Overview

Useful For
Screening for chromosomal aneuploidies of chromosomes 13, 15, 16, 18, 21, 22, X, and Y when fresh tissue is not available

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>PBCT</td>
<td>Probe, +2</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>PADD</td>
<td>Probe, +1</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>PB02</td>
<td>Probe, +2</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>PB03</td>
<td>Probe, +3</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>IL25</td>
<td>Interphases,</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>IO99</td>
<td>Interphases, 25-99</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
<tr>
<td>I300</td>
<td>Interphases, &gt;=100</td>
<td>No, (Bill Only)</td>
<td>No</td>
</tr>
</tbody>
</table>

Testing Algorithm
This test does not include a pathology consult. If a pathology consultation is requested, PATHC / Pathology Consultation should be ordered and the appropriate FISH test will be ordered and performed at an additional charge.

This test includes a charge for application of the first probe set (2 FISH probes) and professional interpretation of results.

Additional charges will be incurred for all reflex probes performed. Analysis charges will be incurred based on the number of cells analyzed per probe set. If no cells are available for analysis, no analysis charges will be incurred.

Special Instructions
- Final Disposition of Fetal/Stillborn Remains
- Informed Consent for Genetic Testing
- Informed Consent for Genetic Testing (Spanish)

Method Name
Fluorescence In Situ Hybridization (FISH)

NY State Available
Yes

Specimen

Specimen Type
Tissue

Advisory Information
If a fresh specimen or specimen in fixative is submitted, ANPAT / Anatomic Pathology Consultation, Wet Tissue will
be added by the laboratory, at an additional charge, to facilitate the performance of this test.

**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**

Submit only 1 of the following specimens:

**Specimen Type:** Tissue

**Preferred:** Tissue block

**Collection Instructions:** Formalin-fixed, paraffin-embedded tissue block containing fetal or placental (including chorionic vili) tissue.

**Additional Information:** A pathology report must be submitted 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.

**Acceptable:** Slides

**Collection Instructions:** 6 consecutive, unstained, 5-micron-thick sections placed on positively charged slides, and 1 hematoxylin and eosin-stained slide.

**Additional Information: Do not** send the entire fetus.

**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 an [Oncology Test Request](T729) (T729) with the specimen.

3. [Final Disposition of Fetal/Stillborn Remains](T729) in Special Instructions.

**Specimen Minimum Volume**

Formalin-fixed, paraffin-embedded tissue block

Four consecutive, unstained, slides and 1 hematoxylin and eosin-stained slide.

**Reject Due To**

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

**Specimen Stability Information**

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tissue</td>
<td>Ambient (preferred)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Refrigerated</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Clinical and Interpretive

Clinical Information

Products of conception (POC) are tissues created at conception that spontaneously miscarry; these tissues include chorionic villi, fetal membranes, or fetal tissue. Spontaneous miscarriages occur in 15% to 20% of all recognized human conceptions. While there are many possible causes for miscarriages, chromosome anomalies can be identified in up to 50% of first-trimester miscarriages. It is important to determine a possible chromosomal cause of the pregnancy loss as this information impacts patient management and facilitates understanding of the reason for the loss.

Chromosomal aneuploidy, the gain or loss of chromosomes, is a major cause of early fetal demise. Trisomy is the most common type of chromosome abnormality in spontaneous abortions and has been observed for most chromosomes, with 13, 15, 16, 18, 21, 22, X, and Y being the most common.

Chromosomal microarray analysis of POC (CMAMT / Chromosomal Microarray, POC, FFPE) is available when a more comprehensive assessment for chromosome abnormalities is desired.

Reference Values

An interpretive report will be provided.

Interpretation

Aneuploidy is detected when the percent of cells with an abnormality exceeds the normal reference range for any given probe.

Cautions

This test has not been approved by the U.S. Food and Drug Administration and is best used as an adjunct to existing clinical and pathological information.

