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Bertolotti Syndrome: Role of SPECT/CT in Distinguishing Benign Anatomical Variants from Suspicious Bone Lesions

S. E. Abaid[1]*, H. Alaoui[1], M. A. Bsiss[1], A. Matrane[1]

[1]Department of Nuclear Medicine, Hematology Oncology Centre, Mohammed VI University Hospital, Marrakesh, Morocco

***Corresponding author:** S. E. Abaid
Department of Nuclear Medicine, Hematology Oncology Centre, Mohammed VI University Hospital, Marrakesh, Morocco

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Abstract: **Case Report**

Bertolotti syndrome corresponds to a congenital lumbosacral transitional anomaly, characterized by a partial or complete articulation between the L5 mega-transverse process and the sacrum or iliac wing. Although common, it often remains underdiagnosed and can represent a true imaging pitfall, particularly in patients monitored for neoplastic disease, where any osseous uptake is initially suggestive of a secondary lesion. We report the case of a female oncology patient in whom technetium-99m bone scintigraphy revealed a suspicious lumbar focus of increased uptake. The contribution of SPECT/CT allowed correlation of the osteoblastic hyperactivity with a neo-articulation between L5 and the sacrum, thus confirming Bertolotti syndrome and ruling out metastatic involvement. This case highlights the major value of SPECT/CT in the differential diagnosis of atypical bone uptakes. By integrating morphological and functional information, SPECT/CT enables differentiation between a benign anatomical variant and a tumor lesion, thereby avoiding invasive investigations or inappropriate therapeutic decisions.

Keywords: Bertolotti syndrome, Lumbosacral transitional vertebra, Low back pain, SPECT/CT, Differential diagnosis.

INTRODUCTION

Bertolotti syndrome, first described by Mario Bertolotti in 1917, corresponds to the presence of a lumbosacral transitional vertebra (LSTV), generally characterized by an enlarged transverse process of L5 partially or completely articulating with the sacrum or iliac wing. This congenital anomaly, although relatively common, often remains underdiagnosed or unrecognized, particularly in the context of chronic low back pain [1, 2].

Studies report highly variable prevalence rates of LSTV in the population, ranging from approximately 4 % to over 30 %, depending on the population studied and the imaging modalities used [3]. Recent work has even identified a 24.9 % prevalence of LSTV in a European cohort using computed tomography [4]. Although many LSTVs are asymptomatic, the pathophysiological link between LSTV and low back pain suggests several mechanisms, including degenerative changes at the pseudo-articulation, hypermobility at the lumbosacral junction, and abnormal mechanical stress leading to pain [5].

Diagnosis of these anomalies can be challenging: conventional morphologic techniques (radiography, CT) show the bony structure but do not always allow differentiation between benign mechanical fixation and secondary lesions, particularly in patients with neoplastic disease. This

diagnostic challenge may lead to misinterpretations and inappropriate management.

Single-photon emission computed tomography combined with computed tomography (SPECT/CT) provides a decisive functional-clinical dimension by correlating tracer uptake (indicating bone activity) with precise anatomical structures. This modality increases diagnostic specificity, allowing differentiation of benign fixations, such as those associated with Bertolotti syndrome, from true pathological lesions.

In this context, we present the case of an oncology patient in whom SPECT/CT allowed reclassification of a lumbar focus of increased uptake, initially suspected, as benign Bertolotti syndrome, highlighting the importance of this approach in the differential diagnosis of bone anomalies in patients with neoplastic disease.

Case Report

We report the case of a 51-year-old female patient, followed for infiltrating ductal carcinoma of the breast as part of a staging workup, who was referred to the Nuclear Medicine Department of the Mohammed VI University Hospital Center in Marrakech. Collected data included age, sex, neoplastic history, any clinical symptoms, and results from scintigraphic and morphologic imaging studies. Bone scintigraphy was performed using a Symbia Pro.specta X3 SPECT-CT gamma camera (Siemens), including whole-body planar acquisitions, followed by thoraco-abdomino-pelvic SPECT/CT (TAP) two hours after intravenous injection of 740 MBq (20 mCi) of 99mTc-MDP.

Planar acquisitions (Figure 1) revealed multiple foci of increased uptake, including a focal lumbar uptake initially considered suspicious due to the oncologic context. This osseous hyperactivity appeared localized at L5, but planar images alone could not determine whether it corresponded to a metastatic lesion or a benign osteoarticular anomaly. Prior morphologic imaging had not clearly visualized the transverse megaprocess of L5 or the corresponding neo-articulation with the sacral ala. The observed osseous changes were nonspecific and did not exclude the possibility of a secondary lesion.

SPECT/CT – TAP acquisition (Figure 2) enabled more precise characterization of this focus. Fusion slices demonstrated that the lumbar uptake corresponded to a left L5 transverse megaprocess contacting the sacral ala, forming a neo-articulation typical of Bertolotti syndrome. The associated CT revealed localized degenerative changes, including mild cortical thickening, articular irregularities, and reactive sclerotic changes. The concordance between morphological anomalies and scintigraphic hyperfixation confirmed the mechanical, non-tumoral nature of the uptake.

