subsequent degeneration their-of are painful and as a result they may show a cold spot on bone scintigraphy. They postulated that it is the hypermobility

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rectification between the rostral lumbar segment and sacrum which relieve the LBA following resection of neo-articulation[34]. Pain arising from the facet joint contra-lateral to the pseudo-joint also has been claimed. Jeffrey S. *et al* treated one case of BS by prior confirmation with injection of anesthetic agents and subsequent excision of neo-articulation site and had successful pain relief at the contralateral facet site at one-year follow-up after[40]. Minimally invasive technique also employed to resect the overgrown transverse process of L5 and reported having good pain relief in short term follow –up[41]. Li *et al* treated 7 patients of BS using minimally invasive tubular resection of overgrown L5 TP. 3 out of 7 patients reported complete relief in symptoms, 2 out of 7 reported reduced pain intensity and 2 of 7 reported initial pain relief but recurrence of pain at 1 and 4 years of surgery was observed. Almeida *et al* treated 5 patients of BS with prior infiltration of neo-articulation with anesthetic agent. 3 out of 5 got partial relief and 2 out of had significant relief. The later were subjected to surgical resection of transverse mega-apophysis and got total pain relief in the long run. He theorized the principle of low back pain care in presence of bertolotti's syndrome[42]. General norm of low backache with BS starts from conservative management in form of NSAIDS, physiotherapy and chiropractic manipulations etc. Those who do not respond to this regime are potential candidate for diagnostic and sometimes therapeutic injection of anesthetic agents at neo-articulation site or the contralateral facet joint, although, no previous studies reported any prognostic value of this procedure. The candidates who have had partial/significant pain relief from injection, may be enrolled for surgical resection of the overgrown L5 transverse process. In association with significant degeneration of contralateral transverse process, a fusion procedure at L5 and S1 in form of TLIF/PLIF is a better choice. Chang il ju *et al* studied 256 patients of bertolotti's syndrome and chose to give steroid injections at pseudo articulation in 87 case. 26 cases were excluded from study analysis because of elimination of confounding factors like presence of other spinal diseases viz. spinal stenosis, disc herniation and spondylolisthesis. Group A (39 cases) received local steroid and anesthetic agent injection at pseudo articulation site and in group B (22 cases) received selective L4 nerve root block. After confirming temporary relief, all 61 patients were subjected to L5 transverse processectomy. In group A, preoperative VAS score was 7.59± 0.93 and post-operative VAS score was 3.82± 1.59. in group B, pre-surgical average VAS score was 7.50±0.86 and post-surgical VAS score was 2.05± 1. Average follow up duration was 10 months. In their study, effective pain relief with the injection was received only in 25% cases. They emphasized that most persistent pain could originate from other site other than pseudo articulation site such as far out syndrome, foraminal stenosis, etc. In addition, they also pointed out that the LBP and radicular pain could also arise from L4 nerve compression because of overgrown L5 TP. They advocated additional decompression of L4 nerve root while bisecting the L5 transverse process[43].

Table 1: Classification of lumbosacral junction according to O'driscoll *et al*[33]

Type	Status of first sacral intervertebral disc(S1-S2)
1	No Disc material
2	A small residual disc with AP length less than that of the sacrum
3	A well-formed disc extending the entir AP length of the sacrum
4	A well-formed disc with the addition of squaring of the first sacral vertebra(S1)

Figure 1:

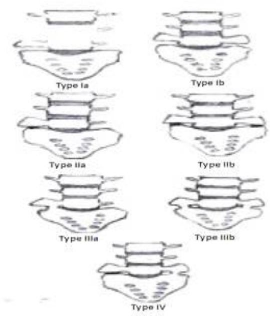

Figure 2:

Figure 3:

Figure 4:

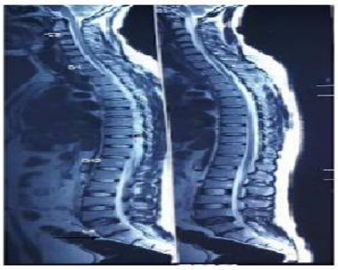

Figure 5:

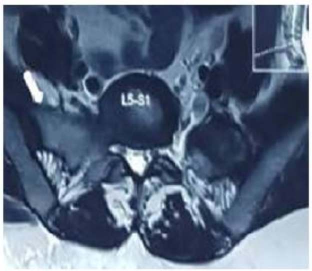

Figure 6:

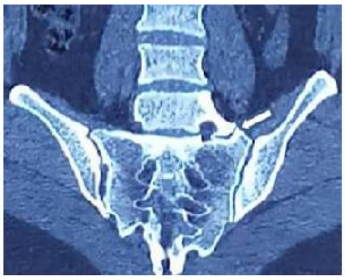

Figure 7:

Figure1: Antero-posterior pelvic radiograph with beam directed 30^0 cephalad showing unilateral neo-articulation between overgrown transverse process of L5 and sacral ala and ilium, falling under category of Castellvi type IIa, **Figure 2:** A drawing showing various types of LSTV as per Castellvi *et al*[1] **Figure 3:** castellvi type of III b with completed bilateral Fusion of enlarged TP of L5 to sacrum and ilium in an asymptomatic 26 years old manm, **Figure4:** 3D reconstructed image of Castellvi type III b, complete assimilation of L5 into sacrum in the same man as depicted in fig. 3, **Figure 5:** T2 weighted sagittal sections of MRI whole spine showing decreased L5-S1 disc space with early degeneration of superjacent disc at L4-L5 level in a 26 years old patient with Catellvi type III b LSTV, **Figure 6:** T2 weighted axial image of LS spine showing overgrown Rt side transverse process of L5, **Figure 7:** Coronal section of NC-CT scan showing pseudo-joint between TP and sacrum with sclerosis.

CONCLUSION

Bertolotti's syndrome with LBA may pose great diagnostic dilemma to a clinician. The origin of pain is multifactorial. A diagnostic search for pain points should be done using an array of investigations viz. x-ray, MRI; CT scan, SPECT and bone scan. Initial treatment starts from conservative management just as with LBA with no BS. Along with pain medications, physiotherapy should be employed. In refractory cases, diagnostic injection of steroid and anesthetic agent can be given. If the pain relief is partial and short lived, surgical means in form of resection or posterolateral fusion may be considered. A resection only fairs better in cases where facet joints are not so degenerated, while fusion surgery appears to be worthwhile approach if advanced facet joint degeneration, spondylolysis and spondylolisthesis are present at transitional vertebral level. Need of individualization of treatment appears of great deal. Further studies of higher evidence with large sample size are needed to delineate a definite management plan.

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