MAYO CLINIC LABORATORIES

TEST CHANGE

Notification Date: December 30, 2025
Effective Date: February 3, 2026

Uniparental Disomy, Varies

Test ID: UNIPD

Explanation: On the effective date, extracted DNA will be acceptable for testing. Formatting of acceptable specimen types will also be standardized. Reflex testing will be updated to include MATCC, Maternal Cell Contamination testing for cord blood specimens.

Current Algorithm	New Algorithm
Polymerase chain reaction amplification of microsatellite markers on the chromosome of interest are used to test DNA from the parents and the child for the presence of uniparental disomy. **For prenatal specimens only:** If an amniotic fluid specimen or cultured amniocytes is received, amniotic fluid culture for genetic testing will be performed at an additional charge. If a chorionic villus specimen or cultured chorionic villi is received, fibroblast culture for a genetic test will be performed at an additional charge. For more information see: -Prader-Willi and Angelman Syndromes: Laboratory Approach to Diagnosis -Beckwith-Wiedemann and Russell-Silver Syndromes: Laboratory Approach to Diagnosis	Polymerase chain reaction amplification of microsatellite markers on the chromosome of interest are used to test DNA from the parents and the child for the presence of uniparental disomy. **For prenatal specimens:** If an amniotic fluid specimen or cultured amniocytes is received, amniotic fluid culture for genetic testing will be performed at an additional charge. If a chorionic villus specimen or cultured chorionic villi is received, fibroblast culture for a genetic test will be performed at an additional charge. For any prenatal specimen that is received, maternal cell contamination testing will be performed at an additional charge. **Cord blood:** For cord blood specimens that have an accompanying maternal blood specimen, maternal cell contamination studies will be performed at an additional charge. For more information see: -Prader-Willi and Angelman Syndromes: Laboratory Approach to Diagnosis -Beckwith-Wiedemann and Russell-Silver Syndromes: Laboratory Approach to Diagnosis

Current Testing Requirements	New Testing Requirements
None	**All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on both the prenatal specimen and maternal specimen as separate orders.

Current Reflex Tests			
Test ID	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Testing	Yes	No
CULAF	Amniotic Fluid Culture/Genetic Testing	Yes	No

New Reflex Tests			
Test ID	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Testing	Yes	No
CULAF	Amniotic Fluid Culture/Genetic Testing	Yes	No
MATCC	Maternal Cell Contamination, B	Yes	No

Current Specimen Required

Specimens from both parents and the child or fetus are recommended for optimal interpretation of results. Each specimen must have a separate order for this test. Only the proband specimen will be charged.
Testing can be performed if only one parent specimen is submitted, however, biparental inheritance and some types of uniparental disomy (UPD) cannot be definitively established in the absence of one parent. Additionally, there is a higher likelihood for uninformative or inconclusive results.
If all required specimens are not received within one month of ordering, testing will be canceled.

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: Any anticoagulant
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

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

New Specimen Required

Specimens from both parents and the child or fetus are recommended for optimal interpretation of results. Each specimen must have a separate order for this test. Only the proband specimen will be charged.
Testing can be performed if only one parent specimen is submitted, however, biparental inheritance and some types of uniparental disomy (UPD) cannot be definitively established in the absence of one parent. Additionally, there is a higher likelihood for uninformative or inconclusive results.
If all required specimens are not received within one month of ordering, testing will be canceled.

Patient Preparation: A previous hematopoietic stem cell transplant from an allogenic donor will interfere with testing. For information about testing patients who have received a hematopoietic stem cell transplant, call 800-533-1710.

Submit only 1 of the following specimens:

Specimen Type: Whole blood
Container/Tube: Lavender top (EDTA) or 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**.
3. Whole blood collected postnatal from an umbilical cord is also acceptable. See Additional Information
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 are met, the requested volume must be submitted.

Specimen Stability Information: Refrigerated (preferred)/Ambient
Additional information: If amniotic fluid or culture amniotic fluid is received, CULAF / Culture for Genetic Testing, Amniotic Fluid will be added at an additional charge.

Specimen Type: Chorionic villi (CVS)
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 mg
Specimen Stability Information: Refrigerated
Additional Information: If CVS or cultured CVS is received, CULFB / Fibroblast Culture for Biochemical or Molecular Testing will be added at an additional charge.

Acceptable:
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 (<24 hours)

Testing may be canceled if DNA requirements are inadequate.
3. For postnatal umbilical cord whole blood specimens, maternal cell contamination studies are recommended to ensure test results reflect that of the patient tested. A maternal blood specimen is required to complete maternal cell contamination studies. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on both the cord blood and maternal blood specimens under separate order numbers.

Specimen Type: Extracted DNA
Container/Tube:
Preferred: Screw Cap Micro Tube, 2mL with skirted conical base
Acceptable: Matrix tube, 1mL
Collection Instructions:
1. The preferred volume is at least 100 mcL at a concentration of 75 ng/mcL.
2. Include concentration and volume on tube.
Specimen Stability Information: Frozen (preferred) 1 year/Ambient/Refrigerated
Additional Information: DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

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: Specimen will only be tested after culture.
1. Specimens are preferred to be received within 24 hours of collection. Culture and 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 are 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 flask **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 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 CULFB / Fibroblast Culture for Biochemical or Molecular Testing. 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**: Chorionic villi **Container/Tube**: 15-mL Tube containing 15 mL of transport media **Specimen Volume**: 20 mg **Specimen Stability Information**: Ambient (preferred) <24 hours/Refrigerated <24 hours **Additional Information**: Specimen will only be tested after culture. 1. Specimens are preferred to be received within 24 hours of collection. Culture and 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 CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks are required to culture fibroblasts 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**: Cultured chorionic villi **Container/Tube**: T-25 flasks **Specimen Volume**: 2 full flasks **Collection Instructions**: Submit confluent cultured cells 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 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 CULFB / Fibroblast Culture for Biochemical or Molecular Testing. 3. **All prenatal specimens must be accompanied by a maternal blood specimen;** order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Current Specimen Retention Time	New Specimen Retention Time
Whole blood: 2 weeks (if available); Extracted DNA: 3 months	Whole blood: 28 days (if available); Extracted DNA: 3 months

Questions

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