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Prevalence of sacral spina bifida occulta with lumbosacral transitional vertebra in a skeletal collection of a South African population

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ABSTRACT

In this study, complete Sacral Spina Bifida Occulta (SSBO) and Lumbosacral Transitional Vertebrae (LSTV) with their various subtypes based on the Castellvi classification were appraised in a South African population sample. Adult human skeletons representing the three largest South African population groups, namely South African African, South African of Mixed Ancestry; South African of European descent; both biological sexes; and age range between 21 and 90 years at death were evaluated for both spinal anomalies.

The number of preselected skeletons (n = 1798) from the largest collection in Africa of modern human skeletons (N = 2630) provided a strong representative South African sample. The observational study looked at lumbar and sacral vertebrae in each skeleton and any anomalous features were captured in an Excel spreadsheet.

Complete SSBO with no LSTV was observed in eight subjects (8/1798; 0.44 %), while incomplete SSBO was observed in one subject (1/1798; 0.06 %), and one individual (1/1798; 0.06 %) exhibiting complete SSBO with LSTV (Type IIIB, Castellvi classification). No significant differences were observed when comparing the prevalence of the LSTV with SSBO in the male and female groups of the different population affinity groups. The number of individuals with SSBO was found in more South African Africans than in the other population groups with no significant difference between groups.

This research revealed the simultaneous presence of two morphological anomalies (SSBO and LSTV) at the same vertebral segment that could result in low back pain. Variant morphology awareness is crucial for clinicians across all modalities to prevent misdiagnosis, leading to better treatment plans, and avoiding injury.

1. Introduction

The prevalence of a structural anomaly or variations of the vertebral column elements may have a clinical impact on a patient. Any particular vertebra may exhibit an anomaly in a part of the bone or a particular vertebral region. The deficiency of a particular part of a bone could reflect and present itself as specific signs and symptoms in an individual. These vertebral anomalies originate from neural tube defects (NTDs) during fetal development and may also be disabling in some patients [1–3]. In this study, the prevalence of two vertebral anomalies, namely Sacral Spina Bifida Occulta (SSBO) and Lumbosacral Transitional Vertebra (LSTV), were appraised. Spina bifida represents the failure in developing components associated with the neural tube and its coverings in the posterior elements of the spine [4]. Data from animal models suggests that spina bifida and anencephaly can result from disturbances in cell adhesion or alterations in neural plate shaping or bending that prevent the apposition of the neural folds [5].

Smith [4] similarly defined Spina bifida and identified three types of Spina bifida, viz. (1) rachischisis, which is the most severe type that exhibits an incomplete closure of the neural canal, the everted Iining of the neural tube forming a reddish band down the middle of the back; (2) spina bifida cystica which is sometimes referred to as “spina bifida”, which shows a herniation of central nervous system elements through a vertebral arch defect; and (3) spina bifida occulta (SBO) is considered to be the mildest type of vertebral defect, without any observed external malformation of the spinal cord, associated nerves, and their coverings [4]. The different NTDs were identified to be influenced by genetic and environmental factors [2,5].

Several studies looked at spina bifida and the clinical implications of this anomaly on pediatric, adolescent, and adult patients, and these

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ranged from mild low back pain (LBP), sexual dysfunction, motor or sensory loss and enuresis [6–9].

Infants may present with a cutaneous dimple (usually associated with a tuft of hair) in the gluteal and lower back regions, with extreme presentation representing a large cystic protuberance (Fig. 1) [10].

Spina bifida occulta (SBO) is a neural arch cleft defect that occurs in one or more levels of the lamina or the spinous processes of vertebrae and is usually found in the lumbosacral region, thus classifying SSBO affecting the sacrum with different degrees of spinous process non-fusion [11]. SBO is generally considered the mildest form of spina bifida [2,12]. Early cases of SBO have been reported in the literature with patients exhibiting pain in the low back and lower limb weakness or pain [7,13–17]. In some instances, these were determined to be co-incidental [18]. SBO occurs due to the non-fusion of the posterior elements of the vertebral arch [19]. Unlike more severe forms of spina bifida, it has no neurological pathological condition resulting from a prior disease, injury, or attack. It is usually asymptomatic, and although this anomaly can occur at any level of the vertebral column, the malformation of the last lumbar vertebra and the first sacral vertebra is the most routinely observed, studied, and reported [20].

