Overview

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

Reflex Tests

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
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<tr>
<td>_M15A</td>
<td>Metaphases, 1-14</td>
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<td>_STAC</td>
<td>Ag-Nor/CBL Stain</td>
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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 referral 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
Specimen Required
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.

Submit only 1 of the following specimens:

**Specimen Type:** Whole blood

**Container/Tube:** Green top (sodium heparin)

**Specimen Volume:** 5 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 whole 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 in Special Instructions:

- [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**
Clinical and 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, mental retardation, 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


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 harvest 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.(Dewald GW, Michels VV: Recurrent miscarriages: cytogenetic causes and genetic counseling of affected families. Clin Obstet Gynecol 1986;29:865-885; 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
Test Definition: CHRCB
Chromosomes, Congenital, Blood

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
10 days

Maximum Laboratory Time
10 days

Specimen Retention Time
Four weeks

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
88230, 88291- Tissue culture for Lymphocytes, Interpretation and report
88262 w/modifier 52-Chromosome analysis less than 15 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

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