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MUSCULOSKELETAL IMAGING

ORIGINAL ARTICLE

Sacroiliac joint involvement in osteochondromatosis: identifying its prevalence and characteristics from cross-sectional imaging

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PURPOSE

Apart from a few case reports, sacroiliac joint (SIJ) involvement in osteochondromatosis has not been studied. We aimed to determine the prevalence and characteristics of such involvement using cross-sectional imaging.

METHODS

In this retrospective study, three observers (one junior radiologist and two musculoskeletal radiologists) independently reviewed computed tomography (CT) or magnetic resonance imaging (MRI) of patients in our database who had osteochondromatosis (≥2 osteochondromas across the skeleton) for SIJ involvement. The final decision was reached by the consensus of the two musculoskeletal radiologists in a later joint session.

RESULTS

Of the 36 patients with osteochondromatosis in our database, 22 (61%) had cross-sectional imaging covering SIJs (14 females, 8 males; age range 7–66 years; mean age 23 years; 13 MRI, 9 CT). Of these, 16 (73%) had intra-articular osteochondromas. For identifying SIJ osteochondromas on cross-sectional imaging, interobserver agreement was substantial [κ = 0.67; 95% confidence interval (CI): 0.34, 1.00] between the musculoskeletal radiologists and moderate (κ = 0.59; 95% CI: 0.23, 0.94) between the junior radiologist and the final consensus decision of the two musculoskeletal radiologists. In the cohort with cross-sectional imaging, the anatomical variations of the accessory SIJ (n = 6, 27%) and iliosacral complex (n = 2, 9%) were identified in six different patients with (n = 2) and without (n = 4) sacroiliac osteochondromas.

CONCLUSION

Cross-sectional imaging shows frequent (73%) SIJ involvement in osteochondromatosis, which, although a rare disorder, nevertheless needs to be considered in the differential diagnosis of such SIJ anatomical variants as the accessory SIJ and iliosacral complex. Differentiating these variants from osteochondromas is challenging in patients with osteochondromatosis.

KEYWORDS

Computed tomography, magnetic resonance imaging, osteochondroma, osteochondromatosis, sacroiliac joint

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Although solitary osteochondroma is the most common bone tumor, osteochondromatosis, also known as hereditary multiple exostoses, is a rare disease that manifests with the occurrence of multiple (≥2) osteochondromas in bones featuring endochondral ossification.[1] It has an autosomal dominant inheritance pattern with a slight male preponderance and a reported prevalence of 1/1,000 to 1/50,000.[2] Since the disease has different penetrance rates between sexes (almost complete penetrance in males and incomplete penetrance in females), not all patients with osteochondromatosis have a family history,[2] and diagnosis is usually made upon detection of multiple osteochondromas in patients with or without familial history. Osteochondromas remain clinically silent unless they cause a palpable mass, compression of the nearby structures, bone deformity, or fractures. Although rare with solitary osteochondromas (1%), malignant transformation may occur in 3% to 25% of cases with osteochondromatosis.[3]

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Recently, we observed sacroiliac joint (SIJ) involvement in several patients with osteochondromatosis. We also realized that some SIJ anatomical variations (namely, the accessory SIJ and iliosacral complex), which can mimic sacroiliitis and are being increasingly recognized,[4] might be challenging (and might even be mistaken for) in the diagnosis of SIJ involvement in osteochondromatosis. Although flat bones, in particular the ilium and scapula, are commonly affected in patients with osteochondromatosis,[2] there is almost no data in the literature, apart from a few case reports,[5,6] regarding the prevalence of SIJ involvement in osteochondromatosis. In this study, we aimed to investigate the prevalence of SIJ involvement in patients with osteochondromatosis and how such involvement compares with SIJ anatomical variations.

