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Adult-Onset Satoyoshi Syndrome and Axial Spondyloarthritis

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Dear Editor,

A 23-year-old male presented with a 2-month history of frequent recurring episodes of severe painful muscular spasms involving the lower limbs (worse on the right) that were restricting his activities of daily living. He has been evaluated for alopecia when he developed progressive scalp hair loss 6 years previously. Concurrently, he also experienced intermittent diarrhea about once a week and lower back pain for the past 6 months. The insidious lower back pain was worse at night and relieved after physical activity, and was associated with gradual restriction of his spinal mobility. He did not experience dysphagia, speech disturbances, blurring of vision, tremors of the limbs, or seizures. He reported no significant family history of any movement disorder or similar type of spasm.

On examination, he had marked alopecia for his age and a mildly stooped head posture with flexion of the hips and knees when standing erect. There was no uveitis, conjunctivitis, or arthritis. A neurological examination showed no muscle atrophy, weakness, abnormal sensation, or pathological reflexes of the upper or lower extremities. Episodes of painful spasms of the right lower limb (Supplementary Video 1 in the online-only Data Supplement) with clawing of the toes, forced flexion of the knee, and dorsiflexion and inversion of the foot lasting for 3−5 minutes were noted. A gait examination showed a normal stride length with no dystonic posturing. Chest expansion and spine movement were both reduced. The modified Schoeber index was less than 10 cm (normal: >15 cm), and the FABER test was positive.

Investigations revealed positivity for HLA-B27 with radiographic features of the early formation of syndesmophytes over the lumbar spine and ill-defined sacroiliac joints suggestive of sacroiliitis. Radiography of the hands showed shortening (brachydactyly) and metaphyseal flaring of the middle phalanx of the little fingers bilaterally without osteolytic lesions (Fig. 1). The findings of a nerve conduction study of the lower limbs were normal, and electromyography of the left vastus medialis revealed fasciculations and continuous motor unit activity. MRI of the brain and lumbosacral spine was unrevealing.

Based on the combination of painful recurrent muscle spasms, alopecia, and diarrhea, a diagnosis of Satoyoshi syndrome and ankylosing spondylitis was made. The sequential development of alopecia followed by painful muscle spasms, diarrhea, and inflammatory back pain was also consistent with Satoyoshi syndrome. Alopecia usually precedes the constellation of other symptoms, although the concurrent appearance of alopecia and muscle spasms has been reported. Despite being present in all patients, a characteristic pattern of alopecia has not been identified, with alopecia areata, totalis, and universalis all being reported.[1] A full autoimmune panel including antinuclear antibodies and rheumatoid factor was unrevealing (Supplementary Table 1 in the online-only Data Supplement). The patient was prescribed a trial of phenytoin at 100 mg three times daily and oral prednisolone at 40 mg daily, which resulted in the gradual improvement of his diarrhea and muscle spasms without any recurrence. The diagnosis of Satoyoshi syndrome in this case is still a clinical one con-

