

# Ankylosing Spondylitis Genotyping (*HLA-B27*)

## Indications for Ordering

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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

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Ankylosing Spondylitis (*HLA-B27*) Genotyping

- Polymerase chain reaction/fluorescence monitoring

*HLA-B27*

- Qualitative flow cytometry

## Tests to Consider

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[Ankylosing Spondylitis \(\*HLA-B27\*\) Genotyping 0050392](#)

- May assist in the diagnosis of ankylosing spondylitis, juvenile rheumatoid arthritis, and Reiter's syndrome
- Tests are not diagnostic for these disorders and should be correlated with other clinical signs and symptoms

[HLA-B27 0095840](#)

- Flow cytometry is the preferred test for most patients since sensitivity and specificity are nearly equivalent to the *HLA-B27* PCR genotyping test

## Disease Overview

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**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

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**Genes** – *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 non-self in the immune system
- Strong genetic association between *HLA-B27* and AS

### Mutations

At least 49 subtypes identified

## Test Interpretation

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### 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 mutations