## Overview

### Useful For

Screening for chromosomal aneuploidies of chromosomes 13, 18, 21, X, and Y in prenatal specimens

### Reflex Tests

| Test ID | Reporting Name | Available Separately | Always Performed |
|---|---|---|---|
| _PBCT | Probe, +2 | No, (Bill Only) | No |
| _PADD | Probe, +1 | No, (Bill Only) | No |
| _PB02 | Probe, +2 | No, (Bill Only) | No |
| _PB03 | Probe, +3 | No, (Bill Only) | No |
| _IL25 | Interphases, | No, (Bill Only) | No |
| _I099 | Interphases, 25-99 | No, (Bill Only) | No |
| _I300 | Interphases, >=100 | No, (Bill Only) | No |

### Testing Algorithm

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.

The following algorithms are available in Special Instructions:

-High-Risk Pregnancy Based on Abnormal Fetal Malformations: Laboratory Testing Algorithm

-Prenatal Aneuploidy Screening and Diagnostic Testing Options

### Special Instructions

- Informed Consent for Genetic Testing
- High-Risk Pregnancy Based on Abnormal Fetal Malformations: Laboratory Testing Algorithm
- Prenatal Aneuploidy Screening and Diagnostic Testing Options
- Informed Consent for Genetic Testing (Spanish)

### Method Name

Fluorescence In Situ Hybridization (FISH)

### NY State Available

Yes

## Specimen

### Specimen Type

Varies

## Advisory Information

This test does not detect aneuploidy of chromosomes other than 13, 18, 21, X, or Y. This test does not detect other chromosomal or structural anomalies.

Low levels of mosaicism involving chromosomes 13, 18, 21, X, or Y may not be detected by this procedure.

## Necessary Information

Provide a reason for referral and gestational age with each specimen, and verify the specimen source. 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:**

**Preferred:**

**Supplies:** Refrigerate/Ambient Mailer, 5 lb (T329)

**Specimen Type:** Amniotic fluid

**Container/Tube:** Amniotic fluid container

**Specimen Volume:** 20-25 mL

**Collection Instructions:**

1. Optimal timing for specimen collection is during 14 to 18 weeks of gestation, but specimens collected at other weeks of gestation are also accepted. Provide gestational age at the time of amniocentesis.

2. Discard the first 2 mL of amniotic fluid.

3. Place the tubes in a Refrigerate/Ambient Mailer, 5 lb (T329).

4. Fill remaining space with packing material.

**Additional Information:**

1. Unavoidably, about 1% to 2% of mailed-in specimens are not viable.

2. Bloody specimens are undesirable.

3. If the specimen does not grow in culture, you will be notified within 7 days of receipt.

4. Results will be reported and also telephoned or faxed, if requested.

**Acceptable:**

**Supplies:** CVS **Media** (RPMI) and Small Dish (T095)

**Specimen Type:** Chorionic villi

**Container/Tube:** 15-mL tube containing 15 mL of transport media

**Specimen Volume:** 20-30 mg

**Collection Instructions:**

1. Collect specimen by the transabdominal or transcervical method.

2. Transfer chorionic villi 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.

## 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

Amniotic Fluid: 2 mL; Chorionic Villi: 2 mg; If ordering in conjunction with other testing: If ordered with CHRAF: 12 mL; with CHRCV: 12 mg; with CMAP: 12 mL or 12 mg; with CHRAF/CHRCV and CMAP: 26 mL or 26 mg

## Reject Due To

No specimen should be rejected.

## Specimen Stability Information

| Specimen Type | Temperature | Time |
|---|---|---|
| Varies | Refrigerated (preferred) | |
| | Ambient | |

## Clinical and Interpretive

## Clinical Information

Approximately half of clinically recognizable spontaneous abortions have a major chromosomal anomaly.

Up to 95% of chromosomal abnormalities diagnosed prenatally involve aneuploidy (gain or loss of whole chromosome) of chromosomes 13, 18, 21, X, and Y.