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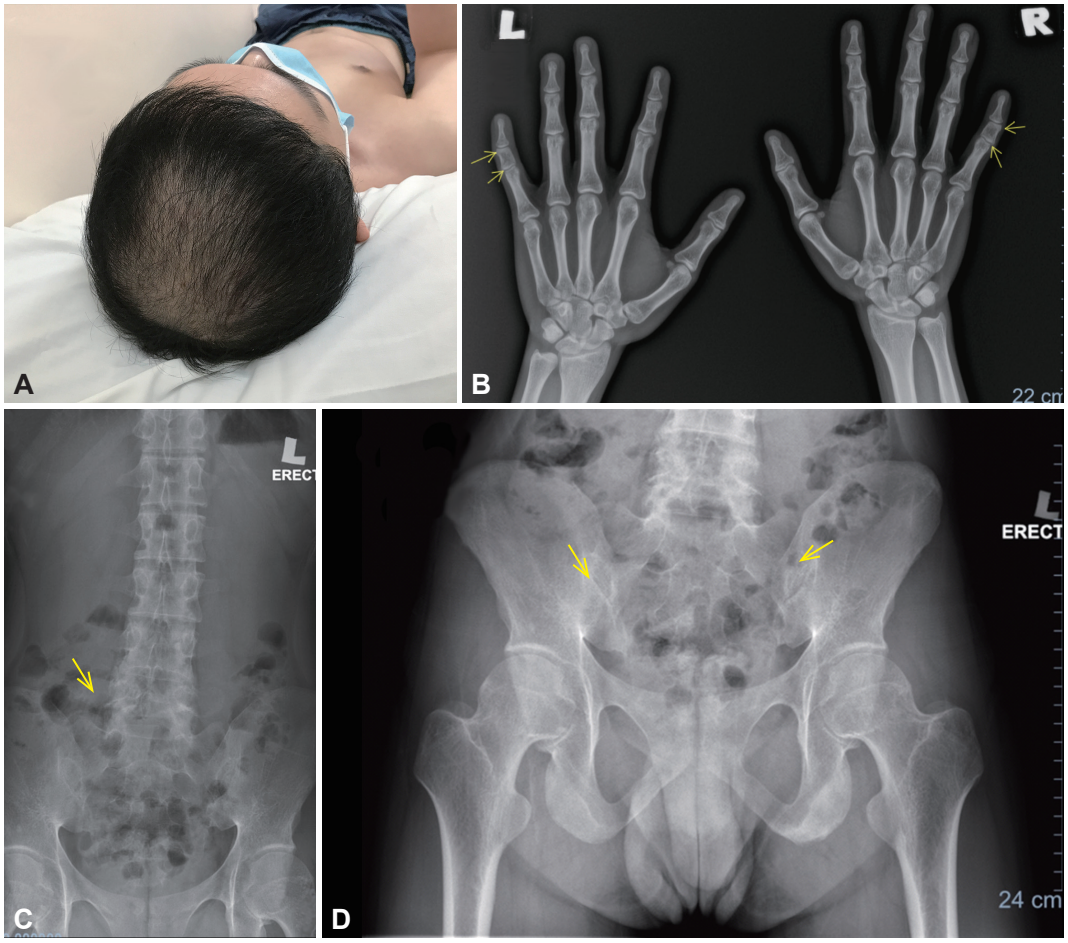

Fig. 1. A: Our patient with Satoyoshi syndrome showing alopecia involving the scalp hair. B: X-ray of the hands (AP view) showing shortening and non-specific metaphyseal flaring of the middle phalanx of the little fingers bilaterally (arrows). C: Arrow indicates lateral bony bridging at the L4/L5 and L5/S1 levels suggestive of early formation of syndesmophytes. D: Arrows indicate ill-defined sacroiliac joints bilaterally suggestive of joint fusion (ankylosis). AP, anterior-posterior.

sidering the chronology and cluster of symptoms, characteristic skeletal abnormalities, and favorable response to steroid therapy.

Satoyoshi syndrome was first described in 1967 by Satoyoshi and Yamada, and constitutes a multisystem disease characterized by progressive painful muscular spasms, alopecia, diarrhea, endocrinopathy, and secondary skeletal abnormalities.[2] An autoimmune basis is implicated through the association with other autoimmune conditions, presence of autoantibodies, and successful treatment of symptoms with immunosuppressants.[3]

This is the first report of an association between Satoyoshi syndrome and axial spondyloarthritis, which include ankylosing spondylitis. Skeletal abnormalities such as joint deformities, epiphyseal separation, fatigue fractures, and metaphyseal lesions may appear with Satoyoshi syndrome.[4] It has been postulated that repeated traumatic physical injuries to the growth plates, epiphyses, and tendon attachments due to recurrent muscle spasms could be responsible for some of the skeletal problems encountered.[5] However, the implicated relationship of severe muscle spasm with skeletal changes cannot be explained by the presence of bilateral symmetrical sacroiliitis and HLA-B27 positivity in our patient. Furthermore, joint destruction and growth plate changes are unlikely to be solely due to repetitive trauma, given that the painful spasms in our patient first started in late adulthood when the growth plates were already fused.

The pathogenesis of Satoyoshi syndrome remains uncertain. Our findings of ankylosing spondylitis and HLA-B27 positivity suggest that genetic factors are important in the susceptibility to Satoyoshi syndrome. The present report of this novel association could have pathophysiological implications as well as be helpful in the clinical management of this rare disease. The purpose of this commentary is to increase familiarity with this rare condition and with the possibility of other autoimmune diseases—such as Satoyoshi syndrome—being associated with alopecia, diarrhea, and recurrent painful muscular spasms.

Supplementary Video Legend

Video 1. The intense muscular spasms were characterized by painful dorsiflexion and inversion in the right foot.

Supplementary Materials

The online-only Data Supplement is available with this article at https://doi.org/10.3988/jcn.2021.17.4.593.

Ethics Statement

Written informed consent from the patient has been obtained for publication of this research.

Availability of Data and Material

All data generated or analyzed during the study are included in this published article (and its supplementary information files).

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Conflicts of Interest

The authors have no potential conflicts of interest to disclose.

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