


**Autosomal dominant undifferentiated spondyloarthropathy not related to the HLA system**

We were interested to read the article by Kidd and colleagues1 on familial aggregation for undifferentiated spondyloarthropathy associated with HLA-B7. The authors described a single family in whom numerous members had a relapsing, negative arthritis or enthesopathy, or both, which fulfilled the European Spondyloarthropathy Study Group criteria for spondyloarthropathy2 in the absence of the HLA-B27 tissue type, coexistent psoriasis, or inflammatory bowel disease. They suggested that 'undifferentiated' spondyloarthropathy can be associated with genetic factors other than HLA-B27. We are in total agreement.

A few years ago, we studied a French family of 83 members distributed over five generations divided into four main branches, among whom 18 adult members had destructive arthropathy and enthesopathic changes.3 4 In all patients, the disease began between the ages of 18 and 32 years. It affected predominantly the wrists, fingers, shoulders, and peripheral entheses and progressed as an oligoarthropathy, with intermittent inflammatory episodes lasting for one to three months. Axial involvement of the cervical and lumbar spine and the sacroiliac joints was also seen, but was not prominent. The sites of involvement seemed to be influenced by mechanical factors. The right wrist was generally the first joint to be affected. Destructive abnormalities, followed by bony proliferation, and intra- or extra-articular bony ankylosis were the main radiological features of this familial arthropathy (figure).

The transmission of the disease was dominant and autosomal, with 100% penetrance. The clinical and radiological features were strikingly similar in all patients with successive generations and different branches of the genealogical tree, suggesting monogenic transmission. HLA typing of 12 patients and 13 healthy family members was performed. No HLA antigen was linked to the disease. None of the affected subjects had antigens B27, DR4 or DR7. The disease was not transmitted with any particular HLA haplotype.

Tests for rheumatoid factor yielded negative results. There was no history of psoriasis or chronic entheropathy in the members of this family. In none of the 18 patients did the arthropathy fulfill the American Rheumatism Association criteria.3 A diagnosis of ankylosing spondylitis was also eliminated, because the New York criteria were not fulfilled.5 This familial arthropathy could belong to the class of undifferentiated spondyloarthropathies proposed by Bums and Calin.6 The spondyloarthropathies have in common a non-specific inflammation of the entheses, involving both the chondrified and the calcified parts. After a destructive phase, which causes bony erosion, repair takes the form of ossification.6 7 An inherited abnormality in the collagen matrix of the entheses may predispose to destructive arthropathy and enthesopathic changes. Linkage analysis of COL2A1, the gene responsible for the disease causing locus in this family,8 further studies are needed to identify the genetic locus responsible for the disease.

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