It is also known that different sacral levels exhibit arch non-closure [16,21–23]. SSBO has also been classified by looking at the particular sacral vertebra level and the combination of levels affected by the non-closure of the vertebral arch.

On the other hand, a Lumbosacral Transitional Vertebra (LSTV) refers to the dysplastic changes of the transverse process with incorporation of the last lumbar vertebra into the sacral complex (sacralization) or the first sacral vertebra into the lumbar complex (lumbarization), with various types based on the Castellvi et al. classification [24].

LSTV as described by Castellvi et al. [24], classification (Fig. 2) contains 4 distinct morphological Types. These variations present with a unilateral presentation denoted as ‘A’, either right or left, and bilateral presentation denoted as ‘B’.

- Type I represents an enlargement of the transverse vertebral process (TVP), unilateral right or left or bilateral presentation. Type I morphology demonstrates lack of clinical relevance [24–27].
- Type II represents an enlargement of the TVP with contact to the ala of the sacrum, unilateral right or left, or bilateral presentation.
- Type III represents an osseous fusion of the TVP to the ala of the sacrum, unilateral right or left, or bilateral presentation.
- Type IV represents bilateral enlargement of the TVP of the last lumbar vertebra with a unilateral articulation/contact with the ala of the sacrum on one side and fusion of the lumbar TVP to the ala of the sacrum on the contralateral side.

As happened with SSBO, LBP has been associated with LSTV presence [29], and various studies have emphasized the need for an accurate diagnosis and management of LSTV [30].

Thus, SSBO and LSTV can be related to LBP, and it is known that at some point during their lives, between 70 % and 85 % of the population will experience an episode of LBP for over three months [31]. LBP is well-documented as the leading cause of disability and sick leave in most industrialized countries, especially when it becomes a chronic presentation [32,33]. It has been estimated that between 5 % and 10 % of acute LBP cases will develop into chronic LBP [33]. Chronic LBP is defined as the presence of pain on at least half of the days in the last six months, explicitly allowing short periods without pain during those six months [34]. Chronic LBP is one of the major causes of productivity loss and disability (permanent, partial, or complete inability to work) in the world of work in Western countries [35].

In this context of the relationship between SSBO, LSTV, and LBP, the visual examination of human skeletons provides a unique opportunity to discover and describe the prevalence of SSBO and LSTV and their possible simultaneous presence in the same subject since such cadaveric-derived studies are usually considered the gold standard for assessing the presence and the anatomical characteristics of anatomic variations of the vertebrae [36].

Following this, the present study aimed to carry out large-size research in order to detect the presence of SSBO and LSTV in a South African population sample because of their possible causal relationship between LBP, SSBO, and LSTV. Official reports of the United Nations [37] revealed that sub-Saharan Africa is projected to become the most populous of the eight geographic regions in the late 2060s, surpassing both Eastern and South-Eastern Asia and Central and Southern Asia in size. A precise knowledge of SSBO and LSTV may prevent surgical complications and serve to improve the treatment of patients.

