

Overview

Useful For

Ruling out the presence of maternal cell contamination within a fetal specimen

Required for all prenatal testing performed in Mayo Clinic Laboratories' Molecular and Biochemical Genetics laboratories

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 |
| _STR1 | Comp Analysis using STR (Bill only) | No, (Bill only) | No |
| _STR2 | Add'l comp analysis w/STR (Bill Only) | No, (Bill only) | No |

Genetics Test Information

Required in conjunction with molecular and biochemical prenatal testing only.

Testing Algorithm

Prenatal specimens:

If an amniotic fluid specimen (nonconfluent cultured cells) is received, amniotic fluid culture will be performed at an additional charge.

If a chorionic villi specimen (nonconfluent cultured cells) is received, a fibroblast culture will be performed at an additional charge.

To complete this testing, both maternal and proband/fetal samples must be received, and testing must be ordered under separate order numbers. If proband and maternal samples are received under the same order number, a new test order will be added for the maternal sample, per laboratory protocol.

If this test is ordered in conjunction with CMAP / Chromosomal Microarray, Prenatal, Amniotic Fluid/Chorionic Villus Sampling or CMAPC / Chromosomal Microarray, Autopsy, Products of Conception, or Stillbirth, and no other molecular testing is ordered, the test will be changed to PPAP / Parental Sample Prep for Prenatal Microarray Testing, Blood. If both CMAP / Chromosomal Microarray, Prenatal, Amniotic Fluid/Chorionic Villus Sampling or CMAPC / Chromosomal Microarray, Autopsy, Products of Conception, or Stillbirth, and other molecular testing is ordered, PPAP will be added, per laboratory protocol.

Special Instructions

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

Method Name

Polymerase Chain Reaction (PCR) based comparison of Microsatellite Markers

NY State Available

Yes

Specimen**Specimen Type**

Varies

Ordering Guidance

If a prenatal specimen has already been submitted as part of another diagnostic test, a second prenatal specimen is not required. If a prenatal specimen has not yet been submitted, submit only 1 prenatal specimen.

Specimen Required

Both maternal blood and prenatal specimens are required for testing; order this test on both specimens under separate order numbers.

Maternal Specimen

Specimen Type: Maternal 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.**

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. Testing may be canceled if DNA requirements are inadequate.

Prenatal/Proband 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: Cord blood

Container/Tube: Lavender top (EDTA) or yellow top (ACD)

Specimen Volume: 3 mL

Collection Instructions:

1. Collect whole blood from an umbilical cord either prenatally or postnatally.
2. Invert several times to mix blood.
3. Send whole blood specimen in original tube. **Do not aliquot.**

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. Testing may be canceled if DNA requirements are inadequate.

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.

Specimen Type: Cultured amniocytes

This does not include cultured chorionic villi.

Container/Tube: T-25 flask

Specimen Volume: 2 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.

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.

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.

Forms

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 \(T826\)](#)

Specimen Minimum Volume

See Specimen Required

Reject Due To

All specimens will be evaluated by Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

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

Clinical & Interpretive

Clinical Information

One of the risks associated with prenatal testing is maternal cell contamination (MCC), which can occur when a fetal specimen comes into contact with maternal blood or tissue. The risk of MCC is associated with procedures such as chorionic villus sampling, amniocentesis, or extraction of fetal blood from the umbilical cord (cord blood). If MCC is present, the maternal DNA may mask the results of any genetic testing performed on the fetal DNA. Therefore, the results of prenatal testing may be compromised.

To rule out the presence of MCC, a maternal blood specimen is necessary for comparison of maternal and fetal chromosomal markers. The presence of both maternal and nonmaternal alleles for each fetal marker indicates the fetal specimen is not contaminated. MCC is confirmed when both alleles in the fetus are maternal.

Reference Values

An interpretative report will be provided.

Interpretation

The interpretive report includes an overview of the findings as well as the associated clinical significance.

Cautions

This test does not rule out the presence of low-level maternal cell contamination (<5%).

Clinical Reference

1. Nagan N, Faulkner NE, Curtis C, Schrijver I; MCC Guidelines Working Group of the Association for Molecular Pathology Clinical Practice Committee. Laboratory guidelines for detection, interpretation, and reporting of maternal cell contamination in prenatal analyses a report of the association for molecular pathology. *J Mol Diagn.* 2011;13(1):7-11. doi:10.1016/j.jmoldx.2010.11.013

2. Smeekens SP, Leferink M, Yntema HG, Kamsteeg EJ. Maternal cell contamination in postnatal umbilical cord blood samples implies a low risk for genetic misdiagnoses. *Prenat Diagn.* 2024;44(11):1304-1309. doi:10.1002/pd.6595

Performance

Method Description

The maternal blood genotype is compared to the fetal genotype, derived from amniocyte DNA or chorionic villus DNA, utilizing a polymerase chain reaction-based assay with a set of microsatellite repeat markers.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Varies

Report Available

10 to 11 days

Specimen Retention Time

Whole blood: 28 days (if available); Extracted DNA: 3 months

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

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

- 81265
- 88233-(if appropriate)
- 88240-(if appropriate)
- 88235-(if appropriate)
- 81266-each additional specimen

LOINC® Information

| Test ID | Test Order Name | Order LOINC® Value |
|---------|--------------------------------|--------------------|
| MATCC | Maternal Cell Contamination, B | 40704-9 |

| Result ID | Test Result Name | Result LOINC® Value |
|-----------|---------------------|---------------------|
| 53285 | Result Summary | 50397-9 |
| 53286 | Result | 40704-9 |
| 53287 | Interpretation | 69047-9 |
| 53288 | Reason for referral | 42349-1 |
| 53289 | Specimen | 31208-2 |
| 53290 | Source | 31208-2 |
| 53291 | Released By | 18771-6 |
| 55150 | Method | 85069-3 |