Methods

Ethics approval

This retrospective observational study conducted in a tertiary health care center was approved by the Institutional Review Board with a waiver of informed consent (protocol number: GO 21/521). All procedures performed in this study involving human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

The reporting of this study conforms to the Strengthening the Reporting of Observational Studies in Epidemiology guidelines.[7]

Consent to participate/consent for publication

Approval from the Institutional Review Board was obtained, and in keeping with the policies for a retrospective review, informed consent was not required.

Main points

- Our study demonstrated a high prevalence of intra-articular involvement in osteochondromatosis (73% for the sacroiliac joints), which is considered to be a rare condition.
- Differentiation of sacroiliac joint (SIJ) osteochondromas from anatomical variants is challenging on cross-sectional imaging.
- Knowledge of the frequent SIJ involvement can change the management of patients with osteochondromatosis presenting with low-back pain.

Study population

We searched our institutional electronic patient records for osteochondromatosis (i.e., ≥2 osteochondromas across the skeleton) over the 88-month period from January 2014 through April 2021 by using the following key words: osteochondromatosis, multiple osteochondromas, exostoses, or hereditary exostoses. Having multiple osteochondromas (i.e., ≥2) with or without familial history was used as the diagnostic criterion for osteochondromatosis. Our hospital information system featuring electronic patient records was used to investigate the family history, clinical follow-up, and surgical history of identified patients.

Imaging assessment

SIJ involvement with osteochondromas was investigated on cross-sectional imaging [computed tomography (CT) or magnetic resonance imaging (MRI)] independently by three observers (one junior radiologist, who had just finished a 5-year residency, and two radiologists with 14 and 25 years of dedicated musculoskeletal radiology experience, respectively). The final decision for the presence of osteochondromas within the SIJs was reached by consensus of the two musculoskeletal radiologists in a later joint session. The SIJ variations of the accessory SIJ and iliosacral complex, as defined by Prassopoulos et al.[8] and El Rafei et al.[9], were also noted during the consensus session of the two musculoskeletal radiologists.

An SIJ osteochondroma was defined on CT or MRI either as a sessile or pedunculated cartilage-capped bony overgrowth (with intralesional continuity of the medullary cavity of the parent bone) from the iliac or sacral side of the SIJ protruding into the synovial (cartilaginous) and/or ligamentous portions of the joint.[10]

All imaging studies (i.e., radiographs, CT, and MRI) in all patients with osteochondromatosis were reviewed by the junior radiologist to detect the number and location of osteochondromas across the skeleton. Locations outside the SIJs were labeled as craniofacial, spine (including the sacrum outside the coverage of the SIJs), chest wall, shoulder girdle, elbow (including the three long bones around the elbow), hand and wrist, pelvic girdle (including the ilium outside the coverage of the SIJs), knee (including the patella and the three long bones around the knee), and foot and ankle.

Statistical analysis

Data analysis was performed by using IBM SPSS Statistics 23.0 (Armonk, NY, USA) and free online resources on the GraphPad Software (San Diego, CA, USA) website (www.graphpad.com/quickcalcs). Descriptive analyses were based on frequencies and means of the variables. The Mann–Whitney U test was used to compare the differences of non-categorical continuous data (i.e., age and number of osteochondromas) between independent groups. Fisher's exact test was used to compare the differences in categorical data (i.e., sex and family history) between independent groups. A *P* value less than 0.05 was considered statistically significant. Interobserver agreement was assessed with kappa statistics.[11]

Results

Thirty-six patients with osteochondromatosis (23 females, 13 males) were identified in our database. Their ages ranged between 5 and 71 years (mean, 21 years; median, 16 years); the age data of patients were taken as their age at the time of their cross-sectional imaging covering the SIJs or, where such imaging was not available, from the time of their latest imaging study. Each patient had at least two osteochondromas (range, 2–76; mean, 32) across their skeleton (Table 1).