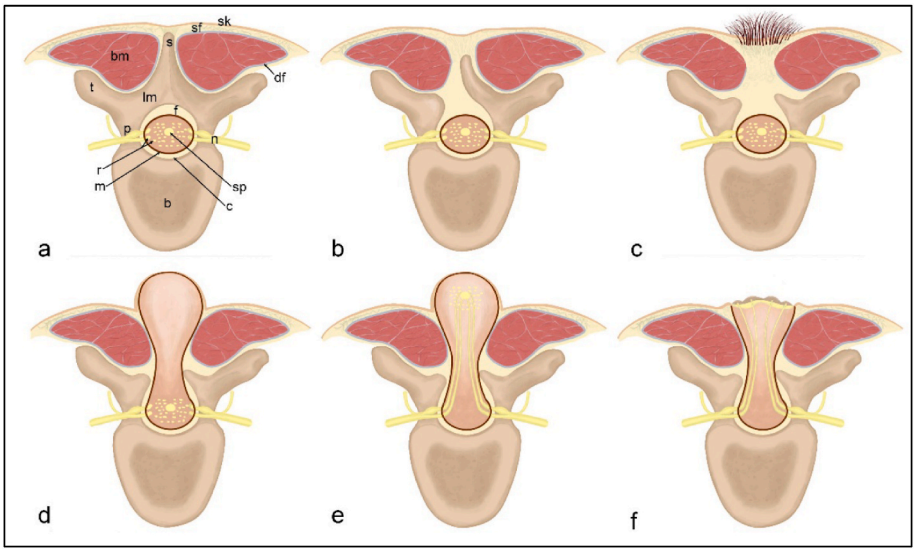

Fig. 1. Transverse views of the patterns of lumbosacral spina bifida. a. Normal lumbosacral anatomy (L3), b. Spina bifida occulta resultant from agenesis of posterior vertebral arch and little pit on the covering skin, c. Spina bifida occulta (complete posterior arch defect) showing hair tuft, d. Meningocele, e. Myelomeningocele and f. Myeloschisis (rachischisis). (Abbreviations for labels in diagram “a”: b, vertebral body; bm, back musculature; c, vertebral canal; f, fibroadipous tissue in vertebral canal; lm, Vertebral lamina; m, meninges; n, spinal nerve; p, Vertebra pedicle; r, spinal nerve roots; sf, superficial fascia; df, deep fascia; sp, spine; s, spinous process; sk, skin; t, transverse process). Adapted from [10].

