

Case Study

## Coexistence of osteopoikilosis with seronegative spondyloarthritis and spinal stenosis

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**Abstract.** [Purpose] Osteopoikilosis is a rare hereditary bone disease that is usually asymptomatic. It is generally diagnosed incidentally on plain radiography. The coexistence of osteopoikilosis with seronegative spondyloarthritis or spinal stenosis is rarely reported. Here, we report the case of a 27-year-old male patient with osteopoikilosis, seronegative spondyloarthritis, and spinal stenosis. [Subject] A 27-year-old male patient with buttock pain and back pain radiating to the legs. [Methods] A plain anteroposterior radiograph of the pelvis revealed numerous round and oval sclerotic bone areas of varying size. Investigation of the knee joints showed similar findings, and the patient was diagnosed with osteopoikilosis. Lumbar magnetic resonance images showed spinal stenosis and degenerative changes in his lumbar facet joints. Magnetic resonance images of the sacroiliac joints showed bilateral involvement with narrowing of both sacroiliac joints, nodular multiple sclerotic foci, and contrast enhancement in both joint spaces and periarticular areas. HLA B-27 test was negative. [Results] The patient was diagnosed with osteopoikilosis, seronegative spondyloarthritis, and spinal stenosis. Treatment included asetmetasin twice daily and exercise therapy. [Conclusion] Symptomatic patients with osteopoikilosis should be investigated for other possible coexisting medical conditions; this will shorten the times to diagnosis and treatment.

**Key words:** Osteopoikilosis, Spinal stenosis, Spondyloarthritis

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### INTRODUCTION

Osteopoikilosis (OPK) is a rare hereditary bone dysplasia<sup>1,2)</sup>. This disease exhibits autosomal dominant inheritance, and its clinical course is usually asymptomatic<sup>2,3)</sup>. OPK is generally diagnosed incidentally on plain radiography. Its radiological findings are typical and include diffuse, small round or ovoid, symmetric sclerotic bone areas<sup>4)</sup>. The coexistence of OPK with spondyloarthritis<sup>1,5,6)</sup> or spinal stenosis<sup>7-9)</sup> is rarely reported. Here, we report the case of a 27-year-old male who suffered from back and leg pain and was diagnosed with OPK, spondyloarthritis, and spinal stenosis.

### SUBJECT AND METHODS

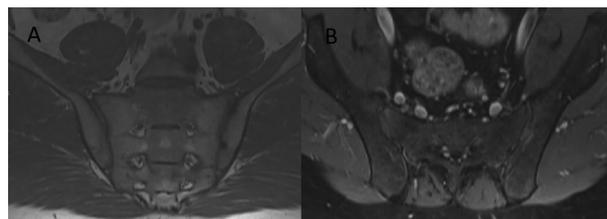
A 27 year-old male patient was referred to the Physical Medicine and Rehabilitation Outpatient Clinic with a history of posterior bilateral buttock pain and back pain radiating to the legs for one month. His radicular back pain was inter-

mittent and aggravating by walking or prolonged standing. The patient did not report morning stiffness at that time. He had no pain while resting. Physical examination revealed limited mobility of the lumbar spine in all planes. The result of the straight leg raising test was negative. The Schober test result was normal. Neurological examination was normal. Sacroiliac joint compression test results were positive. As spinal stenosis was considered as a preliminary diagnosis, magnetic resonance imaging (MRI) of the lumbar spine was performed. Lumbar MRI showed obvious degenerative changes in the lumbar facet joints incompatible with his age; bulging discs at the L3–4, L4–5, and L5–S1 levels; and spinal stenosis. Lumbar MRI also showed compression of bilateral L5 and S1 nerve roots and relative spinal stenosis. His pain improved with nonsteroidal anti-inflammatory drugs (NSAIDs) and physical therapy. However, his symptoms reappeared three weeks later. He described his pain level as moderate according to a 4-point Likert scale. Upon reexamination, test results for sacroiliac joints were positive. A plain anteroposterior radiograph of pelvis was taken and revealed numerous round and oval sclerotic bone areas of varying size at the ilea, ischia, sacrum, around the acetabulum of the pelvis, and proximal femur bones (Fig. 1A). Investigations of knee joints showed similar findings, and the patient was therefore diagnosed with OPK (Fig. 1B). The sacroiliac joints appeared narrowed and sclerotic on the pelvic radiograph. MRI of the sacroiliac joints showed bilateral involvement with narrowing of both sacroiliac joints and multiple

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**Fig.1.** A. Pelvic radiograph showing bilateral sclerotic bone areas at the ilea, ischia, sacrum, around the acetabulum of the pelvis, and proximal femur. B. Bilateral knee radiograph showing multiple sclerotic bone lesions