In liveborn infants, about 8/1,000 have a major chromosome anomaly, of which 6.5/1,000 involve aneuploidy of the 5 chromosomes analyzed by this test. Therefore, aneuploidy of chromosomes 13, 18, 21, X, and Y accounts for 81% to 95% of major chromosome anomalies in liveborn infants.

Techniques to detect aneuploidy include standard chromosome analysis and FISH. Standard chromosome analysis

from amniotic fluid cells or chorionic villi requires 5 to 9 days for culture, harvest, and analysis. FISH, which uses DNA probes and can be performed on cultured and uncultured cells, can rapidly detect aneuploidy of 13, 18, 21, X, and Y in uncultured amniotic fluid cells or chorionic villi. FISH-based analysis may be helpful in medically urgent evaluations of newborn infants suspected to have aneuploidy of any of these chromosomes.

## Reference Values

An interpretive report will be provided.

## Interpretation

An interpretive report will be provided.

## Cautions

The use of these probes has been approved by the Food and Drug Administration as a stand-alone test. However, we recommend that complete chromosome analysis (CHRAF / Chromosome Analysis, Amniotic Fluid or CHRCV / Chromosome Analysis, Chorionic Villus Sampling) or chromosomal microarray (CMAP / Chromosomal Microarray, Prenatal, Amniotic Fluid/Chorionic Villus Sampling) be performed in conjunction with this FISH test. In cases where the FISH analysis is normal, a chromosome analysis or chromosomal microarray allows for the potential identification of more complex abnormalities and the less common numeric abnormalities of other chromosomes. In cases where the FISH study is abnormal, chromosome analysis can determine whether the abnormality is due to aneuploidy or a complex structural abnormality, allowing for recurrence risk information for the family.

Interfering factors:

-Inadequate amount of specimen may not permit adequate analysis

-Exposure of the specimen to temperature extremes (freezing or greater than 30 degrees C) may kill cells and 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. (2007). ACOG Practice Bulletin No. 88, December 2007. Invasive prenatal testing for aneuploidy. Obstet Gynecol 110:1459-1467

2. Ward BE, Gersen SL, Carelli MP, et al: Rapid prenatal diagnosis of chromosomal aneuploidies by fluorescence in situ hybridization: Clinical experience with 4,500 specimens. Am J Hum Genet 1993;52:854-865

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;20:432-444

# Performance

## Method Description

This test is performed using probes for the centromere regions of chromosome X (DXZ1), Y (DYZ3), and 18 (D18Z1), and locus-specific probes for 13q14 and 21q22. For each probe set, 2 technologists each analyzed 50 interphase nuclei (100 total). Aneuploidy of chromosomes 13, 18, 21, X, and Y is reported.(Unpublished Mayo method)

### PDF Report

No

### Day(s) and Time(s) Test Performed

Samples processed Monday through Sunday. Results reported Monday through Friday, 8 a.m.-5 p.m. CST.

### Analytic Time

3 days

### Maximum Laboratory Time

4 days

### Specimen Retention Time

Until reported

### 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 has been modified from the manufacturer's instructions. 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 x 1-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)

88275-Interphase in situ hybridization, 100 to 300 cells, each probe set (if appropriate)

### LOINC® Information

# Test Definition: PADF

Prenatal Aneuploidy Detection, FISH

| Test ID | Test Order Name | Order LOINC Value |
|---|---|---|
| PADF | Prenatal Aneuploidy Detection, FISH | In Process |

| Result ID | Test Result Name | Result LOINC Value |
|---|---|---|
| 51937 | Result Summary | 50397-9 |
| 51939 | Interpretation | 69965-2 |
| 54553 | Result | 57317-0 |
| CG695 | Reason for Referral | 42349-1 |
| CG696 | Specimen | 31208-2 |
| 51940 | Source | 31208-2 |
| 51941 | Method | 49549-9 |
| 51938 | Additional Information | 48767-8 |
| 53861 | Disclaimer | 62364-5 |
| 51942 | Released By | 18771-6 |