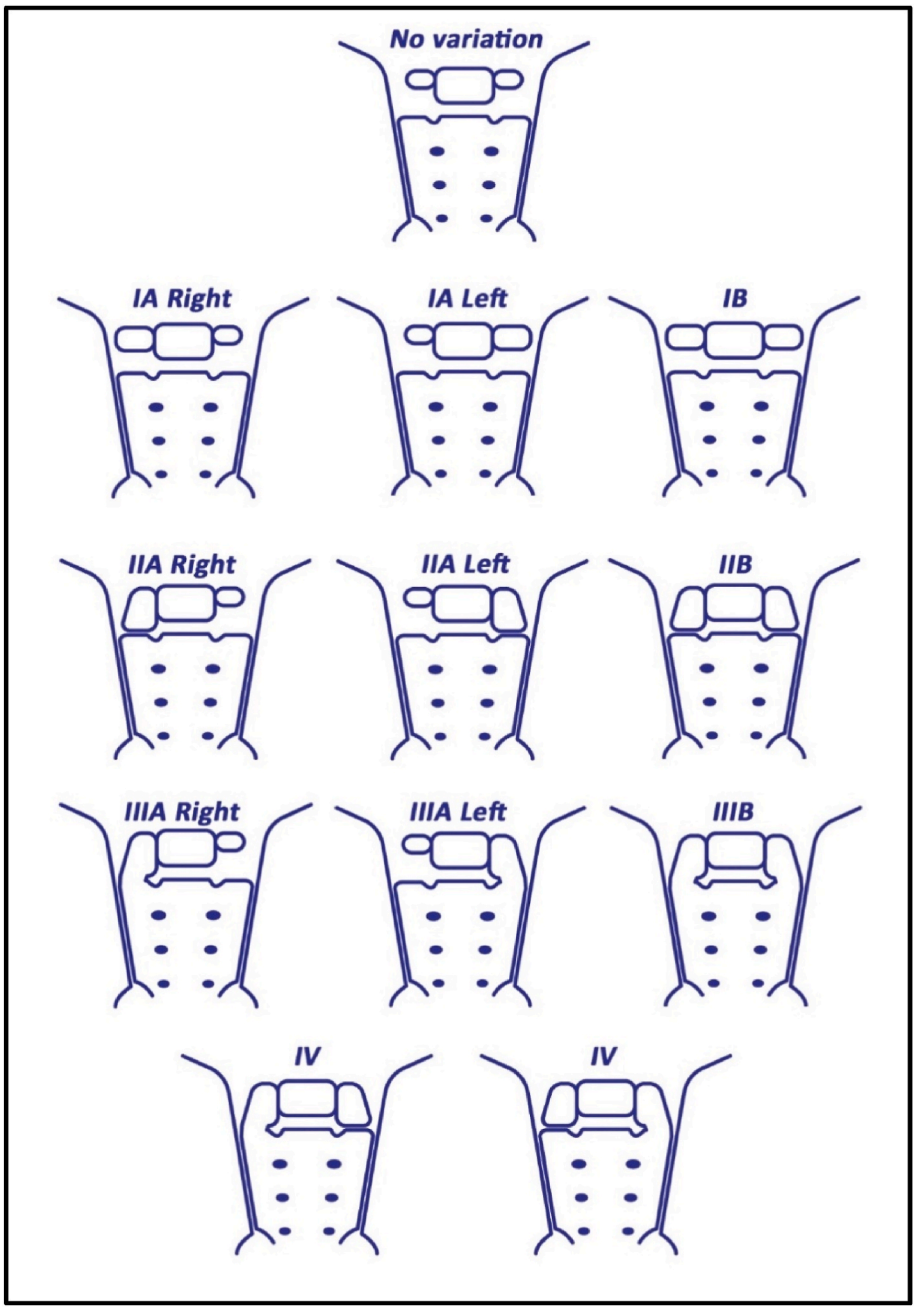

Fig. 2. Castellvi et al. [24] classification after Paton et al. [28], Top to bottom: No variation represents no change in transverse process TVP height. Type I (included for illustrative purposes only). Types II, II, and IV.

2. Material and methods

This study investigated the prevalence of SSBO and LSTV in a South African population sample through the appraisal of adult human skeletons (n = 1798), thus representative of the three largest population groups, both sexes, across a broad age range from the Raymond A. Dart Collection of Modern Human Skeletons (hereto referred to as the Dart Collection), School of Anatomical Sciences, University of the Witwatersrand, South Africa. The Dart Collection is composed of modern human skeletons primarily derived from bodies that were deemed to be "unknown" and more recently, from "donor bodies" that undergo dissection by students registered for medical and allied medical offerings at the Medical School. Each skeleton is cataloged and has its associated demographic information, and this facilitates research on the materials from this collection. The Dart Collection of skeletons was used because of their known Age at Death (AD), biological sex, and population affinity, making it a special South African, African, and world reference anatomical skeletal collection [38].

This large number of preselected skeletons (n = 1798) from one of the largest collections of modern human skeletons in South Africa (N = 2630) provides a representative South African sample. The margin of error was 1.21 % (n = 1798; N = 2630; osteological population proportion: 68 %; Confidence level = 95 %). Table 1 reflects the breakdown of the population sample used in this study.

The Population affinity of the sample (Table 1) and terminology associated with the research sample population is based on the comprehensive description of and the currently accepted nomenclature (South African African, South African of Mixed Ancestry, South African of European Descent) of the samples from the Dart Collection [38,39].

Table 1
The population sample used in this study with the indicated prevalence of the SSBO and LSTV per population group and biological sex.