Additionally, SPECT/CT allowed differentiation of other foci observed on planar images. Uptake in the sternum, D7, and L1 corresponded to previously identified osteolytic or mixed lesions on CT, consistent with secondary osseous localizations. Diffuse degenerative-appearing uptake, notably at the shoulders, knees, and right mandible, exhibited typical benign features without ambiguity.

In this case, SPECT/CT played a decisive role by discriminating an atypical lumbar focus that appeared suspicious on planar images alone, enabling a definitive diagnosis of Bertolotti syndrome.

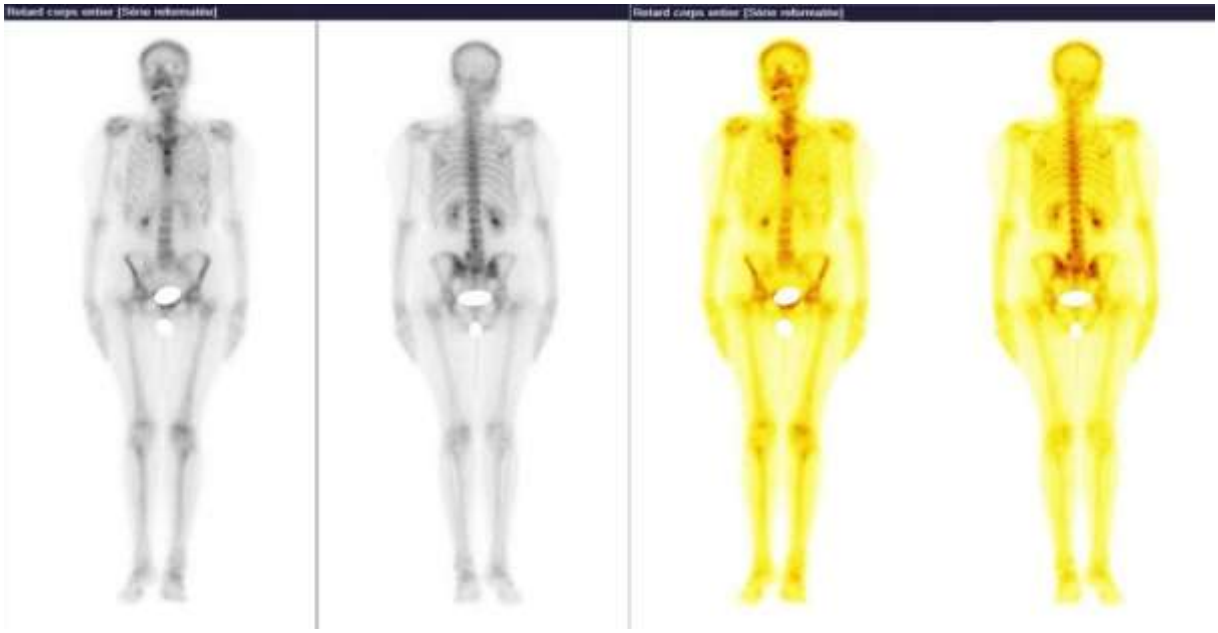

Figure 1: Whole-body planar image in anterior and posterior views

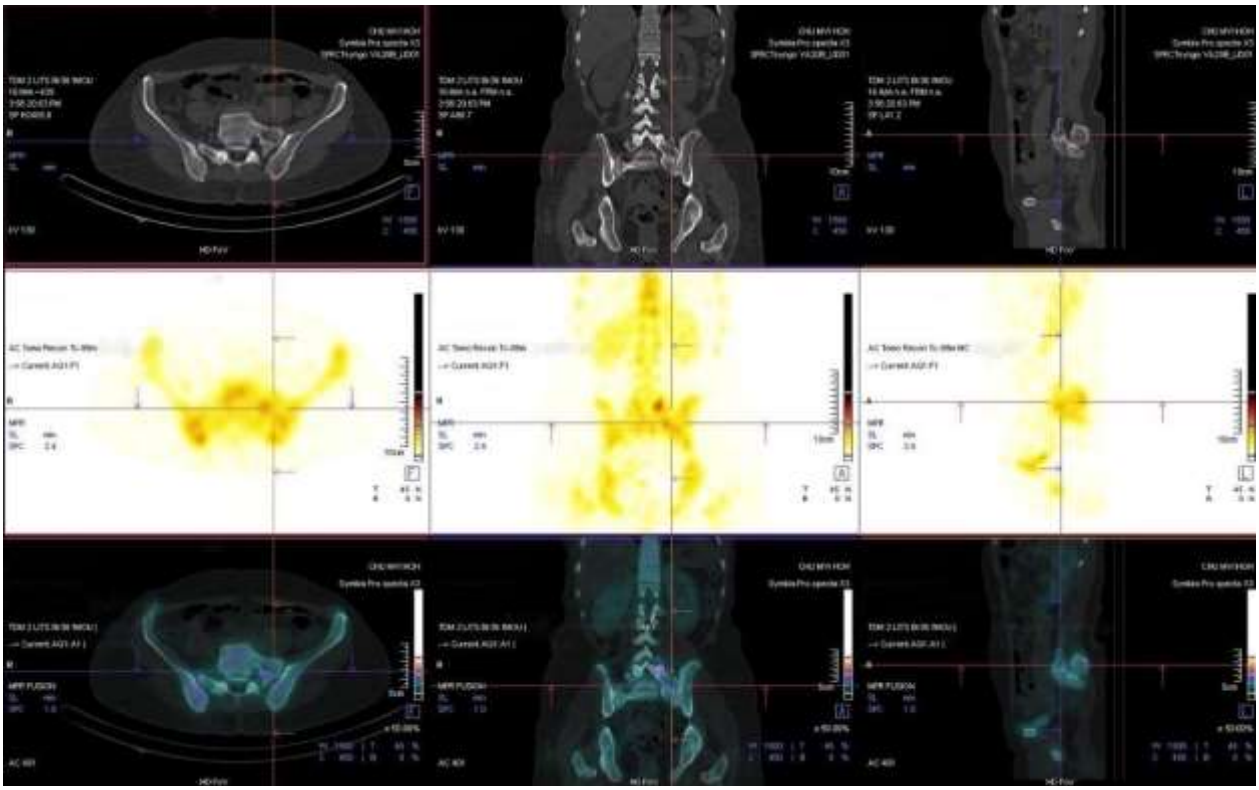

Figure 2: SPECT/CT acquisition of the lumbar spine and sacrum

DISCUSSION

In this case report, SPECT/CT allowed precise characterization of a suspicious lumbar hyperfixation focus, demonstrating that it corresponded to Bertolotti's syndrome. This diagnosis is particularly relevant, as lumbosacral transitional vertebrae (LSTV) are common and can pose an imaging challenge, especially in oncological contexts where any osseous hyperactivity is immediately suspected to be metastatic.

Several studies have shown that the prevalence of LSTV in the general population is highly variable, likely due to differences in imaging modalities and populations studied. A

European study using computed tomography reported an LSTV rate of 24.9%, and highlighted that certain types, particularly according to Castellvi's classification, are associated with more pronounced functional impairment [6]. Similarly, literature reviews indicate that LSTVs alter spinal biomechanics, especially at pseudo-articulations, potentially promoting degenerative changes [3].

The association between LSTV and low back pain is not systematic, but sufficiently documented to justify thorough investigation. In a cohort analyzing sacroiliac joint MRIs, Ten *et al*., reported a significant prevalence of symptomatic patients with various Castellvi-type LSTVs, suggesting that this anomaly may be an underestimated cause of low back pain [7].

Regarding functional imaging, SPECT/CT appears to be an extremely useful complementary tool. As reported in the literature, SPECT/CT can accurately localize metabolic stress zones at pseudo-articulations or transverse processes, which is not always evident on radiographs or CT alone [8, 9]. In our case, the correlation between hyperfixation and bone morphology confirmed that the anomaly was mechanical rather than neoplastic. Multiple studies emphasize the growing importance of functional imaging in diagnosing Bertolotti's syndrome. Al Riyami *et al*., reported a case of chronic low back pain in which SPECT/CT identified a pseudo-articulation between the L5 transverse mega-apophysis and the sacrum, showing metabolic hyperactivity, allowing successful micro-endoscopic resection of the pseudo-articulation [10].

