



# HLA-B27 DNA Test

Spondyloarthropathies are chronic inflammatory disorders that include ankylosing spondylitis, Reiter's syndrome, reactive arthritis, psoriatic arthritis, Crohn disease, and ulcerative colitis. This group of disorders is associated with human leukocyte antigen (HLA) B27 suggesting a possible autoimmune etiology. The strength of the association with the HLA-B27 allele varies among the different spondyloarthropathies and among ethnic groups. The strongest association is with ankylosing spondylitis. HLA-B27 (alleles B\*2701-B\*2723) is present in >90% of Caucasians with ankylosing spondylitis compared to an 8% frequency in the general Caucasian population.

Ankylosing spondylitis is the most common inflammatory disorder of the axial skeleton. Men are affected 2-3 times more frequently than women. Disease onset typically occurs in young adulthood manifesting as lower back pain and/or stiffness progressing to chronic pain with marked axial immobility or deformity. Sacroilitis is the hallmark feature of this disorder. In addition, ocular involvement, most commonly acute uveitis, is seen in 40% of patients, while 60% of patients have asymptomatic irritable bowel disease.

DNA testing for detection of HLA-B27 is extremely accurate and considered the gold standard. Identification of HLA-B27 by DNA testing supports the diagnosis of ankylosing spondylitis in symptomatic individuals and a negative result excludes the diagnosis. Early detection of ankylosing spondylitis through genetic and radiographic testing may provide an opportunity for treatment before permanent limitation of spinal mobility.

## Indications for HLA-B27 DNA Testing:

- Suspected clinical diagnosis of ankylosing spondylitis and/or related spondyloarthropathy
- Sacroilitis
- Inflammatory back pain for more than 3 months
- Limitation of motion of the lumbar spine and/or of chest expansion
- History of enthesitis, asymmetric arthritis, anterior uveitis, irritable bowel disease, and/or aortic insufficiency
- Ambiguous result from flow cytometry for HLA-B27
- Family history of ankylosing spondylitis

## Our HLA-B27 DNA Test Service Provides:

- PCR analysis for HLA-B27 (alleles B\*2701 to B\*2723)
- Detailed reports with genetic interpretation, recommendations, and education
- Free genetic counseling for physicians, patients, and families
- Free shipping via FedEx

## Specimen Requirements:

- 5 ml blood in an EDTA (lavender top) tube, room temperature

## Turnaround Time:

- 1-2 business days

Please call Kimball Genetics for more information.

**kimball genetics**

101 University Blvd., Suite 350, Denver, Colorado 80206 • Tel: 800-320-1807 • 303-320-1807 • Fax: 303-388-9220

© Kimball Genetics Inc. 9/03