

Overview

Useful For

Evaluation of patients presenting with mosaicism, confined placental mosaicism, or Robertsonian translocations

Evaluation of patients presenting with features of disorders known to be associated with uniparental disomy (eg, Russell-Silver syndrome)

Evaluation of disease mechanism in individuals with rare autosomal recessive disease and only one carrier parent

Reflex Tests

| Test Id | Reporting Name                      | Available Separately | Always Performed |
|---------|-------------------------------------|----------------------|------------------|
| CULFB   | Fibroblast Culture for Genetic Test | Yes                  | No               |
| CULAF   | Amniotic Fluid Culture/Genetic Test | Yes                  | No               |

Genetics Test Information

Specimens from fetus or child and at least one parent are required for testing. Specimens from both parents are recommended for optimal interpretation of the results. Chromosome of interest **must be specified** on request form.

Testing 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.

Special Instructions

- [Informed Consent for Genetic Testing](#)
- [Informed Consent for Genetic Testing \(Spanish\)](#)
- [Molecular Genetics: Uniparental Disomy Patient Information](#)

Method Name

Polymerase Chain Reaction (PCR)/Microsatellite markers

NY State Available

Yes

Specimen

Specimen Type

Varies

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 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

**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)

**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 Minimum Volume**  
Blood: 0.5 mL  
Amniotic Fluid: 10 mL  
Chorionic Villi: 5 mg

**Reject Due To**  
All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

**Specimen Stability Information**

| Specimen Type | Temperature | Time | Special Container |
|---------------|-------------|------|-------------------|
| Varies        | Varies      |      |                   |

**Clinical & Interpretive**

**Clinical Information**  
Uniparental disomy (UPD) occurs when a child inherits 2 copies of a chromosome from only one parent and no copies of that chromosome from the other parent. This is typically due to an error in cell division during the formation of egg or sperm cells (meiosis). When an error causing UPD occurs during meiosis I, both chromosome homologs from a single parent are transmitted, resulting in uniparental heterodisomy. When the error causing UPD occurs during meiosis II or as a postzygotic event, and a single parental homolog is transmitted to offspring in duplicate, isodisomy results. Meiotic

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recombination events within the context of UPD often result in a mixture of regions of heterodisomy and isodisomy.

When UPD occurs, the imbalance of maternal versus paternal genetic information for the involved chromosome can be associated with clinical symptoms in the affected child. However, UPD does not always impart an abnormal clinical phenotype. In fact, while isodisomy can result in disease due to a recessive allele, heterodisomy is not expected to result in an abnormal clinical phenotype unless the involved chromosome or chromosomal segment includes imprinted genes. Imprinted genes demonstrate differential expression depending on parent of origin. Disorders that result from UPD of imprinted genes are not due to a defect in the imprinting mechanism itself, but rather they are due to an unbalanced parental contribution of normally imprinted alleles that results in altered expression of imprinted genes. For example, when maternal UPD 15 occurs (2 copies of the maternal chromosome 15 instead of 1 maternal and 1 paternal copy of chromosome 15), it causes Prader-Willi syndrome due to the lack of paternally expressed genes at the imprinted site.

UPD has been described for many but not all chromosomes. In addition to the rare cases of autosomal recessive disease that result from isodisomy, clinical syndromes associated with UPD have been described for only a few chromosomes, including chromosomes 6, 7, 11, 14, 15 and 20.

UPD cannot be identified by gross cytogenetic analysis and requires molecular DNA-based analysis using multiple polymorphic markers spanning the chromosome of interest.

For optimal interpretation of results, specimens from both parents and the child or fetus are recommended. If only one parent specimen is submitted, testing can be performed; however, biparental inheritance and some types of UPD cannot be definitively established. Additionally, the likelihood for uninformative or inconclusive results is higher.

**Reference Values**

An interpretive report will be provided.

**Interpretation**

Microsatellite markers are compared between the proband and parental samples for the chromosome of interest. The pattern of the microsatellite markers will be classified as demonstrating uniparental disomy or biparental inheritance when sufficient informative markers are identified.

**Cautions**

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in the interpretation of results may occur if the information given is inaccurate or incomplete.

This test will detect nonpaternity.

Uniparental disomy (UPD) may not be detected by this assay in cases where there is low-level mosaicism for a particular chromosome.

This test only rules out whole-chromosome UPD and cannot reliably detect cases of segmental UPD. If testing is desired to rule out UPD 11 for Beckwith-Wiedemann syndrome or Russell-Silver syndrome, BWRS / Beckwith-Wiedemann Syndrome/Russell-Silver Syndrome, Molecular Analysis, Varies is the recommended test as it will also detect cases caused by segmental UPD.

Although UPD testing is available for all chromosomes, prenatal testing for UPD for chromosomes other than those

associated with known phenotypes should be done only after genetic counseling involving adequate discussion of risks, benefits, and limitations of testing.

### Clinical Reference

1. Del Gaudio D, Shinawi M, Astbury C, et al. Diagnostic testing for uniparental disomy: a points to consider statement from the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2020;22(7):1133-1141. doi:10.1038/s41436-020-0782-9
2. Kotzot D, Utermann G. Uniparental disomy (UPD) other than 15: phenotypes and bibliography updated. Am J Med Genet. 2005;136(3):287-305. doi:10.1002/ajmg.a.30483
3. Kotzot D. Prenatal testing for uniparental disomy: indications and clinical relevance. Ultrasound Obstet Gynecol. 2008;31(1):100-105. doi: 10.1002/uog.5133
4. Engel E. A fascination with chromosome rescue in uniparental disomy: Mendelian recessive outlaws and imprinting copyrights infringements. Eur J Hum Genet. 2006;14(11):1158-1169. doi:10.1038/sj.ejhg.5201619

### Performance

#### Method Description

A polymerase chain reaction-based assay, using multiple microsatellite markers (dinucleotide repeats) for the specific chromosome being tested, is used to test DNA from parents and child for the presence of uniparental disomy.(Vnencak-Jones CL. Molecular testing for inherited diseases. Am J Clin Pathol. 1999;112[1 Suppl 1]:S19-S32)

#### PDF Report

No

#### Day(s) Performed

Monday and Wednesday

#### Report Available

5 to 21 days

#### Specimen Retention Time

Whole blood: 2 weeks (if available); Extracted DNA: 3 months

#### Performing Laboratory Location

Rochester

### Fees & Codes

#### Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

81402

LOINC® Information

| Test ID | Test Order Name    | Order LOINC® Value |
|---------|--------------------|--------------------|
| UNIPD   | Uniparental Disomy | 36917-3            |

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