Test Definition: UNIPD
Uniparental Disomy

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

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

Reflex Tests

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Testing Algorithm
Uniparental disomy testing is available for all chromosomes. PCR and microsatellite markers on the chromosome of interest are used to test DNA from parents and child for the presence of uniparental disomy.

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)

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 draw.

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 (UNIPD / Uniparental Disomy). 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

Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing. Prenatal specimens can be sent Monday through Thursday and must be received by 5 p.m. CST on Friday in order to be processed appropriately.

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 by Mayo Clinic Laboratories for test suitability.

**Specimen Stability Information**

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**Clinical and 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. UPD does not always impart an abnormal clinical phenotype however. 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 chromosome 14.

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


Performance

Method Description
A 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. (Vencak-Jones CL: Molecular testing for inherited diseases. Am J Clin Pathol 1999;112[1 Suppl 1]:S19-S32)

PDF Report
No

Day(s) and Time(s) Test Performed
Monday and Wednesday; 2 p.m.

Analytic Time
5 days

Maximum Laboratory Time
21 days

Specimen Retention Time
Whole Blood: 2 weeks (if available) Extracted DNA: 3 months

Performing Laboratory Location
Rochester

Fees and 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 U.S. Food and Drug Administration.

CPT Code Information
81402

LOINC® Information

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