Clinically, this distinction is crucial. In oncological contexts, any osseous hyperfixation raises concerns for metastasis, potentially leading to over-investigation, unnecessary biopsies, or inappropriate treatments. Recognition of benign Bertolotti syndrome via SPECT/CT not only avoids these procedures but also guides symptomatic management if needed. In surgical contexts, the presence of an LSTV is not negligible, a study demonstrated that among patients undergoing lumbosacral surgery, the prevalence of LSTV was very high (46.2%), with an increased association of degenerative pathologies such as spinal stenosis and spondylolisthesis [11].

Finally, our observation underscores the need for increased awareness of this anatomical and clinico-functional entity. Although Bertolotti's syndrome is not always symptomatic, its potential for diagnostic error in imaging should prompt nuclear medicine, radiology, and oncology practitioners to consider it, particularly when scintigraphic findings are atypical.

CONCLUSION

Bertolotti's syndrome represents a common but often underrecognized cause of chronic low back pain, and its diagnosis relies on a multimodal approach integrating both morphological and functional imaging. In our case, bone scintigraphy combined with SPECT/CT proved particularly valuable in highlighting osteoblastic hyperactivity at the pseudo-articulation, thereby confirming the anatomical variant. The functional information provided by this modality offers a decisive advantage over morphological techniques alone, especially in stratifying patients who may benefit from interventional or surgical management. Literature data further support the value of SPECT/CT for detecting LSTVs and emphasize the frequent association of these anomalies with lumbosacral degenerative pathologies.

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Declaration

Conflict of Interest: All the authors (Saad Eddine Abaid, Hamza Alaoui, Mohammed Aziz Bsiss, Aboubaker Matrane) declare that there is no confict of interest that could be perceived as prejudicing the impartiality of the research reported.

Informed consent: Written informed consent was obtained prospectively from the patient to use their clinical data.

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