Of the 36 patients with osteochondromatosis, 22 (61%) had cross-sectional imaging covering SIJs (14 females, 8 males; age range 7–66 years; mean age, 23 years; median age, 17 years; 13 MRI, 9 CT). Nine of these 22 patients had dedicated sacroiliac MRI; the remainder had their SIJs covered in examinations such as abdominopelvic CT, stone protocol abdominal CT, abdominal CT-angiography, spinal MRI, and pelvic/hip MRI. Indications for cross-sectional imaging studies were follow up of painful osteochondromas, low-back pain, abdominal pain, trauma, or post-operative assessment.

Of the 22 patients with SIJ cross-sectional imaging, 16 (73%) had intra-articular osteochondromas (all but two based on the iliac side) involving one (n = 5) or both (n = 11) of the SIJs (Figures 1, 2). The hereditary background of this condition was established in 22 (61%) of the 36 patients with osteochondromatosis [14 (64%) of 22 patients with sacroiliac cross-sectional imaging and 9 (56%) of 16 patients with SIJ involvement]. The characteristics of patients with osteochondromatosis, who had their SIJs covered in a cross-sectional imaging examination, are given in Table 2. All patients with SIJ osteo-

chondromas also had osteochondromas in the pelvic girdle. SIJ osteochondromas were significantly more common in younger patients with osteochondromatosis ($P = 0.012$). The total number of osteochondromas across the skeleton was significantly higher in patients with SIJ osteochondroma than those without ($P = 0.001$). Sex and positive family history were not discriminators for the presence of SIJ osteochondromas ($P = 1.000$ and $P = 0.350$, respectively).

In the cohort with cross-sectional imaging, the anatomical variations of accessory SIJ (n = 6, 27%) and iliosacral complex (n = 2, 9%) were identified in six different patients with (n = 2) and without (n = 4) sacroiliac osteochondromas (Figure 3).

For identifying SIJ osteochondromas on cross-sectional imaging, interobserver agreement was substantial [κ = 0.67; 95% confidence interval (CI): 0.34, 1.00] between the musculoskeletal radiologists and moderate (κ = 0.59; 95% CI: 0.23, 0.94) between the junior radiologist and the final consensus decision of the two musculoskeletal radiologists.

Discussion

This study shows that SIJ involvement is common (73%) in patients with osteochondromatosis. Considering the multiplicity of lesions found within the SIJs, SIJ involvement in osteochondromatosis might even be more prevalent than the occurrence of a solitary (intra-articular) osteochondroma of the SIJ. Thus far, only two solitary osteochondromas of the SIJ have been reported,[5,6] along with a single case of osteochondromatosis with SIJ involvement.[6]

Osteochondromas can increase in size and number during skeletal development;[2] therefore, the prevalence of SIJ involvement can be expected to increase with age. However, in this study, the age of patients with SIJ involvement was significantly lower than that of patients without SIJ involvement, and we detected SIJ osteochondromas in three patients aged 7 and 8 years old. In addition, in this study, the number of osteochondromas across the skeleton was found to be significantly higher in patients with osteochondromatosis involving SIJ osteochondromas than in those without. This finding might imply that SIJ involvement is more common in patients with a greater number of osteochondromas; however, the entire skeleton was not imaged in all of our patients. In addition, smaller sessile osteochondromas might not be amenable to detection on radiographs. Therefore, these figures might not truly reflect the actual number of osteochondromas.