**Fig. 2.** A. T1-weighted MRI showing bilateral involvement with narrowing of both sacroiliac joints and multiple nodular sclerotic foci. B. Post-contrast images showing contrast enhancement in both joint spaces and periarticular areas

nodular sclerotic foci. Post-contrast images showed contrast enhancement in both joint spaces and periarticular areas (Fig. 2). Laboratory evaluations were as follows: erythrocyte sedimentation rate, 2 mm/h; C-reactive protein, 0.13 mg/dL (reference range 0–0.5 mg/dL); leukocytes,  $7.86 \times 10^3/\mu\text{L}$ ; platelets,  $269 \times 10^3/\mu\text{L}$ ; and hemoglobin, 15.4 g/dL. Serum urea, creatinine, alkaline phosphatase, and electrolytes levels were normal, and the HLA B-27 test result was negative. This case study complied with the ethical standards of the Declaration of Helsinki (1975, revised 1983). The ethics committee of our institution approved publishing the case details. The patient provided written informed consent.

## RESULTS

According to the Assessment of SpondyloArthritis International Society (ASAS) criteria<sup>10</sup>, the patient was diagnosed with seronegative spondyloarthritis. Treatment included aemetasin 60 mg twice daily for four weeks and exercise therapy. His pain improved as a result of this treatment. He described his pain level as mild according to a 4-point Likert scale.

## DISCUSSION

Albers-Schönberg first described osteopoikilosis in 1915 as a sclerosing bone dysplasia. Its estimated prevalence is reported to be 1 in 50,000 people. The characteristic radiologic feature is multiple, sclerotic, round or oval foci. These foci are usually distributed symmetrically in periarticular areas within the epiphyseal and metaphyseal regions<sup>4</sup>. As mentioned above, its course is usually asymptomatic. OPK has been reported to be associated with various developmental malformations such as coarctation of aorta, pubertas praecox, diabetes mellitus, dacryocystitis, chondrosarcoma, synovial chondromatosis, otosclerosis, syndactyly, and Klippel-Feil syndrome<sup>8, 9, 11–14</sup>. The number of reported cases of OPK coexisting with rheumatic diseases is increasing. Rheumatic diseases reported in patients with OPK include reactive arthritis<sup>15</sup>, rheumatoid arthritis<sup>16</sup>, juvenile arthritis<sup>2</sup>, discoid lupus erythematosus, spondyloarthritis<sup>1, 5, 6</sup>, psoriatic arthritis<sup>17</sup>, Raynaud phenomenon<sup>1</sup>, antiphospholipid syndrome, livedoid vasculitis, undifferentiated connective tissue disease<sup>18</sup>, familial Mediterranean fever<sup>5, 19</sup>, fibromyalgia<sup>3, 20</sup>, myofascial pain syndrome<sup>3</sup>, adhesive capsulitis<sup>11</sup>, spinal

stenosis<sup>7–9</sup>, and De Quervain syndrome<sup>21</sup>.

Spondyloarthritis comprises a group of inflammatory rheumatic conditions<sup>22</sup>. The group includes ankylosing spondylitis, psoriatic arthritis, reactive arthritis, and enteropathic arthritis. Because of the long delay between symptoms onset and the diagnosis of spondyloarthritis, the ASAS developed new classification criteria for spondyloarthritis; these criteria aim to provide earlier and more accurate diagnosis for both early and established cases. These criteria include MRI as an important tool to detect active inflammation for early diagnosis<sup>10</sup>. We made the diagnosis of spondyloarthritis according to the ASAS criteria in the present case on the basis of the following: low back pain with a duration of at least three months, onset at less than 45 years of age, active inflammatory lesions of sacroiliac joints on MRI, and good response to NSAIDs. The patient had a negative HLA B-27 test result.

In patients with spinal stenosis, the space within the spinal canal narrows due to the reduced volume of the central canal, lateral recesses, and/or neural foramina caused by degenerative changes or congenital causes<sup>23</sup>. Degenerative changes including disc dehydration, disc herniation, disc space narrowing, facet joint hypertrophy, and ligamentum flavum thickening are the most common causes of spinal stenosis. Radiographic changes associated with stenosis are very common with aging, especially in people older than 50 years. The major symptoms include back and buttock pain, weakness, numbness, and neurogenic intermittent claudication<sup>24, 25</sup>. Patients with unusually early onset of degenerative spinal changes, as in the present case, should be investigated for underlying congenital pathologies.

Clinicians should investigate the existence of other possible coexisting medical conditions in symptomatic patients with OPK in order to shorten the time to diagnosis and provision of appropriate treatment.

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