Gene test information

HLA B27

- **Background**

  Ankylosing spondylitis is a chronic rheumatic disease which primarily affects the spine. The early symptomatology of the disease leads to movement limitations of the lumbar spine. Further on in the course of the disease, the symptoms may worsen to the point of hardening of the bones of the spine.

  The fist signs of the disease are frequently nonspecific and are generally not associated with the disease. There is generally a time period of 5-10 years between the initial symptoms and a definite diagnosis. However, an early diagnosis is of crucial importance, since the course of the disease can be influenced better when the disease is diagnosed early on.

  The majority (>90%) of patients with ankylosing spondylitis carry the HLA-B27 gene, which however is only present in 8% of the “healthy” population. Thus the molecular genetic detection of HLA-B27 has a high diagnostic significance for the probability of developing ankylosing spondylitis.

- **Indications for testing**

  - Suspected clinical diagnosis of ankylosing spondylitis and/or related spondyloarthropathy
  - 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

References:

Ramos M, López de Castro JA. HLA-B27 and the pathogenesis of spondyloarthritis. Tissue Antigens. 2002;60:191-205.

Suhler EB, Martin TM, Rosenbaum JT. HLA-B27-associated uveitis: overview and current perspectives. Curr Opin Ophthalmol. 2003;14:378-83.

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