# Spondyloarthropathy: A Common Familial Form of Arthritis

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Ankylosing spondylitis is the most common of a group of related seronegative rheumatic diseases known collectively as the spondyloarthropathies. Other illnesses in this group include Reiter's syndrome, psoriatic arthritis, and the arthropathies of ulcerative colitis and regional enteritis.[1] Recent epidemiologic evidence suggests that these conditions are often concentrated in families, are frequently associated with the HLA-B27 tissue-typing antigen, are more common than previously recognized, and are easily confused with other diseases.[2] A case report of a young boy with spondyloarthropathy will serve to highlight the clinical features at onset, diagnostic problems, family involvement, and the clinical course.

## CASE REPORT

A 12-year-old boy was seen at the University of Missouri Family Medical Care Center in December 1980 because of left hip pain. The pain worsened with exercise, awakened him at night, and was associated with morning stiffness. There was no personal or family history of tendonitis, rash, fever, ocular complaints, gastrointestinal symptoms, or involvement of other joints. Two years previously he had a similar complaint, diagnosed as "toxic synovitis," which resolved spontaneously. Three weeks previously, an orthopedist diagnosed "traumatic sacroiliitis."

The general physical examination was normal, except that his height and weight were below the third percentile. The ranges of motion of all joints, including the left hip, were normal. No effusion, synovial thickening, or warmth were found. Tenderness to percussion over the left sacroiliac joint was elicited.

Upon referral, a pediatric orthopedist found a normal sedimentation rate, white blood cell count, and hemoglobin level; antinuclear antibodies and rheumatoid factor were absent. Roentgenograms of the spine, sacroiliac joints, and hips were normal, as was a left hip arthrogram. A pediatric rheumatologist then saw the boy and made a presumptive diagnosis of spondyloarthropathy on the basis of recurrent arthritis in the lower extremities in a male patient with associated sacroiliitis; a test for HLA-B27 was positive. The boy was begun on aspirin, 10 grain four times a day, with marked improvement. He was instructed in posture and physical therapy. History from the father revealed long-standing problems with his shoulders, low back, and parasternal joints. A maternal aunt and grandmother were also both subsequently found to have psoriasis.

Over the next year the boy experienced severe, transient arthritis in the left third and fourth metacarpophalangeal joints and left third metatarsophalangeal joint; however, back pain did not recur. Tolmetin, 200 mg four times a day, was added to his treatment, with good response. During the second year of illness he developed ankle arthritis, fingernail pitting, onycholysis, and a scaly, erythematous eruption over his knees; a diagnosis of psoriasis was confirmed. Indomethacin, 25 mg half strength, was added to ameliorate worsening morning stiffness. The boy's height progressively rose to the 25th percentile. He has remained active in sports and is mostly pain-free.

The patient's father developed multifocal costochondritis in 1981 and was also found to be HLA-B27 positive. The patient's younger brother has been

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evaluated twice for plantar fasciitis and recurrent neck pain.

## DISCUSSION

The spondyloarthropathies are a group of chronic inflammatory diseases affecting primarily the axial skeleton and lower extremities, most frequently becoming evident in older children or young adults.[1,2] Ankylosing spondylitis is the prototype; other rheumatoid factor-negative spondyloarthropathies include Reiter's syndrome, psoriatic arthritis, the enteropathic arthropathies, and various reactive arthritidies—all of which are associated to varying degrees (40 to 90 percent) with the HLA-B27 antigen.

