

# TEST SUMMARY: SOMATIC INBORN ERRORS OF IMMUNITY GENETIC PANEL

Nearly 500 genes underlie primary immunodeficiencies (PID). Yet, many patients have no pathogenic variants reported after testing by germline sequencing, including whole exome (WES) and whole genome (WGS) sequencing. Some of these cases will be due to somatic variants in the inborn errors of immunity (IEI) genes, increasingly recognized as an important cause of immunodeficiency. However, detecting somatic variants is challenging; WES/WGS does not sequence deeply enough to detect variants with low allele frequencies. Furthermore, the relevant immunity genes mostly do not overlap with existing cancer genetic panels. Finally, the turnaround time is generally several weeks and these patients are frequently acutely ill. Thus, in collaboration with members from the NICER Consortium, we designed a somatic NGS genetic panel to fill this clinical testing gap: this test 1) sequences 69 IEI genes at ~5000x mean read depth, 2) with 98.5% sensitivity for variant allele frequency (VAF) of 2% or greater and 100% sensitivity for VAF of 3% or greater, and 3) has a turnaround time of less than 1 week.

Please visit MachaonDiagnostics.com for further information.

References: 1. Aluri J and Cooper M. (2023) Semin Immunol.67:101761. 2. Lopez-Nevado M et al. (2021) Front Immunol.12:656356. 3. Tanaka N et al. (2011) Arthritis Rheum.63(11):3625–32. 4. Beck et al (2020) N Engl J Med. 2020;383(27):2628–38. 5. Aluri et al. (2021) Blood. 2021;137(18):2450–62.

ASR Disclaimer: This test was developed and its performance characteristics determined by Machaon Diagnostics. It has not been cleared or approved by the US Food and Drug Administration; the FDA does not require this test to go through premarket FDA review. This test has been validated by Machaon Diagnostics and is for clinical purposes. It should not be regarded as investigational or for research. Machaon Diagnostics is accredited by the College of American Pathologists (CAP) and accredited under the federal Clinical Laboratory Improvement Amendments (CLIA) program as qualified to perform high complexity clinical laboratory testing.

## **WHY CHOOSE US?**

- 𝒮 Fastest turnaround in the US
- 🔗 Clinical consultation
- 🛇 Critical Results called to physician

#### SPECIMEN REQUIRMENTS

3mL EDTA Whole Blood

## STABILITY

Room Temp: 1 month Refrigerated: 1 month

#### **CPT CODES** 81443

## **METHODOLOGY**

NGS; 69 genes have been sequenced and analyzed for this panel

### **TURNAROUND TIMES**

Routine TAT: <1 week

## **ASSOCIATED TESTING**

Soluble IL-2R alpha (CD25) Level CXCL9 Level IL-18 Level HLH Extended Genetic Panel 3.0



## **ABOUT US:**

Machaon Diagnostics is a clinical reference laboratory, specializing in coagulation, platelets, complement, genetics and rare disease.

2023 Eighth Street Berkeley, CA 94710

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