

Ankylosing Spondylitis (HLA-B27) Genotyping

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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 (i.e., ankylosis). Although spinal involvement is typical to AS, other areas of the body (e.g., 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 alleles²

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

| Result | Interpretive Note |
|------------------------------------|---|
| Positive: HLA-B27 was detected | Result supports a clinical diagnosis of AS or another related disorder (e.g., Reiter syndrome, anterior uveitis, psoriatic arthritis, and inflammatory bowel disease) |
| Negative: HLA-B27 was not detected | Does not rule out AS; refer to clinical sensitivity |

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Method: Polymerase Chain Reaction (PCR)/Fluorescence Monitoring

- Not a diagnostic test for AS
- May assist in the diagnosis of AS only if other clinical signs and symptoms are present
- Test should not be performed for prenatal diagnosis of AS because a positive result is not predictive for the disorder

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

1. Wenker KJ, Quint JM. [Ankylosing spondylitis](#). In: *StatPearls*, StatPearls Publishing. Updated Apr 2022; accessed Aug 2022.
2. Khan MA. [An update on the genetic polymorphism of HLA-B*27 with 213 alleles encompassing 160 subtypes \(and still counting\)](#). *Curr Rheumatol Rep*. 2017;19(2):9.
3. Profaizer T, Dibb K, Bethers H, et al. [Comparison of next-generation sequencing-based human leukocyte antigen typing with clinical flow cytometry and allele-specific PCR melting assays for HLA-B27 genotyping](#). *J Appl Lab Med*. 2021;6(5):1221-1227.

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