MACHAON DIAGNOSTICS

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.

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

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.

V:24FEB2025

WHY CHOOSE US?

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

ABOUT US:

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

2023 Eighth Street
Berkeley, CA 94710
800-566-3462
510-839-5600
510-839-6153
info@machaondiagnostics.com
MachaonDiagnostics.com