Several anatomical variations of the SIJ, including the accessory SIJ, iliosacral complex, isolated synostosis, unfused ossification center, bipartite iliac bony plate, and semicircular defect, have been described.[8,9] Among these variations, the accessory SIJ and iliosacral complex may mimic SIJ osteochondromas, in particular, the sessile (broad-based) type. The accessory SIJ has a reported prevalence of 1.7% to 19.1%,[8,9,12,13] and the prevalence of the iliosacral complex is reportedly 2.6% to 11%.[8,9,13] Both of these variations are usually seen on the iliac surface at the posterosuperior (ligamentous) portion of the SIJ at the level of the first and second sacral foramina. The iliosacral complex, which indicates a marked prominence of the ilium across a concave recess of the sacrum,[9] is mostly bilateral (Figure 4a, b); the accessory SIJ is reported to be associated with degenerative changes (Figure 4c), in contradistinction to the iliosacral complex.[8,9] We observed that iliosacral complexes generally protrude with shallow angles from the iliac bone, whereas osteochondromas are either pedunculated (with a stalk narrower than the bulk of the lesion) or, when sessile, show steeper angles at their take-off from their base than iliosacral complexes. Nevertheless, distinguishing an osteochondroma from an iliosacral complex is not straightforward. Considering the rarity of osteochondromatosis and the frequency of these variations, different SIJ anatomy is more likely to be a variation rather than an osteochondroma. However, patients with osteochondromatosis may be asymptomatic until they are incidentally diagnosed, and they may present with low-back pain and undergo sacroiliac MRI. As the early diagnosis of osteochondromatosis may dramatically change the treatment and follow-up algorithms, the correct identification of SIJ osteochondromas is especially important in

Table 1. Characteristics of all patients (n = 36) with osteochondromatosis

Characteristic	
Age,[a] years [mean, (range)]	21 [5–71]
Sex (F:M)	23:13
Family history, n (%)	22 (61%)
Number of osteochondromas,[b] mean (range)	32 (2–76)
Involvement sites,[c] n (%)	
Craniofacial (n = 6)	0 (0%)
Spine (n = 32)	12 (38%)
Chest wall (n = 31)	17 (55%)
Shoulder girdle (n = 32)	29 (91%)
Elbow (n = 17)	3 (18%)
Hand and wrist (n = 20)	19 (95%)
Pelvic girdle (n = 32)	28 (88%)
SIJ[d] (n = 22)	16 (73%)
Knee (n = 34)	34 (100%)
Foot and ankle (n = 27)	24 (89%)
Malignant transformation,[e] n (%)	3 (8%)
Other indications for surgical treatment,[f] n	
Painful lesion	12 (39)
Bone deformity	9 (12)
Spinal cord compression	2 (2)
Fracture	1
Radial head dislocation	1

[a]Age data of patients was taken as their age at the time of their cross-sectional imaging covering the SIJs or, where such imaging was not available, from the time of their latest imaging study.
[b]Minimum number of osteochondromas across the skeleton identified on available imaging studies.
[c]Numbers in parentheses below denote patients having imaging studies covering these areas. Spine includes the sacrum outside the coverage of the SIJs, elbow includes the three long bones around the elbow, pelvic girdle includes the ilium outside the coverage of the SIJs, and knee includes the patella and the three long bones around the knee.
[d]This only takes into account cross-sectional imaging (CT or MRI), not radiographs, on which it is not possible to reliably ascertain the presence of SIJ osteochondromas.
[e]Four lesions in three patients, who had SIJ osteochondromas, were surgically excised and histologically proven to be secondary chondrosarcoma (scapula, 1; ilium, 1; fibula, 1; toe phalanx, 1). The mean age at the time of malignancy diagnosis was 31 years (range, 15–46 years).
[f]These indications for surgical treatment in 23 patients exclude malignant transformation, mentioned above. Nine patients had more than one surgery for different indications. Numbers in parentheses are the total number of surgeries for the mentioned indications. SIJ, sacroiliac joint; F, female; M, male; CT, computed tomography; MRI, magnetic resonance imaging.

these patients. Therefore, before a diagnosis of variant anatomy is made, the possibility of an SIJ osteochondroma should be considered, and osteochondromas should be searched for elsewhere in the skeleton. In this study, all patients with SIJ osteochondromas had osteochondromas in the pelvic girdle. Thus, patients with osteochondromatosis presenting with low-back pain and featuring osteochondromas in the pelvic girdle should also be evaluated for SIJ osteochondromas along with other causes of low-back pain. In addition, considering the hereditary background of this condition, undiagnosed relatives of osteochondromatosis patients presenting with low-back pain would benefit from evaluation with sacroiliac MRI for possible SIJ osteochondromas.

