Overview

Useful For
Establishing a diagnosis of Bloom syndrome

Genetics Test Information
Diagnostic testing for Bloom syndrome

Reflex Tests

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
</tr>
</thead>
<tbody>
<tr>
<td>_M20A</td>
<td>Metaphases, 1-19</td>
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<td>_M25A</td>
<td>Metaphases, 20-25</td>
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<tr>
<td>_MG26</td>
<td>Metaphases, &gt;25</td>
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</tbody>
</table>

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.

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

Method Name
Cell Culture with Exposure to BrdU Prior to Harvest followed by Statistical Analysis of Metaphases for Spontaneous Sister Chromatid Exchange.

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.

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.

**Additional Information:**

1. Advise Express Mail or equivalent if not on courier service.

2. If submitting with other cytogenetic studies, a minimum of 1 mL is required in addition to other volumes. Specimens will not be rejected if the volume is less than the minimum, but testing may not be possible depending on the amount received and other tests ordered.

**Forms**

1. **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)

2. If not ordering electronically, complete, print, and send a [Hematopathology/Cytogenetics Test Request](#) (T726) with the specimen.

**Specimen Minimum Volume**

1 mL/If submitting with other cytogenetics studies, a minimum of 1 mL is required in addition to other volumes. Specimens will not be rejected if the volume is less than minimum, but testing may not be possible depending on the amount received and other tests ordered.

**Reject Due To**

No specimen should be rejected.

**Specimen Stability Information**

<table>
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<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
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<tbody>
<tr>
<td>Whole blood</td>
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<tr>
<td></td>
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**Clinical and Interpretive**

**Clinical Information**

Sister chromatid exchange analysis is appropriate in individuals with clinical features suggestive of Bloom syndrome.

Bloom syndrome is a genetic disorder associated with various congenital defects and predisposition to acute leukemia, pulmonary fibrosis, and Hodgkin lymphomas. Carcinoma also is commonly seen in these patients. Approximately one-fourth to one-half of patients develop some type of cancer with a mean age of 25 years at onset. The severity and age of onset of cancer varies among patients.
These patients often have prenatal or postnatal growth retardation, short stature, malar hypoplasia, telangiectatic erythema of the face and other regions, hypo- and hyperpigmentation, immune deficiencies, occasional mild mental retardation, infertility, and high-pitched voices.

Bloom syndrome is an autosomal recessive disorder caused by mutations in the *BLM* gene located at 15q26.1. While multiple mutations have been detected, the use of molecular testing to diagnose Bloom syndrome is limited in many ethnic groups. Patients with Bloom syndrome demonstrate a high frequency of chromosome abnormalities when their cells are cultured. Thus, cytogenetic studies can be helpful to establish a diagnosis.

Bloom syndrome results in 2 characteristic cytogenetic abnormalities. First, the cells are at increased risk for random breaks leading to fragments or exchanges between nonhomologous chromosomes. Second, cells in these patients have an increased frequency of sister chromatid exchanges (SCE: exchange of material between homologous chromosomes) of approximately 10-fold to 20-fold higher than average.

This test is diagnostic for Bloom syndrome. This test cannot be used to identify heterozygote carriers for Bloom syndrome and is not appropriate as part of a prenatal screening panel.

A normal result does not rule out the possibility of birth defects, such as those caused by chromosomal abnormalities, molecular mutations, and environmental factors (ie, teratogen exposure). The test does not rule out other numeric or structural abnormalities. If a constitutional chromosome abnormality is suspected, a separate conventional cytogenetic study, CHRCB / Chromosome Analysis, for Congenital Disorders, Blood should be requested.

**Reference Values**

An interpretive report will be provided.

**Interpretation**

A frequency of sister chromatid exchange comparable to a control specimen and historical reference values will be reported as normal.

A 10-fold or more increase in sister chromatid exchange relative to a control specimen and historical reference values will be reported as abnormal. This is consistent with a diagnosis of Bloom syndrome.

**Cautions**

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
- T lymphocytes that do not respond to mitogens used to stimulate T cells to undergo mitosis (rare)
- 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

A portion of the whole blood is transferred to a flask containing media and a cell mitogen. The cells are incubated for 96 hours at 37 degrees C. Twenty-four hours before harvest the cells are exposed to bromodeoxyuridine (BrdU) and protected from light. In the harvest process, the cells are exposed to colcemid and a hypotonic solution, and fixed with glacial acetic acid and methanol. Metaphase cells are dropped onto microscope slide and are treated with acridine orange. Twenty metaphases are examined for sister chromatid exchange (SCE) using a fluorescence microscope system and documented by computer-based imaging. Mean frequency of SCE per metaphase is calculated and compared with SCE per metaphase from a parallel control.(Dicken CH, Dewald G, Gordon H: Sister chromatid exchanges in Bloom's syndrome. Arch Dermatol 1978;114:755-760)

PDF Report

No

Day(s) and Time(s) Test Performed
Samples processed Monday through Sunday. Results reported Monday through Friday, 8 a.m. to 5 p.m. CST.

Analytic Time
13 days

Maximum Laboratory Time
14 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
Test Definition: BLOOM
Chromosomes, Bloom Syndrome

88245 w/modifier 52 â€“ Chromosome analysis for breakage syndromes, less than 20 cells (if appropriate)

88245 - Chromosome analysis for breakage syndromes, 20-25 cells (if appropriate)

88245, 88285 - Chromosome analysis for breakage syndromes, more than 25 cells (if appropriate)

**LOINC® Information**

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