
Reporting Title: Uniparental Disomy**Performing Location:** Rochester**Ordering Guidance:**

This test is only intended to rule out whole-chromosome uniparental disomy (UPD). If testing is desired to rule out UPD 11 for Beckwith-Wiedemann syndrome or Russell-Silver syndrome, the recommended test is BWRS / Beckwith-Wiedemann Syndrome/Russell-Silver Syndrome, Molecular Analysis, Varies, as it will also detect cases caused by segmental UPD.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:

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

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)

Specimen Minimum Volume:

Blood: 0.5 mL

Amniotic Fluid: 10 mL

Chorionic Villi: 5 mg

Forms:

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. Molecular Genetics: Uniparental Disomy Patient Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
CULFB	CG770	Reason for Referral	Plain Text	No
CULFB	CG899	Specimen	Plain Text	No
CULAF	CG767	Reason for Referral	Plain Text	No

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
53356	Result Summary	Alphanumeric		50397-9
53357	Result	Alphanumeric		36917-3
53358	Interpretation	Alphanumeric		69047-9
53359	Reason for Referral	Alphanumeric		42349-1
53360	Specimen	Alphanumeric		31208-2
53361	Source	Alphanumeric		31208-2
53362	Method	Alphanumeric		85069-3
53363	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81402

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
CULFB	Fibroblast Culture for Genetic Test			No	Yes
CULAF	Amniotic Fluid Culture/Genetic Test			No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	52327	Result Summary	Alphanumeric		50397-9
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6
CULAF	52304	Result Summary	Alphanumeric		50397-9
CULAF	52306	Interpretation	Alphanumeric		69965-2
CULAF	52305	Result	Alphanumeric		82939-0
CULAF	CG767	Reason for Referral	Alphanumeric		42349-1
CULAF	52307	Specimen	Alphanumeric		31208-2
CULAF	52308	Source	Alphanumeric		31208-2
CULAF	52309	Method	Alphanumeric		85069-3
CULAF	54641	Additional Information	Alphanumeric		48767-8
CULAF	52310	Released By	Alphanumeric		18771-6

Reference Values:

An interpretive report will be provided.