Osteochondromas typically display intralesional continuity of the medullary cavity of the parent bone and project away from the epiphysis. However, neither the latter feature in a flat bone, such as the ilium (where the overwhelming majority of osteochondromas in our study were based) nor the former characteristic (since both accessory SIJs and the iliosacral complex also feature a protrusion with a continuation of the medullary cavity) are necessarily helpful for diagnosis. A helpful finding for differentiating osteochondromas from the accessory SIJ and the iliosacral complex, which is more common at the ligamentous (rather than the cartilaginous) portion of the SIJ, is the cartilage "cap" on osteochondromas. However, prominent vessels on the surface of the iliosacral complex described in a previous study[9] and edematous changes at the accessory SIJ should not be confused with the cartilage cap. The mushroom shape of pedunculated osteochondromas is also useful in differential diagnosis. Nevertheless, some accessory SIJs

Table 2. Characteristics of osteochondromatosis patients with a cross-sectional imaging study covering SIJs (n = 22)

Characteristic		*P*
Patients with SIJ osteochondromas, n (%)		
Overall	16 (73%)	
CT (n = 9)	9	-
MRI (n = 13)	7	
Bilateral	11 (69%)	
Unilateral	5 (31%)	
Age, years [mean, (range)]		
With SIJ osteochondromas	17 [7–42]	0.012[d]
Without SIJ osteochondromas	37 [16–66]	
Sex (F:M)		
With SIJ osteochondromas	10:6	1.000[e]
Without SIJ osteochondromas	4:2	
Patients with family history, n (%)		
With SIJ osteochondromas	9 (56%)	0.350[e]
Without SIJ osteochondromas	5 (83%)	
Number of osteochondromas,[a] mean (range)		
With SIJ osteochondromas	44 (17–76)	0.002[d]
Without SIJ osteochondromas	13 (2–30)	
SIJ anatomical variations, n[b] (%)		
Accessory SIJ (n = 6)[c]	5 (23%)	
With SIJ osteochondromas (n = 3)	2 (13%)	
Without SIJ osteochondromas (n = 3)	3 (50%)	-
Iliosacral complex (n = 2)[c]	2 (9%)	
With SIJ osteochondromas (n = 0)	0 (0%)	
Without SIJ osteochondromas (n = 2)	2 (33%)	

[a]Indicates minimum number of osteochondromas across the skeleton.
[b]Numbers in the right column denote patients.
[c]These numbers denote all instances of anatomical variations seen either on the right or the left side.
[d]Mann–Whitney U test.
[e]Fisher's exact test. SIJ, sacroiliac joint; F, female; M, male; CT, computed tomography; MRI, magnetic resonance imaging.

