Ankylosing Spondylitis (HLA-B27) Genotyping

Ankylosing spondylitis (AS) is a chronic form of arthritis in which the sacroiliac joints become inflamed, leading to back pain and, in some cases, the formation of bone between vertebrae (ie, ankylosis). Although spinal involvement is typical to AS, other areas of the body (eg, shoulders, hips, knees, etc.) may also become stiff or inflamed. Although no specific cause of AS has yet been identified, data support a strong correlation between AS and the presence of HLA-B27; 80-90% of AS patients are HLA-B27 positive, compared to 5-10% of the general population.\(^1\)

### Genetics

#### Gene

**HLA-B27 group alleles**

**Allele(s)**

More than 213 known alleles\(^2\)

#### Inheritance

Codominant

### Prevalence

Overall prevalence in North America is 6-8\(^3\) but varies by ethnicity.\(^1\) Notably, there is a descending gradient from north to south of the HLA-B27 allelic prevalence; in particular, the ancestral HLA-B*27:05 allele frequency ranges from >20% in Northern Europe to <1% in sub-Saharan Africa.

### Test Interpretation

#### Sensitivity/Specificity

- Analytic sensitivity/specificity: >99%
- Clinical sensitivity: 90%
- Clinical specificity: <1% in unaffected individuals with no family history of AS

#### Results

<table>
<thead>
<tr>
<th>Result</th>
<th>Interpretive Note</th>
</tr>
</thead>
<tbody>
<tr>
<td>Positive: HLA-B27 was detected</td>
<td>Result supports a clinical diagnosis of AS or another related disorder (eg, Reiter syndrome, anterior uveitis, psoriatic arthritis, and inflammatory bowel disease)</td>
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<tr>
<td>Negative: HLA-B27 was not detected</td>
<td>Does not rule out AS; refer to clinical sensitivity</td>
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</tbody>
</table>

### Limitations

- Contraindicated for prenatal or carrier testing
- Rare diagnostic errors can occur due to primer site variants.
This test does not rule out alleles HLA-B*27:06 or HLA-B*27:09, which are not associated with spondyloarthropathies.

Certain rare alleles present in <1% of the population will not be detected.

Other rare or uncharacterized alleles are possible and may lead to false positive or false negative results.

References


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