Ankylosing Spondylitis Genotyping (HLA-B27)

Indications for Ordering

Use as adjunct test for diagnosis in individuals symptomatic for ankylosing spondylitis (AS)
- Should not be performed in asymptomatic individuals or those who do not have high suspicion for AS based on specific symptoms

Test Description

Ankylosing Spondylitis (HLA-B27) Genotyping
- Polymerase chain reaction/fluorescence monitoring
- Qualitative flow cytometry

Tests to Consider

Ankylosing Spondylitis (HLA-B27) Genotyping 0050392
- Not a diagnostic test for AS
  - May assist in the diagnosis of the condition only if other clinical signs and symptoms are present
- Test should not be performed for prenatal diagnosis of AS; a positive result is not predictive for the disorder

HLA-B27 0095840
- Not a diagnostic test for AS, juvenile rheumatoid arthritis, or Reiter syndrome
  - May assist in the diagnosis of these conditions only if other clinical signs and symptoms are present

Disease Overview

Prevalence: based on ethnicity
Gender: female: male, 1:2

Symptoms
- Lower back pain and stiffness
- Arthritis of spine, knees, hips, shoulders
- Fever
- Fatigue, malaise, weight loss
- Anemia
- Other potential organs involved
  - Eyes
  - Lungs
  - Kidneys
  - Heart

- Confirmation of diagnosis requires classic x-ray findings
  - Blurring of bony margins of joints
  - Bilateral sacroiliac involvement
  - Patchy sclerosis with superficial bony erosions
  - Late squaring of vertebral bodies with bamboo spine and complete ankylosis

Genetics

Gene: HLA-B27
Inheritance: multifactorial
Structure/Function
Located on chromosome 6
- Encodes for protein with many functions
  - Phagocytosis in infectious diseases
  - Graft rejection
  - Ability to recognize self from nonself in the immune system
- Strong genetic association between HLA-B27 and AS

Variants
At least 49 subtypes identified

Test Interpretation

Sensitivity/Specificity
- Clinical sensitivity: 90%
- Clinical specificity in an unaffected individual without a family history: <1%
- Analytical sensitivity/specificity: >99%

Results
- Positive: one copy of HLA-B27 detected
  - Associated with AS and related disorders (Reiter syndrome, anterior uveitis, psoriatic arthritis, and inflammatory bowel disease)
- Negative: no copy of HLA-B27 identified
  - Does not rule out AS, as 10% of affected individuals lack this antigen
  - Clinical sensitivity: 90%

Limitations
- Contraindicated for prenatal or carrier testing
- Other HLA types will not be detected
- Rare diagnostic errors can occur due to probe-site variants