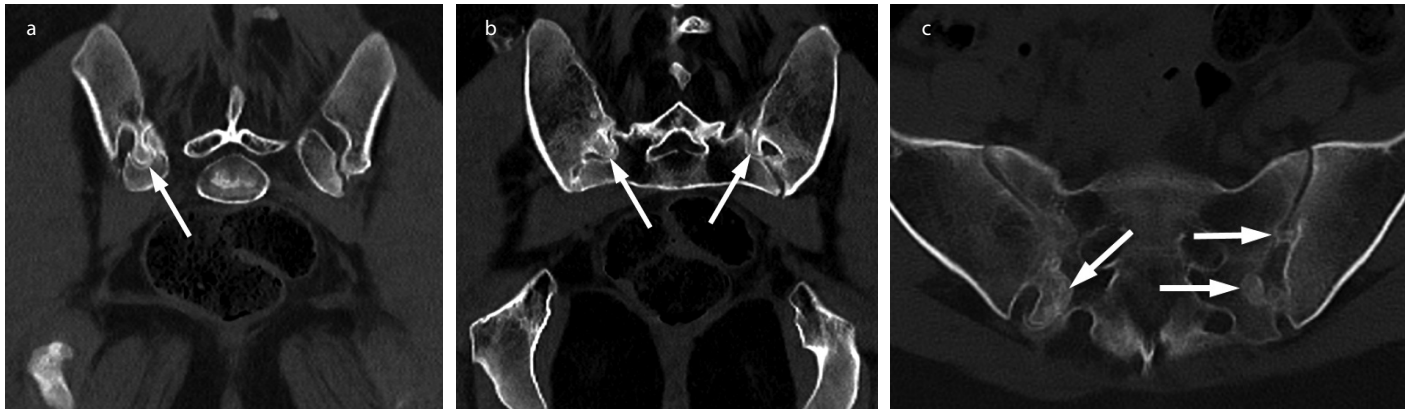

Figure 1. A 16-year-old girl with osteochondromatosis. Oblique coronal reformatted **(a, b)** and axial **(c)** pelvic computed tomography images show ilium-based sacroiliac joint osteochondromas (arrows) on both sides.

mentioned in the literature[8] clearly showed a mushroom shape as well. Furthermore, patients with osteochondromatosis may have SIJ variations. In this study, the anatomical variations of accessory SIJs (n = 6, 27%) and the iliosacral complex (n = 2, 9%) were identified in six different patients with (n = 2) and without (n = 4) sacroiliac osteochondromas. In view of the aforementioned conditions, differentiating these variants from osteochondromas can be challenging in patients with osteochondromatosis. In general, MRI is better than CT at showing the cartilaginous cap in osteochondromas, whereas the greater spatial resolution of CT renders it more helpful than MRI in identifying bony contours of especially small osteochondromas and accessory SIJs.

Although the prevalence of intra-articular involvement in osteochondromatosis is not established, it is considered to be rare.[14,15] The frequent SIJ involvement shown in our study is remarkable in this regard. SIJ involvement has been described in dysplasia epiphysealis hemimelica (Trevor disease),[16,17] which, although featuring intra-articular osteochondroma-like (or, more accurately, osteocarti-

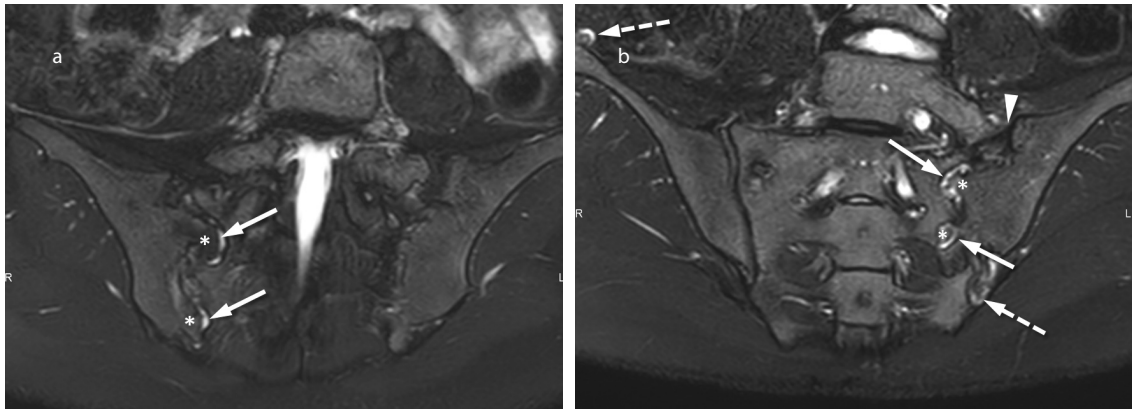

Figure 2. A 17-year-old girl with multiple hereditary exostoses (osteochondromatosis). Coronal T2-weighted magnetic resonance images with fat saturation **(a, b)** show bilateral sacroiliac joint osteochondromas (asterisks), all but one ilium-based (the inferior osteochondroma on the left is sacrum-based). Note cartilage caps on intra-articular (solid arrows) and extra-articular (dashed arrows) osteochondromas. Note also the left-sided pseudoarthrosis (arrowhead) of a lumbosacral transitional vertebra.

