HLA-B*27 testing for ankylosing spondylitis

Record Status
This is a bibliographic record of a published health technology assessment. No evaluation of the quality of this assessment has been made for the HTA database.

Citation
HLA-B*27 testing for ankylosing spondylitis. Lansdale: HAYES, Inc., Genetic Testing Publication. 2011

Authors' objectives
Ankylosing spondylitis (AS) is a chronic inflammatory disease, the prototype of a group of disorders known collectively as spondyloarthritides (SpA). These disorders commonly feature axial skeletal involvement, peripheral arthritis, inflammation of the middle layer of the eye (uveitis), psoriasis, osteoporosis, and inflammatory bowel disease. Enthesitis—inflammation affecting areas where tendon and ligament attach to bone—is a key feature. At the site of enthesitis, bony erosion and growth of bony spurs may occur. These bony spurs can fuse to adjacent vertebral bodies, producing a condition called "bamboo spine." AS occurs in approximately 0.2% to 0.5% of Americans, and occurs worldwide with a prevalence of 0.1% to 1.4%. Disease occurrence is rare in African populations. Two to three times as many males as females are afflicted, with an average onset age of 26 years. Clinically, AS presents as back pain and stiffness, with the shoulders and hips often affected. The reference standard for diagnosis of AS is the modified New York criteria, which include radiologically evident sacroiliitis (inflammation of the sacroiliac joint) as a key criterion. Treatment of AS is designed to address symptoms of stiffness and joint pain and consists of exercise, medication, and in rare cases, surgery. Medications most commonly used to treat AS are nonsteroidal anti-inflammatory drugs (NSAIDs) and tumor necrosis factor alpha (TNF-) antagonists. No treatment has been shown to halt or reverse disease progression. Since the 1970s, studies have linked AS susceptibility to an allele of the major histocompatibility complex (MHC), class I, B gene called HLA-B*27, which is located on the short arm of chromosome 6 at band 21.3 (6p21.3). HLA genes encode cell surface proteins that function in the initiation of immune response by binding and presenting antigens to lymphocytes. The HLA-B*27 allele occurs in approximately 92% of white American AS patients and 50% of black American AS patients, as well as 8% and 4% of healthy white and black American populations, respectively. HLA-B*27 is reported to be associated with disease characteristics such as earlier age of disease onset, shorter delay in time from disease onset to accurate diagnosis, and greater prevalence of acute anterior uveitis. Although many studies link HLA-B*27 to AS, the role that HLA-B*27 may play in disease pathogenesis is not clear and is thought to be mediated by numerous other genetic and environmental factors. Results of twin studies suggest that heritability of susceptibility to disease is in excess of 90%; however, family studies indicate that < 50% of the entire genetic risk is due to HLA-B*27 alone. HLA-B*27 was initially defined using serological methods and was thought to represent one distinct allele of HLA. As DNA sequencing has been used to investigate the MHC in greater detail, however, it has been found that the HLA-B*27 allele comprises more than 90 subtypes, with more subtypes discovered regularly. Some published evidence suggests HLA-B*27 subtypes are differentially associated with disease risk or clinical manifestations of AS; research in this area is ongoing.

Final publication URL
The report may be purchased from: http://www.hayesinc.com/hayes/crd/?crd=12826

Indexing Status
Subject indexing assigned by CRD

MeSH
Hematologic Tests; HLA-B27 Antigens; Spondylitis, Ankylosing

Language Published
English
Country of organisation
United States

English summary
An English language summary is available.

Address for correspondence
HAYES, Inc., 157 S. Broad Street, Suite 200, Lansdale, PA 19446, USA. Tel: 215 855 0615; Fax: 215 855 5218 Email: hayesinfo@hayesinc.com

AccessionNumber
32011001475

Date abstract record published
02/11/2011