MACHAON DIAGNOSTICS

TEST SUMMARY

Dysfibrinogenemia Genetic Panel

This test sequences the exons plus 5bp of the flanking introns of the FGA, FGB, and FGG genes. Additionally, this test was designed to detect the recurrent 11kb deletion in FGA. Sanger sequencing may be used to confirm variants as needed. This test will detect inherited but not acquired dysfibrinogenemia. Mutations in FGA, FGB, or FGG can cause dysfibrinogenemia or afibrinogenemia. Depending on the effect of the mutation, there may be a bleeding, thrombotic, or asymptomatic phenotype. This test may help confirm the diagnosis.

Please visit Machaondiagnostics.com for further information.

Limitations: This test will not detect variants located outside of the targeted DNA regions. This test is not optimized to detect chimerism or somatic mosaicism. This test will detect small indels but may miss larger deletions or duplications. Balanced structural variants will not be detected unless specifically targeted by a custom PCR assay.

References: Neerman-Arbez, M et al. The 11 kb FGA deletion responsible for congenital afibrinogenaemia is mediated by a short direct repeat in the fibrinogen gene cluster. Europ. J. Hum. Genet. 7: 897-902, 1999.

SPECIMEN REQUIRMENTS
3mL EDTA Whole Blood

STABILITY
Room Temp: 1 month
Refrigerated: 1 month

CPT CODE
81479x3

METHODOLOGY
NGS

TURNAROUND TIMES
Routine TAT: 5 days (M-F)
STAT TAT: <48 hours (M-F)

ASSOCIATED TESTS
Dysfibrinogenemia Functional Panel

DRAW KITS AVAILABLE

ABOUT US:

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

V:18FEB2025

WHY CHOOSE US?

- Fastest turnaround in the US
- Draw kits (includes free shipping)
- Clinical consultation
- Critical Results called to physician

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