POPULATION AFFINITY	SUB-TOTAL	SEX	SEX DISTRIBUTION	COMPLETE SSBO	INCOMPLETE SSBO
South African African	1515 (84.30 %)	Male	1135 (74.85 %)	4	0
		Female	380 (25.10 %)	1	0
South African of Mixed Ancestry	103 (5.72 %)	Male	61 (59.80 %)	1	0
		Female	42 (41.20 %)	0	0
South African of European Descent	180 (10.01 %)	Male	111 (61.60 %)	3[a]	1
		Female	69 (38.40 %)	0	0
		Total Male	1257 (69.90 %)	8	1
		Total Female	541 (30.10 %)	1	
TOTAL (N)	**1798**			**9**	**1**

[a] Coexistence of Complete Sacral Spina Bifida Occulta with Lumbosacral Transitional Vertebra.

2.1. Inclusion and exclusion criteria of the population sample

Inclusion: three largest South African population groups, both biological sexes, age range of above 21 years AD; intact lumbar and sacral elements.

Exclusion: Any specimens that showed pre- and post-mortem damage or pathology to the lumbar vertebrae and the sacrum were excluded from the study.

Spina bifida is identified as the absence of fusion of the laminae and the spinous processes of vertebrae. Complete SSBO was identified as the absence of fusion of the spinous processes of all the sacral vertebrae. Incomplete SSBO was classed as any absence of the individual sacral vertebra spinous processes that interrupts the formation of the median sacral crest. To identify the different types of LSTV, the Castellvi classification was used to determine the variant present specifically.

Descriptive statistics as well as the Chi-squared test ($\chi 2$) and Z-test for proportions were used for the data analysis. Statistical analysis was done utilizing IBM SPSS Statistics (Version 27).

3. Results

The average AD for the entire sample group was 45.10 years (standard deviation of 12.23 years; n = 1798). The minimum AD was 21 years, and the maximum was 90 years old for both male and female cohorts. The average AD for the male cohort was 45.63 years (standard deviation of 11.81 years). The average AD for the female cohort was 43.62 years (standard deviation of 13.18 years).

The indicated prevalence (Tables 1 and 2) of the anomalous sacral vertebral anatomy was in 10 individuals (10/1798 = 0.56 %) with complete (9/10 = 90 %, Fig. 3a, 4a and 5b) and incomplete/partial SSBO (1/10 = 10 %, Fig. 3b). Complete SSBO was noted in five South African Africans (four males and one female) and in three male South Africans of European Descent. Only one male of South African of Mixed Ancestry showed complete SSBO. An incomplete SSBO was noted in one male South African of European Descent.

Notably, one individual (1/10 = 10 %; January 1798 = 0.06 %) exhibited complete SSBO with LSTV (Type IIIB, Castellvi classification; one male South African of European Descent aged 88 years old; Fig. 5a).

Table 2
Details of the prevalence in the population sample studied. Individuals with Sacral Spina Bifida Occulta in the population sample studied.

POPULATION AFFINITY	SEX	AGE	SSBO PREVALANCE
South African of Mixed Ancestry (Indian)	Male	60	Complete
South African African	Male	40	Complete
South African African	Male	34	Complete
South African African	Female	20	Complete
South African African	Male	26	Complete
South African of European Descent	Male	38	Complete
South African African	Male	60	Complete
South African of European Descent	Male	51	Incomplete
South African of European Descent	Male	54	Complete
South African of European Descent[a]	Male	88	Complete

[a] Coexistence of Complete Sacral Spina Bifida Occulta with Lumbosacral Transitional Vertebra.

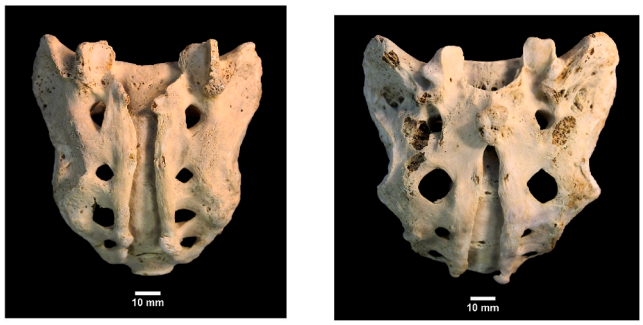

Fig. 3. Sacra that show (a) complete SSBO with no LSTV (South African African (SAA) male 60-year-old) and (b) partial/incomplete SSBO with no LSTV (South African of European Descent (SAED) male 51-year-old) in posterior view.

