

NEW TEST

[Click for Pricing](#)

HLA-A29 Genotyping, Birdshot Chorioretinopathy

3018058, HLA A29

Specimen Requirements:

Patient Preparation:

Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD solution A or B).

Specimen Preparation: Transport 5 mL whole blood. (Min: 3 mL).

Transport Temperature: Refrigerated

Unacceptable Conditions: Specimens collected in green (sodium or lithium heparin).

Remarks:

Stability: Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Methodology: Polymerase Chain Reaction (PCR)/Sequence-Specific Oligonucleotide Probe Hybridization

Performed: Mon-Fri

Reported: 5-7 days

Note:

CPT Codes: 81381

New York DOH Approval Status: This test is New York DOH approved.

Interpretive Data:

Background Information for HLA-A29 Genotyping for Birdshot Chorioretinopathy:

Characteristics: Birdshot chorioretinopathy (BSCR) is a progressive, bilateral, chronic autoimmune inflammatory disease of the eye. It is characterized by posterior uveitis with yellow-white choroid lesions in the fundus that resemble a shotgun splatter. Patients with BSCR may experience decreased vision, floaters, nyctalopia, dyschromatopsia, glare, and photopsia.

Prevalence: BSCR comprises up to 1.5%, of uveitis cases. Its prevalence ranges from 0.1 to 0.6 cases per 100,000 individuals across Europe and the U.S. Particularly prevalent in Caucasians, it is frequently diagnosed in individuals of Northern European ancestry, predominantly affecting middle-aged individuals, (mean onset age of 53 years), with a higher incidence among females.

Inheritance: Multifactorial.

Cause: The disease-causing factors are unknown. HLA-A29 is strongly associated with BSCR, with approximately 80-98% of patients testing positive, compared to about 7% positivity in healthy individuals across different ethnicities. This suggests a negative predictive value of HLA-A29 typing as high as 99%. HLA-A29 is associated with a 50-224 times greater relative risk of developing the disease.

Clinical Sensitivity: Approximately 80-98%, depending on ethnicity.

Methodology: Polymerase Chain Reaction/Sequence-Specific Oligonucleotide Probe Hybridization.

Analytical Sensitivity and Specificity: >99 percent.

Limitations: Other genetic and nongenetic factors that influence BSCR are not evaluated. Other rare, or novel alleles may occur which may lead to false-positive or false-negative results. In cases where an HLA allele cannot be resolved unambiguously, the allele assignment will be reported as the most common, based on allele frequencies from the Common, Intermediate and Well-Documented Alleles Catalogue version 3.0.0 (Hurley CK, et al, 2020).

Alleles tested: HLA-A*29 alleles.

Disclaimer Information:

This test was developed and its performance characteristics determined by the Histocompatibility & Immunogenetics Laboratory at the University of Utah Health under the accreditation guidelines from the American Society for Histocompatibility and Immunogenetics (ASHI).

Performed at: Histocompatibility and Immunogenetics Laboratory, University of Utah Health, 417 Wakara Way, Suite 3220, Salt Lake City, UT 84108.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Reference Interval:

By Report

HOTLINE NOTE: Refer to the Hotline Test Mix for interface build information.