Ankylosing spondylitis is the most common cause of arthritis in boys and young men and has a prevalence equal to rheumatoid arthritis.[1] HLA-B27 is present in 6 to 8 percent of white populations and in 4 percent of blacks. Twenty percent of the HLA-B27-positive male population will eventually develop symptomatic spondylitis,[2,3] and a similar percentage of the female population will have distinctive radiologic changes but less specific physical complaints. The sex distribution is therefore equal, although spondyloarthropathy is diagnosed less frequently in women. Spondyloarthropathies thus have a prevalence of 1 to 1.5 percent in the general population; however, they are diagnosed much less frequently, probably because of underrecognition, the mild nature of the illneses in many patients, and the apparent nonspecificity of complaints. A family history of spondyloarthropathy, related symptoms, or associated illnesses may be elicited in nearly one half of affected children.[4]

The pathologic hallmark of the spondyloarthropathies in children and adults is enthesopathy—inflammation of the bony attachments of ligaments and tendons.[4] In more severely affected persons, erosive osteitis and periostitis may develop; associated rapid and extensive healing of these lesions often causes new bone formation along ligaments, which may result in bony spurs such as at the heel. Ultimately this process in the spinal ligaments results in the so-called "bamboo spine."

Diagnostic criteria for ankylosing spondylitis often have limited utility, especially in the early stages of illness. Decreased mobility of the lumbar spine, low back pain, reduced chest expansion, and radiographic changes in the sacroiliac joints confirm the diagnosis in adults according to the "Rome" and "New York" criteria, but these findings may not be evident in the early years of disease, particularly in children.[4] Hence, many experts rely on the simple evidence of symptomatic sacroiliitis. Testing for HLA-B27 is positive in 70 to 80 percent of children and adults with ankylosing spondylitis and thus may be useful for an early, presumptive diagnosis. However, the test is not needed if the diagnosis is clinically evident.[5] The frequency of HLA-B27 in other spondyloarthropathies correlates with the presence of arthritis, and ranges from 40 percent in psoriasis and the enteropathic arthropathies to 75 to 90 percent in Reiter's syndrome.

In a preselected population the insidious onset of back pain in a patient younger than 40 years, persisting for at least three months, associated with morning stiffness, and improving with exercise was 95 percent sensitive and 85 percent specific for inflammatory spinal disease.[6] In contrast, about one third of patients with spondyloarthropathies experience peripheral arthritis first, usually as asymmetric pauciarticular involvement of the lower extremities.[7] Enthesopathy and arthritis of the first metatarsal-phalangeal joint is a common presentation, but must be carefully sought.[8] Heel pain, plantar fasciitis, sausage digits, inflammation at the anterior tibial tubercle, costochondritis, and other forms of periostitis also occur frequently. Uveitis, aortitis, and pulmonary fibrosis are nonarticular manifestations that should be looked for. It is also essential, particularly in children, to search for the conjunctivitis and urethritis of Reiter's syndrome; the diarrhea, weight loss, and bloody stools of the enteric arthropathies; and the skin and fingernail changes of psoriasis, as demonstrated in this teenage patient.

The prognosis for the spondyloarthropathies is highly variable. The most severe cases develop spinal ankylosis and become susceptible to vertebral fracture. Severe kyphosis may also occur as a result of chronic poor posture, and peripheral arthritis or enthesitis may be severe enough to limit mobility and function. One half of all patients with ankylosing spondylitis, Reiter's syndrome, psoriatic arthropathy, or the enteric arthropathies become significantly and permanently disabled.

The objective of treatment is to relieve pain, decrease inflammation, and to maintain good posture and function. Nonsteroidal anti-inflammatory agents, beginning with aspirin or tolmetin, adding indomethacin and phenylbutazone as indicated, are useful in diminishing symptoms and altering the disease progression. Salicylates seem to work less well but should be tried first. A firm mattress and thin pillow, along with extension exercises, may ameliorate cervical and thoracolumbar deformities. Swimming is also an excellent form of physical therapy.

Family physicians have an excellent opportunity to diagnose the spondyloarthropathies at an early stage and in different family generations. Ankylosing spondylitis, as well as the other less common spondyloarthropathies, should be considered in persons with low back pain and morning stiffness, or in seronegative asymmetric, pauciarticular arthritis when occurring in boys and young men and in patients with enthesopathy. Often a presumptive diagnosis based on

clinical findings and family history must be made and treatment initiated, as other confirmatory features often occur only late in the disease.

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