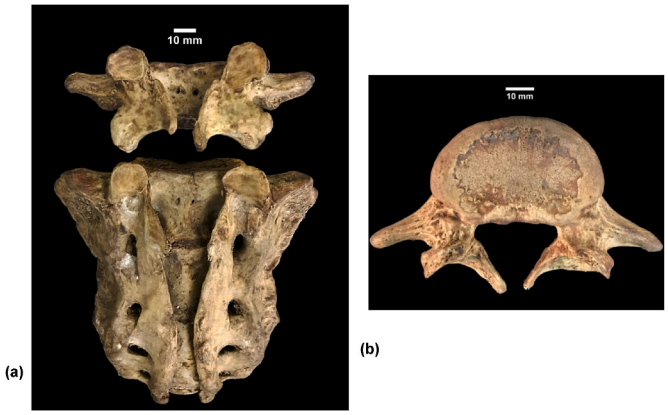

Fig. 4. Lumbosacral spina bifida occulta. An individual (South African African (SAA) male, 26 years old) with (a) complete SSBO (posterior view) and (b) associated separate L5 vertebrae (superior view) also exhibiting an unfused vertebral arch.

Fig. 4a shows an individual with complete SSBO and SBO of the L5. No LSTV was observed in this individual.

Fig. 5a shows an individual exhibiting complete SSBO with LSTV and the complete absence of any fused median sacral crest/spinous processes.

No significant differences were observed when comparing the prevalence of the LSTV with SSBO in the male and female groups of the different population affinity groups (Chi-squared test; $\chi 2 = 6.5636$; $p = 0.248$) (Table 2).

The number of individuals with complete SSBO was found to be more

Fig. 5. Sacra exhibiting SSBO with LSTV (posterior view). Note that the vertebral arch of the incorporated L5 is (a) a complete spinous process (88-year-old South African of European Descent (SAED) male) and (b) an incomplete fused spinous process (60-year-old South African Africans (SAA) male).

prevalent in the South African African population and this was not of significant difference (Chi-squared test; $\chi 2 = 6.5636$; $p = 0.211$).

4. Discussion

Here, the researchers present the prevalence of complete SSBO and LSTV in a large sample of cadaveric vertebral columns within the South African population in an observational descriptive study. A skeletal collection represents populations without the need to perform radiologic examinations for the in vivo SSBO and LSTV detection. In addition, radiologic studies generally have a bias towards subjects with clinical symptoms. However, the asymptomatic population that contain SSBO and LSTV may be at risk of complications, if not detected before lumbar and sacral spine surgery or regional anesthesia. Also, it is of interest for manual practitioners such as chiropractors, osteopaths, physiotherapists to be cognizant the prevalence of SSBO and LSTV because it may necessitate altering therapeutic techniques. The techniques include spinal manipulation, dry needling or interferential current on patients' sacral and lumbar areas, to avoid aggravation of back and gluteal pain. Furthermore, LSTV has been linked to restricted motion of the lumbosacral junction with adjacent segmental degeneration and would be important for clinical decision-making for manual practitioners [24,40,41]. In South Africa, the prevalence of LSTV has been found to be 10 % of the population [42].

Previously, SSBO presence in 27 % of the 10,000-year-old burial comprising Ibero-Maurusian skeletons was studied [43]. Henneberg and Henneberg [44] studied the frequency of SSBO in the past population of Pompeii (AD 74), with results showing that the overall frequency of 11 % falls within the ranges reported for other populations. Different frequencies were also observed at different sacral vertebral levels, ranging from 3 % (S1 to S5) to 47 (S5 only). Albrecht [45] tabulated a summary of findings by other studies on the incidence of spina bifida occulta of the sacrum as differently classified at the time, with observed frequencies of prevalence ranging from 1 % to 27 % in the different populations of the different studies. The incidences of SSBO were found to be between 4 % and 34.5 % [17,46–51].

