

Notification Date: November 19, 2024 Effective Date: December 24, 2024

Prader-Willi/Angelman Syndrome, Molecular Analysis, Varies

Test ID: PWAS

Explanation: On the effective date, this assay will incorporate changes to the Specimen Required, as shown below. The Whole Blood specimen type will no longer accept any anticoagulant and will be limited to the anticoagulants shown below. In addition, all Specimen Types will include updates to the Stability and/or Additional Information fields.

Current Specimen Required

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Preferred: Lavender top (EDTA) tube or yellow top

(ACD) tube

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)/Refrigerated

Prenatal Specimens

Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 20 mL

Specimen Stability Information: Refrigerated

(preferred)/Ambient

Specimen Type: Confluent cultured cells

Container/Tube: T-25 flask Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured

cells from another laboratory.

Specimen Stability Information: Ambient

(preferred)/Refrigerated

New Specimen Required

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

Submit only 1 of the following specimens:

Specimen Type: Whole blood

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

Acceptable: None 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 4 days/Frozen 4 days

Additional Information:

- 1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
- 2. To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.

PRENATAL SPECIMENS

Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type: Amniotic fluid

Container/Tube: Amniotic fluid container

Specimen Volume: 20 mL

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

Additional Information:

- 1. Specimens are preferred to be received within 24 hours of collection. Culture and/or extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
- 2. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid. An additional 2 to 3 weeks is required to culture amniotic fluid before genetic testing can occur.
- 3. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type: Confluent cultured amniocytes

Container/Tube: T-25 flasks Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured

amniocytes from another laboratory.

Specimen Stability Information: Ambient (preferred) <24

hours/Refrigerated <24 hours **Additional Information:**

- 1. Specimens are preferred to be received within 24 hours of collection. Culture and/or extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
- 2. A separate culture charge will be assessed under CULAF / Culture for Genetic Testing, Amniotic Fluid.
- 3. All prenatal specimens must be accompanied by a maternal blood specimen; order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Questions

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