Test Definition: CHRCV

Chromosome Analysis, Chorionic Villus Sampling

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.

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
_ML15	Metaphases, <15	No, (Bill Only)	No
_M15	Metaphases, 15	No, (Bill Only)	No
_MG14	Metaphases, >15	No, (Bill Only)	No
_KTG1	Karyotypes, >1	No, (Bill Only)	No
_STAC	Ag-Nor/CBL Stain	No, (Bill Only)	No

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 800-533-1710.

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.

Special Instructions

- Informed Consent for Genetic Testing
- Informed Consent for Genetic Testing (Spanish)

Method Name

Cell Culture Followed by Chromosome Analysis

NY State Available

Yes

Specimen

Specimen Type
Tissue

Ordering Guidance
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.

Portions of the specimen may be used for other tests such as molecular genetic testing, biochemical testing, and fluorescence in situ hybridization (FISH) testing (including PADF / Prenatal Aneuploidy Detection, FISH). If additional molecular genetic or biochemical genetic testing is needed, order CULFB / Fibroblast Culture for Genetic Test so that cell cultures may be set up specifically for the use in these tests.

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
Specimen Type: Chorionic villi
Supplies: CVS Media (RPMI) and Small Dish (T095)
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.
4. If ordering with PADF / Prenatal Aneuploidy Detection, FISH, submit a minimum of 14 mg.
5. If ordering with CMAP / Chromosomal Microarray, Prenatal, Amniotic Fluid/Chorionic Villus Sampling, submit a minimum of 24 mg.
6. If ordering with both PADF and CMAP, then submit a minimum of 26 mg.

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
The following is the minimum volume when only this test is ordered:
12 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
Tissue	Refrigerated (preferred)		
	Ambient		

Clinical & 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 those for amniocentesis, and may include advanced maternal age, abnormal prenatal screen, and family history of a chromosome abnormality.

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 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 specimens (CVS) are considered nearly 100% accurate for the detection of non-mosaic whole chromosome abnormalities. Most sub-chromosomal abnormalities greater than 5-20Mb are also easily detectable dependent on size, location, and quality of the metaphase chromosomes. Very subtle or cryptic abnormalities involving microdeletions/duplication can typically only be detected by chromosomal microarray or targeted fluorescence in situ hybridization testing.

Approximately 3% of CVS samples 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, pathogenic molecular variants, 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:
-Abnormal results from CVS analysis may represent confined placental mosaicism and may not reflect the fetal karyotype.
-Only large abnormalities visible by manual inspection are detectable; subtle structural chromosome abnormalities may be missed
-Artifacts of cell culture may very rarely be misinterpreted as mosaicism in the sample.

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

1. American College of Obstetricians and Gynecologists Committee on Genetics. Committee Opinion No. 581: the use of chromosomal microarray analysis in prenatal diagnosis. Obstet Gynecol. 2013;122:1374-1377
2. Society for Maternal-Fetal Medicine (SMFM). The use of chromosomal microarray for prenatal diagnosis. Am J Obstet Gynecol. 2016;215:B2-B9
3. Committee Opinion, 640. Cell-free DNA screening for fetal aneuploidy. American College of Obstetricians and Gynecologists Committee on Genetics. Obstet Gynecol. 2015;123:e31-e37
4. Wilson KL, Czerwinski JL, Hoskovec JM, et al. NSGC practice guideline: prenatal screening and diagnostic testing options for chromosome aneuploidy. J Genet Couns. 2013;22:4-15

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, harvested, and analyzed using the In Situ Culture and Analysis (ISCA) method. In the harvest procedure, the cells are exposed to colcemid and hypotonic solution, and then fixed with glacial acetic acid and methanol. Metaphase preparations are routinely stained by G-banding, but other staining methods are frequently employed as needed. Twenty metaphases from 3 or more primary cultures are examined. In cases of pseudomosaicism or when true mosaicism is suspected, up to 6 different 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.(Arsham, Marilyn S., et al. editors. The AGT Cytogenetics Laboratory Manual. 4th ed. Wiley-Blackwell; 2017; Spurbeck JL, Carlson RO, Allen JE, Dewald GW. Culturing and robotic harvesting of bone marrow, lymph nodes, peripheral blood, fibroblasts, and solid tumors with in situ techniques. Cancer Genet

Cytogenet. 1988;32:59-66)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
9 to 14 days

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

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
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

Test ID	Test Order Name	Order LOINC® Value
CHRCV	Chromosomes, Chorionic Villus Samp	62350-4

Result ID	Test Result Name	Result LOINC® Value
52319	Result Summary	50397-9

52321	Interpretation	69965-2
52320	Result	82939-0
CG769	Reason for Referral	42349-1
52322	Specimen	31208-2
52323	Source	31208-2
52325	Method	85069-3
52324	Banding Method	62359-5
54624	Additional Information	48767-8
52326	Released By	18771-6