

## 3-Methylglutaconic Aciduria Panel, Varies

**Test ID:** 3MGAP

### Explanation:

On the effective date, the changes below will occur, including additional acceptable specimen types and possible reflex tests.

### Reflex Tests added:

| Test ID | Reporting Name                   | Available Separately | Always Performed |
|---------|----------------------------------|----------------------|------------------|
| FIBR    | Fibroblast Culture               | Yes                  | No               |
| CRYOB   | Cryopreserve for Biochem Studies | Yes                  | No               |

### Method Name updated:

| Current Method Name   | New Method Name  |
|---|--|
| Custom Sequence Capture and Targeted Next-Generation Sequencing followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing | Sequence Capture and Targeted Next-Generation Sequencing followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing |

### Additional Specimens accepted:

| Current Specimens Accepted | New Specimens Accepted   |
|----------------------------|--|
| Whole Blood                | Whole Blood<br>Skin Biopsy<br>Cultured Fibroblasts<br>Blood Spot<br>Saliva |

### New Specimen Requirements:

Submit only 1 of the following specimens:

|                       |   |
|-----------------------|---|
| <b>Specimen Type:</b> | Whole Blood                             |
| <b>Preferred:</b>     | Lavender top (EDTA) or yellow top (ACD) |
| <b>Acceptable:</b>    | Any anticoagulant                       |

|  |  |
|--|--|
| <b>Specimen Volume:</b>                | 3 mL   |
| <b>Collection Instructions:</b>        | <ol style="list-style-type: none"> <li>1. Invert several times to mix blood.</li> <li>2. Send specimen in original tube. Do not aliquot.</li> </ol>  |
| <b>Specimen Stability Information:</b> | Ambient (preferred) 4 days/Refrigerated 14 days  |
| <b>Minimum Volume:</b>                 | 1 mL   |
| <b>Specimen Type:</b>                  | <b>Skin Biopsy</b>   |
| <b>Supplies:</b>                       | Fibroblast Biopsy Transport Media (T115)   |
| <b>Container/Tube:</b>                 | Sterile container with any standard cell culture media (e.g., minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.  |
| <b>Specimen Volume:</b>                | 4-mm punch   |
| <b>Specimen Stability Information:</b> | Refrigerated (preferred)/Ambient   |
| <b>Specimen Type:</b>                  | <b>Cultured fibroblast</b>   |
| <b>Container/Tube:</b>                 | T-25 flask   |
| <b>Specimen Volume:</b>                | 2 Flasks   |
| <b>Collection Instructions:</b>        | <p>Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory.</p> <p>Cultured cells from a prenatal specimen will not be accepted.</p>   |
| <b>Specimen Stability Information:</b> | Ambient (preferred)/Refrigerated (<24 hours)   |
| <b>Additional information</b>          | A separate culture charge will be assessed under FIBR / Fibroblast Culture, Tissue. An additional 4 weeks is required to culture fibroblasts before genetic testing can occur.   |
| <b>Specimen Type:</b>                  | <b>Blood spot</b>  |
| <b>Supplies:</b>                       | Card-Blood Spot Collection (Filter Paper) (T493)   |
| <b>Preferred:</b>                      | Collection card (Whatman Protein Saver 903 Paper)  |
| <b>Acceptable</b>                      | PerkinElmer 226 (formerly Ahlstrom 226) filter paper, or blood spot collection card  |
| <b>Specimen Volume:</b>                | 5 Blood spots  |
| <b>Collection Instructions:</b>        | <ol style="list-style-type: none"> <li>1. An alternative blood collection option for a patient older than 1 year of age is fingerstick.</li> <li>2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.</li> <li>3. Do not expose specimen to heat or direct sunlight.</li> <li>4. Do not stack wet specimens.</li> <li>5. Keep specimen dry.</li> </ol> |
| <b>Specimen Stability Information:</b> | Ambient (preferred)/Refrigerated   |
| <b>Additional information</b>          | Due to lower concentration of DNA yielded from blood spots, it is possible that additional specimen may be required to complete testing.   |
| <b>Specimen Type:</b>                  | <b>Saliva</b>  |
| <b>Patient Preparation:</b>            | Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.  |

|  |   |
|--|---|
| <b>Supplies:</b>                       | Saliva Swab Collection Kit (T786)   |
| <b>Specimen Volume</b>                 | 1 Swab  |
| <b>Collection Instructions:</b>        | Collect and send specimen per kit instructions.   |
| <b>Specimen Stability Information:</b> | Ambient 30 days   |
| <b>Additional information</b>          | Due to lower concentration of DNA yielded from saliva, it is possible that additional specimen may be required to complete testing. |

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### ***Testing Duration Changed:***

| <b>Current Report Available</b> |
|---------------------------------|
| 3 to 4 weeks                    |

| <b>New Report Available</b> |
|-----------------------------|
| 4 to 6 weeks                |

### ***Additional CPT Codes Added:***

| <b>Current CPT Codes</b> |
|--------------------------|
| 81443                    |

| <b>New CPT Codes</b>   |
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| 81443<br>88233-Tissue culture, skin, solid tissue biopsy (if appropriate)<br>88240-Cryopreservation (if appropriate) |

## **Questions**

Contact Michelle Rath, Laboratory Technologist Resource Coordinator at 800-533-1710.