Singh [52] compared different types of SBBO in relation to frequency and percentages between Australian and Indian Populations and found equivalence between the populations studied.

Bennett [22] found a high frequency of SSBO with LSTV (referring to the "sacralization" of L5 and therefore identified "L6") in a "proto-historic" population of Modoc Indians with a 90 % prevalence of the 33 skeletons studied. Barkley [53] found five skeletons with sacralization, with one individual showing a unilateral right fusion with SSBO and four individuals showing complete fusion.

Taskaynatan et al. [54] report an incidence of lumbosacral malformations in 88 of 881 patients studied. Of these, two individuals exhibited SBO with LSTV (0.2 %). George et al. [55] reported on LSTV with SSBO observed on a dry bone sacrum of a Caucasian ("White") male and highlighted the importance of awareness of this anomaly. Sharma et al. [56] observed a higher prevalence of the co-existence of SBO and LSTV in patients suffering from LBP.

SBO is a more frequent anomaly observed in the lumbosacral region of the vertebral column, as noted by Li et al. [57], with a 61.49 % (93/148) incidence in their study, with observation at variable vertebral levels ranging from 89 % (83/93) at S1 to 2 % (2/93) observed between S1–S4. Tamas-Csaba et al. [58] noted that SBO was more frequently localized to vertebra S1, especially in females.

Morimoto et al. [59] looked at SBO and its association with other vertebral column anomalies. They suggested a connection between spinal irregularities and spondylolysis and SBO, potentially shedding light on how spondylolysis develops and enhancing its treatment and outlook. Recognizing that individuals with SBO of the lumbar spine possess a higher chance of having a spinal anomaly could aid in averting mistakes in spinal surgery related to alignment.

Sharma et al. [56] observed that patients presenting with lower back pain also exhibited the presence of SBO at different lumbar and sacral levels together with LSTV. Of the 27 cases with SBO and LSTV (according to the Castellvi classification) reported in their study, they observed that three patients (11.1 %) had Type I, 18 patients had Type II (66.6 %), four patients had Type III (14.8 %), and 2 (7.4 %) patients had Type IV, respectively. SSBO was observed in 10 of the 27 cases (37 %), SBO at L5 in 12 cases (44 %), and SBO at S1 in 5 (19 %) reported cases. Taskaynatan et al. [54] reported a prevalence of 2 individuals of the 881 patients (0.2 %). Nevertheless, this current study shows a very low prevalence of SSBO combined with LSTV compared to other studies (Table 3).

Bertolotti's syndrome remains a controversial topic of discussion with it being both disputed and supported in the literature [24,40,60–75]. Bertolotti [29] described concomitant findings of an LSTV and LBP and coined the term 'Bertolotti's syndrome'. The prevalence of Bertolotti's syndrome is 4.6 % in the general population and 11.4 % in patients under 30 [61]. A study of 8280 patients seeking care for LBP found that 10.6 % had LSTV types II, III, or IV, with 5.3 % of cases being sacralization and 5.3 % being lumbarization [27]. The prevalence of LSTV in patients seeking care for LBP ranges from 4.6 % to 35.6 % [60,62]. It is theorized that pain generation associated with LSTV predominately affects the site of accessory articulation, namely the Type II and IV morphology, as defined by Castellvi et al. [24]. This is believed to result from abnormal motion and weight bearing, leading to degeneration of the accessory articulation. Interestingly, the literature supports unilateral Type III morphology, which may develop stress fractures due to changes associated with mechanical stress applied to the unilateral site of fusion between the LSTV and the sacrum [75]. Over time, it is understood that Bertolotti's syndrome is now considered multifactorial in etiology as varying causes may be present at one or multiple locations

Table 3
Summary of past studies conducted on SSBO showing prevalence (presented as a percentage).

