The spondyloarthropathies share a number of common features, of which the most important may be an association with the histocompatibility antigen, HLA-B27. Although there are significant clinical differences between these conditions, the common clinical manifestations, along with the association with HLA-B27, imply that they might form a group of conditions with a related pathogenic mechanism. The spondyloarthropathies include ankylosing spondylitis, Reiter’s syndrome, reactive arthritis, spondylitis associated with psoriasis and inflammatory bowel disease, and a variety of less clearly defined conditions termed undifferentiated spondyloarthropathies. In each of these conditions, there is infiltration of affected structures by inflammatory cells, including lymphocytes, plasma cells, macrophages, polymorphonuclear leukocytes, and mast cells. The etiopathogenesis of none of these conditions is clearly understood, although HLA-B27 is likely to play a central role in each.

Transgenic rats expressing the human HLA-B*2705 gene product spontaneously develop an inflammatory condition that resembles the spondyloarthropathies. These features include peripheral and axial arthritis, gastrointestinal inflammation and diarrhea, psoriasiform skin changes, and male urogenital inflammation. Histologically, the joint, gut and skin lesions closely resemble the lesions seen in HLA-B27-associated disease in humans. A number of transgenic rats expressing HLA-A2 or HLA-B7 fail to develop inflammatory disease, indicating that HLA-B27 is likely to be specifically involved in the development of this inflammatory disease. These observations provide direct evidence for the participation of the HLA-B27 molecule in disease pathogenesis, and should prove useful in elucidating the pathogenetic role of HLA-B27. Of note, gastrointestinal inflammation is an early and consistent finding that precedes the other disease manifestations and may be important in the pathogenesis of the entire disease process. This is consistent with the observation that gastrointestinal and articular inflammation is intimately related in the spondyloarthropathies.

Conventional HLA B27 typing is based on the detection of HLA antigens at the cell surface, usually by the classical method of complement-dependent cytotoxicity using Terasaki plates. For routine purposes, screening for HLA B27 antigen expression has been based on flow cytometry. A major disadvantage to serological methods for HLA B27 antigen detection is the cross-reactivity of the antibodies used with different HLA antigens. To diminish the potential risk of false positive results, alternative methods were developed based on analysis of the genes coding for the HLA-B antigens.

Allele-specific PCR for HLA B27 is a direct genotyping method based on specific primer recognition of a unique HLA B27 gene sequence. It uses a HLA B27 unique primer (E91s) and a primer specific for HLA B alleles (E136as). The 134 bp HLA B27 product is clearly detected using a mini-gel format. An internal amplification control is provided by a second set of primers (Human Growth Hormone- HGH) in each tube. This assay provides a rapid and accurate method for identification of carriers of the HLA B27 genotype.