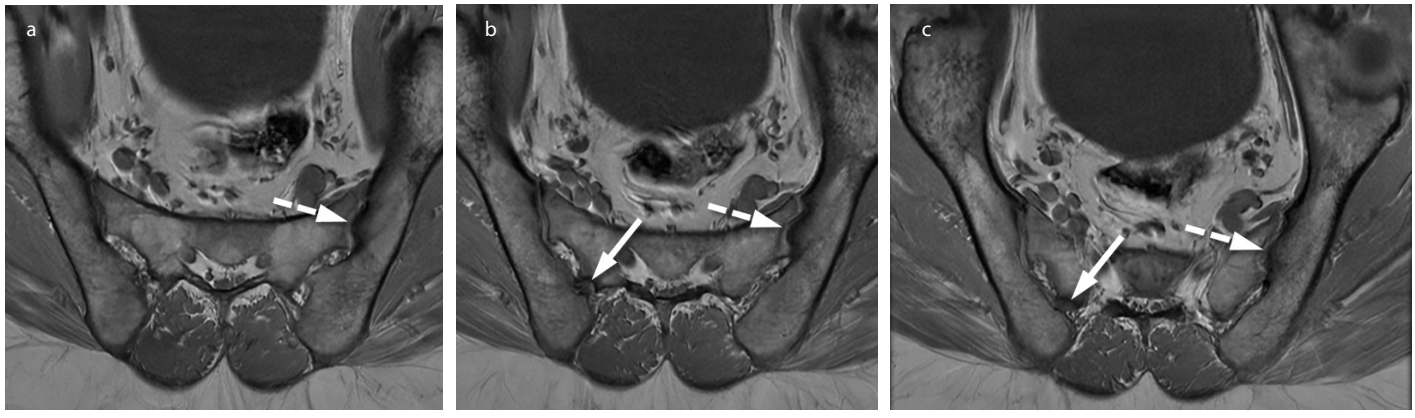

Figure 3. A 31-year-old man with multiple hereditary exostoses (osteochondromatosis). Oblique axial T1-weighted magnetic resonance images **(a-c)** show an accessory sacroiliac joint on the right (solid arrows) and an iliosacral complex on the left (dashed arrows). No sacroiliac joint osteochondromas were identified.

