

FILE DEFINITION TEST CHANGE

Notification Date: January 3, 2022 Effective Date: February 3, 2022

Urea Cycle Disorders Gene Panel, Varies

Test ID: UCDP

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

Λ

Current Method Name

CRYOB Cryopreserve for Biochem Studies	Yes	No				
Method Name updated:						

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 Cultured Fibroblasts Blood Spot Saliva

New Specimen Requirements:

Submit only 1 of the following specimens:

Specimen Type: Whole Blood

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 3 mL **Collection Instructions:** 1. Invert several times to mix blood.

2. Send specimen in original tube. Do not aliquot.

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated 14 days

Minimum Volume: 1 mL

Specimen Type: Cultured fibroblast

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 FIBR / Fibroblast Culture,

Tissue. An additional 4 weeks is required to culture fibroblasts before

genetic testing can occur.

Specimen Type: Blood spot

Supplies: Card-Blood Spot Collection (Filter Paper) (T493)

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable PerkinElmer 226 (formerly Ahlstrom 226) filter paper, or blood spot collection card

Specimen Volume: 5 Blood spots

Collection Instructions: 1. An alternative blood collection option for a patient older than 1 year of age is

fingerstick.

2. Let blood dry on the filter paper at ambient temperature in a horizontal position for

a minimum of 3 hours.

3. Do not expose specimen to heat or direct sunlight.

4. Do not stack wet specimens.

5. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Additional information Due to lower concentration of DNA yielded from blood spots, it is possible that

additional specimen may be required to complete testing.

Specimen Type: Saliva

Patient Preparation: Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.

Supplies: Saliva Swab Collection Kit (T786)

Specimen Volume 1 Swab

Collection Instructions: Collect and send specimen per kit instructions.

Specimen Stability Information: Ambient 30 days

Additional information Due to lower concentration of DNA yielded from saliva, it is possible that additional

specimen may be required to complete testing.

Testing Duration Changed:

Current Report Available		New Report Available
3 to 4 weeks		4 to 6 weeks

Additional CPT Codes Added:

Current CPT Codes	New CPT Codes
81443	81443 88233-Tissue culture, skin, solid tissue biopsy (if appropriate) 88240-Cryopreservation (if appropriate)

Questions

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