

# The Genetics of Spondyloarthropathies

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## Abstract

**Introduction:** Spondyloarthropathies are a heterogenous group of rheumatic disorders that commonly present with axial skeleton or sacroiliac joints involvement. Ocular involvement like uveitis, iritis and conjunctivitis can be present in up to a third of the patients. Genetic factors play a part in the pathogenesis of spondyloarthropathies. Association with the HLA-B27 gene, especially that between ankylosing spondylitis and HLA-B27 antigenic positivity, is one of the strongest association seen between a disease and a Class I antigen. This paper aims to review the frequencies of HLA-B27 gene and its subtypes in different population groups, possible mechanisms leading to the development of joint inflammation and the risk it confers for development of spondyloarthropathies. **Methods:** The MEDLINE database was searched using keywords: HLA-B27, spondyloarthropathy, molecular mimicry, arthritogenic peptides, reactive arthritis and ankylosing arthritis. Related articles for selected papers were also consulted. Books on HLA-B27 and spondyloarthropathy were obtained through the NUS Medical Library's LINC system. **Results:** The genetic subtypes and susceptibility to development of disease vary in different population groups. Other HLA genes and non-HLA genes also play a part in the development of spondyloarthropathies, especially in those who are HLA-B27 negative. HLA-B27-positive relatives of spondyloarthritics have a higher risk of developing a similar condition. The presence of the HLA-B27 gene may serve as an aid to diagnosis or prognosis for clinicians. In juvenile arthritic patients, it is a poor prognostic factor, predicting for disease severity. It is also associated with poor outcomes for patients with anterior uveitis. However screening of asymptomatic individuals for the HLA-B27 gene is not recommended. **Conclusion:** The polygenic nature of the disease needs further elucidation and study.

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**Key words:** Ankylosing spondylitis, Enthesitis, HLA-B27 gene, Sacroiliitis, Uveitis

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