

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB: 1/20/1987
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

HLA-A29 Genotyping, Birdshot Chorioretinopathy

ARUP test code 3018058

HLA Class I, Locus A*, Allele 1 02:01

HLA Class I, Locus A*, Allele 2 03:01

HLA A29 Interpretation

See Note

Negative for HLA-A*29
HLA-A*29, which is strongly associated with birdshot
chorioretinopathy (BSCR), was not detected. Risk for BSCR may be
reduced, but not completely eliminated, since up to 2-20% of
patients may not carry the HLA-A*29 allele. Medical screening
and management of this patient should rely on clinical findings.

H=High, L=Low, *=Abnormal, C=Critical

**INTERPRETIVE INFORMATION: HLA-A29 Genotyping, 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 percent 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 percent of patients testing positive, compared to about 7 percent positivity in healthy individuals across different ethnicities. This suggests a negative predictive value of HLA-A29 typing as high as 99 percent. HLA-A29 is associated with a 50-224 times greater relative risk of developing the disease.

Clinical Sensitivity: Approximately 80-98 percent, 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 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.

H=High, L=Low, *=Abnormal, C=Critical

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
HLA Class I, Locus A*, Allele 1	24-295-400469	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
HLA Class I, Locus A*, Allele 2	24-295-400469	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
HLA A29 Interpretation	24-295-400469	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 24-295-400469
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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