MAYO CLINIC LABORATORIES

Test Definition: CHRCB

Chromosome Analysis, Congenital Disorders, Blood

Overview

Useful For

Diagnosis of congenital chromosome abnormalities, including aneuploidy, structural abnormalities, and balanced rearrangements

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
_M15A	Metaphases, 1-14	No, (Bill Only)	No
_M19	Metaphases, 15-20	No, (Bill Only)	No
_MG19	Metaphases, >20	No, (Bill Only)	No
_KTG2	Karyotypes, >2	No, (Bill Only)	No
_STAC	Ag-Nor/CBL Stain	No, (Bill Only)	No

Testing Algorithm

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.

This test is not appropriate for detecting acquired chromosome abnormalities. If this test is ordered with a reason for testing indicating a hematologic disorder, the test will be cancelled and CHRHB / Chromosome Analysis, Hematologic Disorders, Blood will be performed as the appropriate test.

A chromosomal microarray study (CMACB / Chromosomal Microarray, Congenital, Blood) is recommended as the first-tier test (rather than a congenital chromosome study) to detect clinically relevant gains or losses of chromosomal material for individuals with multiple anomalies not specific to well-delineated genetic syndromes, individuals with apparently nonsyndromic developmental delay or intellectual disability, and individuals with autism spectrum disorders.

Special Instructions

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

Method Name

Cell Culture with Mitogens followed by Chromosome Analysis

NY State Available

Yes

Specimen

Specimen Type
Whole blood

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

Necessary Information
Provide a reason for testing 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
Submit only 1 of the following specimens:

Specimen Type: Whole blood
Container/Tube: Green top (sodium heparin)
Specimen Volume: 4 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Other anticoagulants are not recommended and are harmful to the viability of the cells.
3. Label specimen as whole blood.

Specimen Type: Cord blood
Container/Tube: Green top (sodium heparin)
Specimen Volume: As much as possible
Collection Instructions:
1. Invert several times to mix blood.
2. Other anticoagulants are not recommended and are harmful to the viability of the cells.
3. Label specimen as cord blood.

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
2 mL

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

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Ambient (preferred)		
	Refrigerated		

Clinical & Interpretive

Clinical Information

Chromosome analysis is appropriate for individuals with clinical features including infertility, multiple miscarriages, delayed puberty, ambiguous genitalia, amenorrhea, or individuals with clinical features suggestive of an aneuploidy syndrome, including Down syndrome, Turner syndrome, Klinefelter syndrome, Trisomy 13 syndrome, and Trisomy 18 syndrome.

A chromosomal microarray study (CMACB / Chromosomal Microarray, Congenital, Blood) is recommended as the first-tier test (rather than a congenital chromosome study) to detect clinically relevant gains or losses of chromosomal material for individuals with multiple anomalies not specific to well-delineated genetic syndromes, individuals with apparently nonsyndromic developmental delay or intellectual disability, and individuals with autism spectrum disorders. Chromosome analysis may be appropriate for this patient population if microarray has been performed with normal results. Some chromosome rearrangements are balanced (no gain or loss of material) and, therefore, not detectable by chromosomal microarray. In rare situations these rearrangements may interrupt gene functioning and have the potential to cause abnormal clinical features.

Limitations: A normal karyotype (46,XX or 46,XY with no apparent chromosome abnormality) does not eliminate the possibility of abnormal clinical features such as those caused by submicroscopic cytogenetic abnormalities, molecular mutations, and environmental factors (ie, teratogen exposure). Chromosomal mosaicism may be missed due to statistical sampling error (rare) and subtle structural chromosome abnormalities can occasionally be missed.

Reference Values

An interpretive report will be provided.

Interpretation

When interpreting results, the following factors need to be considered:
-Some chromosome abnormalities are balanced (no apparent gain or loss of genetic material) and may not be associated with birth defects. However, balanced abnormalities often cause infertility and, when inherited in an unbalanced fashion, may result in birth defects in the offspring.
-A normal karyotype (46,XX or 46,XY with no apparent chromosome abnormality) does not eliminate the possibility of birth defects such as those caused by submicroscopic cytogenetic abnormalities, molecular mutations, and environmental factors (ie, teratogen exposure).

