MAYO CLINIC LABORATORIES

Test Definition: UNIPD

Uniparental Disomy, Varies

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

Samples from fetus or child and both parents are required for analysis. Chromosome of interest must be specified on request form.

Testing Algorithm

Polymerase chain reaction and microsatellite markers on the chromosome of interest are used to test DNA from the parents and the child for the presence of uniparental disomy. Uniparental disomy testing is available for all chromosomes, with the exception of chromosome 11 for certain indications. Contact the laboratory for additional information.

For prenatal specimens only: If amniotic fluid (nonconfluent cultured cells) is received, amniotic fluid culture/genetic test will be added and charged separately. If chorionic villus specimen (nonconfluent cultured cells) is received, fibroblast culture for genetic test will be added and charged separately.

See Prader-Willi and Angelman Syndromes: Laboratory Approach to Diagnosis algorithm in Special Instructions.

Special Instructions

- Molecular Genetics: Congenital Inherited Diseases Patient Information
- Informed Consent for Genetic Testing
- Prader-Willi and Angelman Syndromes: Laboratory Approach to Diagnosis
- 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

Shipping Instructions

Specimen preferred to arrive within 96 hours of collection.

Specimen Required

For optimal interpretation of results, 3 specimens are required to perform this test. In addition to child or fetal specimen, a blood specimen from both parents is required. Each specimen must have a separate order for Uniparental Disomy (this test). Only the proband specimen will be charged.

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

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.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Prenatal Specimens
Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing.

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 mg

Specimen Stability Information: Refrigerated

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

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 in Special Instructions:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. Molecular Genetics: Congenital Inherited Diseases Patient Information (T521) in Special Instructions

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 1 parent and no copies of that chromosome from the other parent. This error in division occurs 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, and heterodisomy results. 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 recombination events within the context of UPD often result in a mixture of heterodisomy and isodisomy. UPD can involve an entire chromosome or only a segment. Mosaicism for UPD also occurs in combination with either chromosomally normal or abnormal cell lines.

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 at any location, 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 Russell-Silver syndrome (UPD 7), Prader-Willi syndrome (UPD 15), Angelman syndrome (UPD 15), transient neonatal diabetes (UPD 6), and UPD of chromosome14.

UPD cannot be identified by gross cytogenetic analysis and requires DNA-based analysis using multiple polymorphic markers spanning the chromosome of interest. Specimens from both parents and the child or fetus are required.

Reference Values

An interpretive report will be provided.

Interpretation

An interpretative report will be provided.

Cautions

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

This test will detect nonpaternity.

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

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. Schaffer LG, Agan N, Goldberg JD, Ledbetter DH, Longshore JW, Cassidy DB: American College of Medical Genetics statement on diagnostic testing for uniparental disomy. Genet Med. 2001;3:206-211. doi: 10.1097/00125817-200105000-00011
2. Kotzot D, Utermann G: Uniparental Disomy (UPD) other than 15: phenotypes and bibliography updated. Am J Med Genet. 2005;136A:287-305. doi: 10.1002/ajmg.a.30483
3. Kotzot D: Prenatal testing for uniparental disomy: indications and clinical relevance. Ultrasound Obstet Gynecol. 2008:31:100-105. 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 Nov;14(11):1158-1169. doi: 10.1038/sj.ejhg.5201619

Performance

Method Description
A polymerase chain reaction (PCR)-based assay, using multiple microsatellite markers (dinucleotide repeats) for the particular 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 Regional Manager. 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. This test 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