RESEARCHER	YEAR	COUNTRY	TOTAL POPULATION	PREVALENCE (%)
Ali et al.,	2019	Pakistan	200	43.50 %
Singh	2013	India	140	60.71 %
Wu et al.,	2009	China	203	28.10 %
Albrecht, Scutter, and Henneberg	2007	Australia	53	100 %
Henneberg and Henneberg	1999	Italy		11.00 %
Schweitzer et al.,	1993	Unknown	246	14.63 %
Avrahami et al.	1994	Israel	1200	17.25 %
Fidas et al.,	1987	Scotland	2064	31.15 %
Ferembach	1963	Morocco	180	27.00 %

around the LSTV segment [68,76]. Sites of pain generation may include zygapophysial joint arthrosis, intervertebral disc degeneration and herniation, nerve root irritation, bone marrow edema, central or foraminal stenosis and sacroiliitis [63,68,71,74,77–82]. Nevertheless, as a limitation, our skeletal research did not detect the possible clinical symptoms associated with SSBO and LSTV.

SBO is evident in 97 syndromic conditions and is frequently observed in individuals without syndromes who present with LBP (typically occurring in the L5 region). Kamanli and Gerc [83] observed that SBO and LSTV were present together in eight healthy individuals of a population of 503 individuals studied, thus representing a prevalence of 1.6 %. LSTV, a congenital anatomical variation of the last lumbar vertebra, is prevalent among individuals without syndromes and those with syndromic conditions, often associated with mutations in the HOX gene. When dealing with syndromic conditions, the initial diagnosis often relies on clinical and phenotypic characterization, complemented by a comprehensive skeletal assessment that includes the lateral skull, AP spine, lateral lumbar, AP pelvis, and, in some instances, AP hands. In a broad sense, vertebral fusion is a recurring phenomenon across many syndromic conditions.

5. Conclusion

The researchers have presented the prevalence of SSBO and LSTV in a large sample representative of the South African population, revealing the simultaneous presence of two malformations at the same vertebral segment that may result in LBP and stiffness. This study is the first skeletal study that researched the prevalence of complete SBO in the sacral region of the vertebral column coupled with LSTV. Previous studies did not look independently at the sacral region but instead as only SBO generalized to the vertebral column, typically lumbar vertebrae. Thus, clinicians must be aware that some patients can present more than one anatomical variant or anomaly in the lumbosacral spine. This will, therefore, require then a structured diagnosis to ensure that any multiple anomalous vertebral observation is factored into an associated treatment plan.

Limitations of this study

As an osteological descriptive study, it does not allow for first person accounts of their possible signs and symptoms of their anatomical anomalies.

There is bias towards male individuals due to the composition of the skeletal repository housing more males than female (71 % vs 29 %) [38].

Availability of data and materials

The datasets used and/or analyzed during the current study are available from the corresponding author upon reasonable request.

FUNDING

Statement: No financial support was received for the study, and all authors declare no conflict of interest-associated biases.

FUNDING Statement

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Ethics Statement

This study was approved by the University of Cape Town, Faculty of Health Sciences, Human Research Ethics Committee, approval number HREC REF: 195/2017. The Raymond A Dart Collection of Modern Human Skeletons is a curated skeletal remains collection with restricted access to only approved projects. The research proposal for this study was submitted and Ethics approval for collection usage was received from the Dart Collection curator through the Access Committee of the School of Anatomical Sciences, University of the Witwatersrand, South Africa. The procedures used in this study adhere to the tenets of the Declaration of Helsinki.

CRediT authorship contribution statement

Shahed Nalla: Writing – review & editing, Writing – original draft, Validation, Resources, Project administration, Methodology, Investigation, Formal analysis, Data curation, Conceptualization. **Juan A. Sanchis-Gimeno:** Writing – review & editing, Validation, Methodology, Investigation, Formal analysis. **Glen J. Paton:** Writing – review & editing, Validation, Resources, Methodology, Formal analysis, Data curation, Conceptualization.

Declaration of competing interest

We declare to have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper. The authors declare that they have no conflict of interest.

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Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.tria.2024.100307.

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