Overview

Useful For
Prenatal diagnosis of chromosome abnormalities, including aneuploidy (ie, trisomy or monosomy) and balanced rearrangements

This test is not appropriate as a first-tier test for detecting gains or losses of chromosomal material in pregnancies with 1 or more major structural abnormalities.

Genetics Test Information
Cultures from this specimen will be discarded 10 days after all cytogenetic test results have been reported. If further testing is desired, call the laboratory at 507-284-1668.

Reflex Tests

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<th>Available Separately</th>
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Testing Algorithm
This test is not appropriate as a first-tier test for detecting gains or losses of chromosomal material in pregnancies with 1 or more major structural abnormalities.

This test includes a charge for cell culture of fresh specimens and professional interpretation of results. Analysis charges will be incurred for total work performed, and generally include 2 banded karyograms and the analysis of 20 metaphase cells. If no metaphase cells are available for analysis, no analysis charges will be incurred. If additional analysis work is required, additional charges may be incurred.

The following algorithms are available in Special Instructions:
- Prenatal Aneuploidy Screening and Diagnostic Testing Options
- High-Risk Pregnancy Based on Fetal Malformations or Positive Serum Screen: Laboratory Testing Algorithm

Special Instructions
- Informed Consent for Genetic Testing
- High-Risk Pregnancy Based on Fetal Malformations or Positive Serum Screen: Laboratory Testing Algorithm
- Prenatal Aneuploidy Screening and Diagnostic Testing Options
- Informed Consent for Genetic Testing (Spanish)

Method Name
Cell Culture Followed by Chromosome Analysis
NY State Available
Yes

Specimen

Specimen Type
Tissue

Advisory Information
This test should be performed for prenatal diagnostic purposes only. A chromosomal microarray (CMAP / Chromosomal Microarray, Prenatal, Amniotic Fluid/Chorionic Villus Sampling) is recommended, rather than chromosomal analysis, to detect clinically relevant gains or losses of chromosomal material in pregnancies with 1 or more major structural abnormalities. Chromosomal microarray can also be considered, rather than chromosome analysis, for patients undergoing invasive prenatal diagnostic testing with a structurally normal fetus.

Shipping Instructions
Advise Express Mail or equivalent if not on courier service.

Necessary Information
Provide a reason for referral with each specimen. The laboratory will not reject testing if this information is not provided, but appropriate testing and interpretation may be compromised or delayed.

Specimen Required
Supplies: CVS Media (RPMI) and Small Dish (T095)

Source: Chorionic villi

Container/Tube: 15-mL tube containing 15 mL of transport media

Specimen Volume: 20-30 mg

Collection Instructions:
1. Collect chorionic villus specimen (CVS) by the transabdominal or transcervical method.
2. Transfer the CVS to a Petri dish containing transport medium (Such as CVS media (RPMI) and Small Dish [T095]).
3. Using a stereomicroscope and sterile forceps, assess the quality and quantity of the villi and remove any blood clots and maternal decidua.

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 in Special Instructions:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

Specimen Minimum Volume
Test Definition: CHRCV
Chromosomes, Chorionic Villus Samp

12 mg
If ordering in conjunction with other testing:
PADF: 14 mg
CMAP: 24 mg
PADF and CMAP: 26 mg

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

Specimen Stability Information

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Clinical and Interpretive

Clinical Information
Although not used as widely as amniocentesis, the use of chorionic villus sampling (CVS) for chromosome analysis is an important procedure for the prenatal diagnosis of chromosome abnormalities. CVS can be collected by either transcervical or transabdominal techniques. The medical indications for performing chromosome studies on CVS are similar to amniocentesis, and may include advanced maternal age, abnormal first-trimester screen, and family history of a chromosome abnormality.

A chromosomal microarray (CMAP / Chromosomal Microarray, Prenatal) is recommended, rather than chromosomal analysis, to detect clinically relevant gains or losses of chromosomal material in pregnancies with one or more major structural abnormalities. Chromosomal microarray can also be considered, rather than chromosome analysis, for patients undergoing invasive prenatal diagnostic testing with a structurally normal fetus.