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

Cautions

This test is not appropriate for acquired hematologic disorders, including the following malignancies: chronic myelocytic leukemia, acute myelocytic leukemia, acute lymphocytic leukemia, chronic lymphocytic leukemia, lymphoma, and leukemia.

This test is not appropriate as a first-tier test for detecting gains or losses of chromosomal material for individuals with intellectual disability, developmental delay, autism, dysmorphic features, birth defects, behavior disorders, learning disability, or cognitive impairment.

Interfering factors:
-Cell lysis caused by forcing the blood quickly through the needle
-Use of an improper anticoagulant or improperly mixing the blood with the anticoagulant
-Excessive transport time
-Inadequate amount of blood may not permit adequate analysis
-Improper packaging may result in broken, leaky, and contaminated specimen during transport
-Exposure of the specimen to temperature extremes (freezing or >30 degrees C) may kill cells and interfere with attempts to culture cells

Clinical Reference

1. McKinlay Gardner RJ, Amor DJ, eds. Amor, Gardner and Sutherland's Chromosome Abnormalities and Genetic Counseling, 5th ed. Oxford Monographs on Medical Genetics, 2018
2. Manning M, Hudgins L. Professional Practice and Guidelines Committee: Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities. Genet Med. 2010;12(11):742-745
3. Sheets KB, Crissman BG, Feist CD, et al. Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: recommendations of the national society of genetic counselors. J Genet Couns. 2011;20(5):432-441
4. Practice Committee of the American Society for Reproductive Medicine. Evaluation and treatment of recurrent pregnancy loss: a committee opinion. Fertil Steril. 2012;98(5):1103-1111
5. Committee opinion no. 605: primary ovarian insufficiency in adolescents and young women. Obstet Gynecol. 2014;124(1):193-197

Performance

Method Description

The cytogenetic procedure to study cells from peripheral blood is designed to reduce the problems from the common interfering factors. A portion of the whole blood is transferred to a flask containing media and a cell mitogen. The cells are incubated for 66 to 72 hours at 37 degrees C. In the harvesting process, the cells are exposed to colcemid, ethidium bromide, and hypotonic solution and are fixed with glacial acetic acid and methanol. Metaphase cells are dropped onto microscope slides and routinely stained by G-banding. Other staining methods are employed as needed. Twenty metaphases are usually examined. In cases with suspected mosaicism, 30 or more metaphases are analyzed. In cases in which testing is ordered for confirmation of a known familial chromosome abnormality, an abbreviated study consisting of the analysis of 5 total metaphases may be performed. Minimal evidence for the presence of an abnormality is defined as 2 or more metaphases with the same structural abnormality or chromosome gain (trisomy), or 3 or more metaphases

lacking the same chromosome. Five or more digitized images of metaphases are stored in a computer-based imaging system and karyograms are made from 2 or more representative metaphases.(Arsham MS, Barch MJ, Lawce HJ, eds. The AGT Cytogenetics Laboratory Manual. 4th ed. John Wiley and Sons; 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[1]:59-66)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
10 days

Specimen Retention Time
4 weeks

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 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
88230, 88291- Tissue culture for Lymphocytes, Interpretation and report
88262 w/modifier 52-Chromosome analysis less than15 cells (if appropriate)
88262-Chromosome analysis with 15 to 20 cells (if appropriate)
88262, 88285-Chromosome analysis with greater than 20 cells (if appropriate)
88280-Chromosome analysis, greater than 2 karyotypes (if appropriate)
88283-Additional specialized banding technique (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value

CHRCB	Chromosomes, Congenital, Blood	81862-5

Result ID	Test Result Name	Result LOINC® Value
52366	Result Summary	50397-9
52368	Interpretation	69965-2
52367	Result	82939-0
CG775	Reason for Referral	42349-1
52369	Specimen	31208-2
52370	Source	31208-2
52372	Method	85069-3
52371	Banding Method	62359-5
54630	Additional Information	48767-8
52373	Released By	18771-6