

# 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

*HLA-B27*

- 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