

Macro/Microthrombocytopenia Gene Panel, Next-Generation Sequencing, Varies

Test ID: GNMTC

Explanation:

On the effective date, GNMTC will be obsolete. The recommended alternative is below.

Recommended Alternative Test:

Platelet Disorders, Comprehensive Gene Panel, Next-Generation Sequencing, Varies

Test ID: GNPLT

Methodology:

Sequence Capture and Targeted Next-Generation Sequencing (NGS) followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing

Genetics Information:

This test utilizes next-generation sequencing to detect single nucleotide and copy number variants in 70 genes associated with a variety of hereditary platelet disorders: *ABCC4*, *ABCG5*, *ABCG8*, *ACTB*, *ACTN1*, *ANKRD26*, *ANO6*, *AP3B1*, *AP3D1*, *ARPC1B*, *BLOC1S3*, *BLOC1S5*, *BLOC1S6*, *CDC42*, *CYCS*, *DIAPH1*, *DTNBP1*, *ETV6*, *FERMT3*, *FLI1*, *FLNA*, *FYB1*, *GATA1*, *GATA2*, *GFI1B*, *GNE*, *GP1BA*, *GP1BB*, *GP6*, *GP9*, *HOXA11*, *HPS1*, *HPS3*, *HPS4*, *HPS5*, *HPS6*, *IKZF5*, *ITGA2B*, *ITGB3*, *KDSR*, *LYST*, *MASTL*, *MECOM*, *MPIG6B*, *MPL*, *MYH9*, *NBEA*, *NBEAL2*, *ORAI1*, *P2RY1*, *P2RY12*, *PLA2G4A*, *PLAU*, *PRKACG*, *PTGS1*, *RASGRP2*, *RBM8A*, *RUNX1*, *SLFN14*, *SRC*, *STIM1*, *STXBP2*, *TBXA2R*, *TBXAS1*, *THPO*, *TPM4*, *TUBB1*, *VIPAS39*, *VPS33B*, and *WAS*.

See [Targeted Genes and Methodology Details for Platelet Disorders Comprehensive Gene Panel](#) and Method Description for additional details.

Identification of a disease-causing variant may assist with diagnosis, prognosis, clinical management, recurrence risk assessment, familial screening, and genetic counseling for a variety of hereditary platelet disorders.

Reflex Tests:

Test ID	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Test	Yes	No

Reference Values:

An interpretive report will be provided.

Specimen Requirements:

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD)

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot.**

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated

Specimen Type: Skin biopsy

Supplies: Fibroblast Biopsy Transport Media (T115)

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Cultured fibroblasts

Container/Tube: T-25 flask

Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory. Cultured cells from a prenatal specimen will not be accepted.

Specimen Stability Information: Ambient (preferred)/Refrigerated (<24 hours)

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Stability Information:

Specimen Type	Temperature	Time
Varies	Varies	

CPT Code:

81443

88233-Tissue culture, skin, solid tissue biopsy (if appropriate)

88240-Cryopreservation (if appropriate)

Day(s) Performed: Varies

Report Available: 28 to 42 days

Questions

Contact Melissa Lonzo, Laboratory Resource Coordinator, at 800-533-1710.