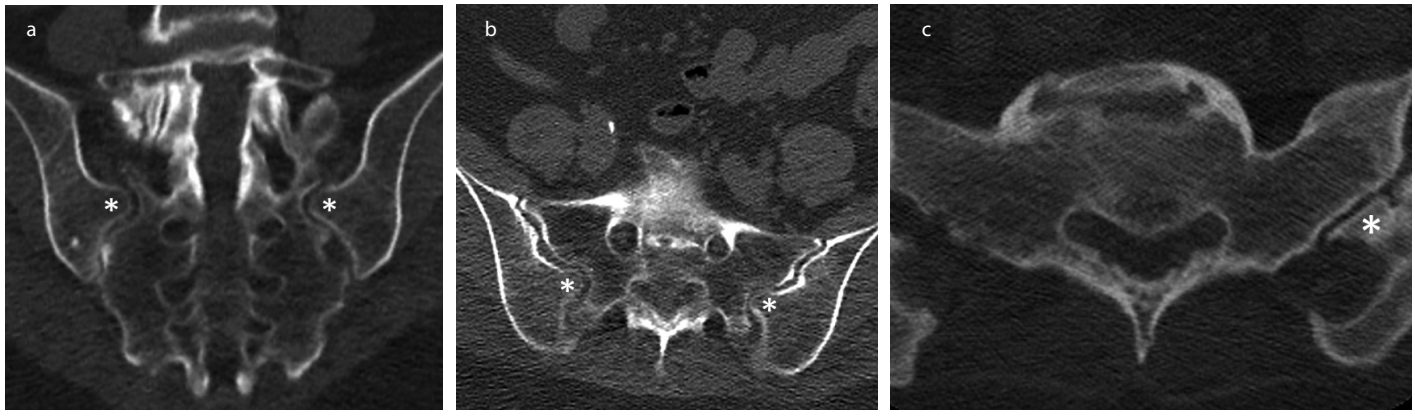

Figure 4. Anatomical variants in two patients without osteochondromatosis (outside the study group). Bilateral iliosacral complexes (a, **b**; asterisks) on oblique coronal reformatted **(a)** and axial **(b)** computed tomography (CT) images in a 66-year-old woman who underwent the pelvic CT exam for total hip arthroplasty control. An accessory sacroiliac joint **(c**; asterisk) on axial CT image in a 38-year-old woman who underwent the lumbar spinal CT exam for posterior spinal fixation hardware control.

laginous) lesions, has a different mechanism than osteochondromatosis.[18] Trevor disease, characterized by osteocartilaginous epiphyseal lesions, is a very rare clinical entity with about 150 reported cases.[18] The most common sites of disease are epiphyses and epiphyseal equivalents of the lower limb. Although bilateral involvement has been reported,[19] the distribution of the lesions usually fits a hemimelic pattern with the involvement of multiple joints in the same extremity. The SIJ is an uncommon location for Trevor disease and has been reported in only two cases.[16,17] Unilateral involvement of the SIJ and multiple lesions on the epiphyses and epiphyseal equivalents in the same extremity appear to be reliable findings in differentiating between osteochondromatosis and Trevor disease.[18] None of our cases featured these findings.

Our study has several limitations. First, the small number of patients with osteochondromatosis who had undergone cross-sectional imaging covering their SIJs in our study limits its power. Nevertheless, as the first study to look into SIJ involvement in osteochondromatosis, it features a cohort of patients (n = 36), 61% of whom had cross-sectional imaging of the SIJs. Second, there is selection bias, as the observers were aware while reviewing the radiological examinations that the patients had osteochondromatosis. However, this did not prevent us from factoring in the already-established anatomical variations of the SIJs that might have mimicked osteochondromatosis. Third, the presence of osteochondromas involving the SIJs might have been obscured by some of the SIJ anatomical variations. Fourth, the presented cases were treated in a tertiary health care center; therefore, our study may not reflect the true prevalence of SIJ involvement in osteochondromatosis. Fifth, we do not have histologic proof of osteochondromas involving the SIJ, as none of these patients needed to undergo surgery in this area. However, when an iliosacral complex is located at the cartilaginous portion of the joint, it would also be covered with a cartilage cap (the joint cartilage), and even histology might not be very helpful in differentiating it from a sessile osteochondroma. The same is valid for an accessory SIJ, which might be overlined by cartilage (in a synchondrotic accessory joint). Finally, the relationship between SIJ osteochondromas and low-back pain could not be definitively ascertained due to the retrospective design of the study. Therefore, we were not able to further elaborate on the clinical significance of SIJ involvement in osteochondromatosis.

In conclusion, this study investigated the SIJ involvement in osteochondromatosis. Cross-sectional imaging shows such involvement to be frequent (73%). Although a rare disorder, osteochondromatosis nevertheless needs to be considered during daily radiological reporting practice in the differential diagnosis of such SIJ anatomical variants as the accessory SIJ and iliosacral complex. However, differentiating these variants from osteochondromas can be challenging in patients with osteochondromatosis. Patients with SIJ osteochondromas commonly display other osteochondromas in the pelvis, which might help ascertain osteochondromatosis in a focused imaging examination such as sacroiliac MRI. Knowledge of the frequency of SIJ involvement can change the management of patients with osteochondromatosis presenting with low-back pain.

Conflict of interest disclosure

The authors declared no conflicts of interest.

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