

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:** Male  
**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 01:01

HLA Class I, Locus A\*, Allele 2 29:01

**HLA A29 Interpretation**

See Note

Positive for HLA-A\*29  
HLA-A\*29, which is strongly associated with birdshot  
chorioretinopathy (BSCR), was detected. This result is  
supportive of a clinical diagnosis of BSCR, but by itself does  
not establish a diagnosis. Medical screening and management of  
this patient should be based on clinical findings.

**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: 24-229-102865  
Patient Identifiers: 01234567890ABCD, 012345  
Visit Number (FIN): 01234567890ABCD  
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**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.

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
HLA Class I, Locus A*, Allele 1	24-229-102865	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
HLA Class I, Locus A*, Allele 2	24-229-102865	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
HLA A29 Interpretation	24-229-102865	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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