Fixatives other than formalin (eg Prefer, Bouin) may not be successful for FISH assays, however nonformalin-fixed samples will not be rejected.

Paraffin-embedded tissues that have been decalcified are generally unsuccessful for FISH analysis. The pathologist reviewing the hematoxylin and eosin-stained slide may find it necessary to cancel testing.

Supportive Data

The specificity of these probes is estimated to be 100% with no cross hybridizations. To establish the analytic sensitivity, analysis was based on 37 products of conception specimens. Chromosome analysis was unsuccessful for 8 of the 37 specimens. For the remaining 29 cases, the correct FISH result was obtained when compared to chromosome analysis.

Clinical Reference


Performance

Method Description

This test is performed using probes for the centromere region of chromosomes X (DXZ1), Y (DYZ3), 18 (D18Z1), 16 (D16Z3), and 15 (D15Z4) and locus-specific probes for 13q14, 21q22, and 22q11.2. Formalin-fixed, paraffin-
embedded tissues are cut at 5 microns and mounted on positively charged glass slides. The selection of tissue and the identification of fetal tissue on the hematoxylin and eosin (H and E)-stained slide is performed by a pathologist. Using the H and E-stained slide as a reference, target areas are etched with a diamond tipped etcher on the back of the unstained slide to be assayed. For each probe set, 2 technologists each analyze 50 interphase nuclei (100 total). Aneuploidy of chromosomes 13, 15, 16, 18, 21, 22, X, and/or Y is reported. (Unpublished Mayo method)

**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**

5 days

**Maximum Laboratory Time**

8 days

**Specimen Retention Time**

Slides and H&E used for analysis are retained by the laboratory in accordance to CAP and NYS requirements. Client provided paraffin blocks and extra unstained slides (if provided) will be returned after testing is complete.

**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 using an analyte specific reagent. Its performance characteristics were 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**

88271 x 2, 88291-DNA probe, each (first probe set), Interpretation and report

88271 x 2-DNA probe, each; each additional probe set (if appropriate)

88271-DNA probe, each; coverage for sets containing 3 probes (if appropriate)

88271 x 2-DNA probe, each; coverage for sets containing 4 probes (if appropriate)

88271 x 3-DNA probe, each; coverage for sets containing 5 probes (if appropriate)

88274 w/modifier 52-Interphase in situ hybridization, <25 cells, each probe set (if appropriate)
88274-Interphase in situ hybridization, 25 to 99 cells, each probe set (if appropriate)

**LOINC® Information**

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Test Order Name</th>
<th>Order LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>POCF</td>
<td>POC Aneuploidy Detection, FISH, Ts</td>
<td>In Process</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Result ID</th>
<th>Test Result Name</th>
<th>Result LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>52063</td>
<td>Result Summary</td>
<td>50397-9</td>
</tr>
<tr>
<td>52065</td>
<td>Interpretation</td>
<td>69965-2</td>
</tr>
<tr>
<td>54573</td>
<td>Result</td>
<td>62356-1</td>
</tr>
<tr>
<td>CG729</td>
<td>Reason For Referral</td>
<td>42349-1</td>
</tr>
<tr>
<td>CG730</td>
<td>Specimen</td>
<td>31208-2</td>
</tr>
<tr>
<td>52066</td>
<td>Source</td>
<td>39111-0</td>
</tr>
<tr>
<td>52067</td>
<td>Tissue ID</td>
<td>80398-1</td>
</tr>
<tr>
<td>52068</td>
<td>Method</td>
<td>49549-9</td>
</tr>
<tr>
<td>55120</td>
<td>Additional Information</td>
<td>48767-8</td>
</tr>
<tr>
<td>53842</td>
<td>Disclaimer</td>
<td>62364-5</td>
</tr>
<tr>
<td>52069</td>
<td>Released By</td>
<td>18771-6</td>
</tr>
</tbody>
</table>