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PATIENT REPORT

Patient Age/Sex: 49 years Female

Specimen Collected: 22-Mar-24 14:54

HLA-B51 Genotyping, Behcet Disease **|Received: 22-Mar-24 14:54** **Report/Verified: 22-Mar-24 14:56**

Procedure	Result	Units	Reference Interval
HLA Class I,Locus B*,Allele 1	07:02		
HLA Class I,Locus B*,Allele 2	51:01		
Behcet HLA Interpretation	See Note [f1 i1]		

Result Footnote

f1: Behcet HLA Interpretation
Positive
Positive for HLA-B*51 HLA-B*51, which is strongly associated with Behcet's disease, was detected. HLA-B*51 positivity by itself does not establish a diagnosis for Behcet's disease. Medical screening and management of this individual should rely on clinical findings.

Test Information

i1: Behcet HLA Interpretation
BACKGROUND INFORMATION: HLA-B51 Genotyping, Behcet Disease

Characteristics: Behcet disease (BD) is a multisystem chronic inflammatory disease, caused by vasculitis of arteries and veins of all sizes, involving the skin, mucosa, eyes, joints, cardiovascular, gastrointestinal, and nervous systems.

Prevalence: BD shows worldwide distribution, but it is most common in the Mediterranean basin, Middle East, and East Asian countries. Prevalence is high in Iran and Turkey with 80-370 cases/100,000 individuals, and comparatively low in the U.S. with 5.2 cases/100,000 individuals.

Inheritance: Multifactorial.

Cause: The disease-causing factors are unknown. HLA-B*51 is strongly associated with BD with approximately 60% of patients being positive, as opposed to about 15% positivity in healthy individuals across different ethnicities. Due to low specificity, HLA-B*51 positivity is not diagnostic for BD. It may, however, affect clinical phenotypes of BD as it is more common in patients with ocular involvement, and less common in patients with gastrointestinal involvement.

Clinical Sensitivity: Approximately 50-80 percent, depending on ethnicity.

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

Analytical Sensitivity and Specificity: >99 percent.

Limitations: Other genetic and nongenetic factors that influence BD 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 can not be resolved

Test Information

i1: Behcet HLA Interpretation

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-B*51 alleles.

Disclaimer Information:

This test was developed and its performance characteristics determined by the Histocompatibility & Immunogenetics Laboratory at the University of Utah Health. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research. Histocompatibility & Immunogenetics Laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA-88) as qualified to perform high complexity clinical laboratory testing.

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

CLIA Number: 46D0679773

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