Reference Values
An interpretive report will be provided.

Interpretation
Cytogenetic studies on chorionic villus specimen (CVS) are considered more than 99% reliable for the detection of most fetal chromosome abnormalities. However, subtle or cryptic abnormalities involving microdeletions usually can be detected only with the use of targeted FISH testing.

Approximately 3% of CSVs analyzed are found to have chromosome abnormalities. Some of these chromosome abnormalities are balanced and may not be associated with birth defects. A normal karyotype does not rule out the possibility of birth defects, such as those caused by submicroscopic cytogenetic abnormalities, molecular mutations, and environmental factors (ie, teratogen exposure). For these reasons, clinicians should inform their patients of the technical limitations of chromosome analysis before the procedure is performed, so that patients may make an informed decision about pursuing the procedure.

Limitations:
-False-chromosome mosaicism may occur due to artifact of culture
Test Definition: CHRCV
Chromosomes, Chorionic Villus Samp

- True mosaicism may be missed due to statistical sampling error
- Presence of chromosome abnormalities in placental cells that do not occur in the cells of the fetus (confined placental mosaicism)
- Subtle structural chromosome abnormalities can occasionally be missed

It is recommended that a qualified professional in Medical Genetics communicate all results to the patient.

Cautions
Interfering factors:
- Inadequate amount of specimen may not permit adequate analysis.
- Exposure of the specimen to temperature extremes (freezing or >30 degrees C) may kill cells and severely interfere with attempts to culture cells.
- Improper packaging may result in broken, leaky, and contaminated specimens during transport.
- Transport time should not exceed 2 days.
- Contamination by maternal cells may interfere with attempts to culture cells and may cause interpretive problems.

Clinical Reference

Performance
Method Description
The chorionic villi are thoroughly cleaned using sterile forceps to remove remaining maternal decidua and blood clots. The villi are then treated with trypsin and collagenase. The cells are then grown on culture media. In the harvest procedure, the cells are exposed to colcemid and hypotonic solution, and then fixed with glacial acetic acid and methanol. Metaphase cells are dropped onto microscope slide and are routinely stained by G-banding, but other staining methods are frequently employed as needed. Twenty metaphases from 3 or more primary cultures are analyzed. Five or more metaphases are stored in a computer-based imaging system and karyograms are made from 2 or more representative metaphases. (Breed AS, Mantingh A, Beekhuis JR, et al: The predictive value of cytogenetic diagnosis after CVS: 1,500 cases. Prenat Diagn 1990;10:101-110; Canadian Collaborative CVS-Amniocentesis Clinical Trial Group: Multicentre randomized clinical trial of chorionic villus sampling and amniocentesis. Lancet 1989;1:1-6; Ledbetter DH, Martin AO, Verlinsky Y, et al: Cytogenetic results of chorionic villus sampling: high success rate and diagnostic accuracy in the United States collaborative study. Am J Obstet Gynecol
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Chromosomes, Chorionic Villus Samp


PDF Report
No

Day(s) and Time(s) Test Performed
Specimens are processed Monday through Sunday.

Results reported Monday through Friday; 8 a.m.-5 p.m.

Analytic Time
9 days

Maximum Laboratory Time
10 days

Specimen Retention Time
Any remaining specimen is discarded at the time results are reported.

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
88235, 88291-Tissue culture for amniotic fluid or chorionic villus cells, Interpretation and report

88267 w/modifier 52-Chromosome analysis, amniotic fluid or chorionic villus, <15 cells, 1 karyotype with banding (if appropriate)

88267-Chromosome analysis, amniotic fluid or chorionic villus, 15 cells, 1 karyotype with banding (if appropriate)

88267, 88285-Chromosome analysis, amniotic fluid or chorionic villus, <15 cells, 1 karyotype with banding (if appropriate)

LOINC® Information

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