MAYO CLINIC LABORATORIES

Test Definition: PADF

Prenatal Aneuploidy Detection, FISH

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, <25	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 the probe application, analysis, and professional interpretation of results for one probe set (2 individual fluorescence in situ hybridization probes). Additional charges will be incurred for additional probe sets 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.

Appropriate ancillary probes may be performed at consultant discretion to render comprehensive assessment. Any additional probes will have the results included within the final report and will be performed at an additional charge.

Special Instructions

- 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

Varies

Ordering Guidance

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 and is intended to be ordered in conjunction with chromosomal microarray or chromosome analysis.

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

Additional Testing Requirements

Normal fluorescence in situ hybridization (FISH) results will not exclude the majority of cytogenetically detectable abnormalities.

A chromosomal microarray study, CMAP / Chromosomal Microarray, Prenatal, Amniotic Fluid/Chorionic Villus Sampling is recognized by the American College of Obstetricians and Gynecologists as the most effective test to detect clinically relevant gains or losses of chromosomal material and should be ordered along with this test. This FISH test it does not substitute for complete cytogenetic analysis.(1)

Shipping Instructions

Advise Express Mail or equivalent if not on courier service.

Necessary Information

A reason for testing is requested with each specimen. The laboratory will not reject testing if this information is not provided; however, appropriate testing or interpretation may be compromised or delayed in some instances. If not provided, an appropriate indication for testing may be entered by Mayo Clinic Laboratories.

Specimen Required

Submit only 1 of the following specimens:

Preferred:
Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 to 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. If ordering with CMAP / Chromosomal Microarray, Prenatal, Amniotic Fluid/Chorionic Villus Sampling, submit a minimum of 12 mL.
4. If ordering with CHRAF / Chromosome Analysis, Amniotic Fluid, submit a minimum of 12 mL.
5. If ordering with both CMAP and CHRAF, then submit a minimum of 26 mL.
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.

Acceptable:
Specimen Type: Chorionic villi

Supplies: CVS Media (RPMI) and Small Dish (T095)
Container/Tube: 15-mL tube containing 15 mL of transport media
Specimen Volume: 20 to 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).
3. Using a stereomicroscope and sterile forceps, assess the quality and quantity of the villi and remove any blood clots and maternal decidua.
4. If ordering with CMAP / Chromosomal Microarray, Prenatal, Amniotic Fluid/Chorionic Villus Sampling, submit a minimum of 12 mg.
5. If ordering with CHRCV / Chromosome Analysis, Chorionic Villus Sampling, submit a minimum of 12 mg.
6. If ordering with both CMAP and CHRCV, then submit a minimum of 26 mg.

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

Amniotic fluid: 2 mL; Chorionic villi: 2 mg

Reject Due To

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

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Refrigerated (preferred)		
	Ambient		

Clinical & Interpretive

Clinical Information

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 to 1000 have a major chromosome anomaly, of which 6.5 to 1000 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 fluorescence in situ hybridization (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

When no significant abnormalities are detected by the targeted fluorescence in situ hybridization (FISH) probes, a simple descriptive interpretation is provided. When abnormal results are detected, a detailed interpretation is given, including an overview of the results and of their significance, a correlation to available clinical information, recommendations for additional testing, and contact information for the laboratory if there are additional questions.

A normal result does not rule out the possibility of birth defects, such as those caused by non-targeted chromosome abnormalities, submicroscopic cytogenetic abnormalities, pathogenic molecular variants, and environmental factors (ie, teratogen exposure). For these reasons, clinicians should inform their patients of the technical limitations of FISH analysis before the procedure is performed, so that patients may make an informed decision about pursuing the procedure.

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

Cautions

The use of these probes has been approved by the US 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 fluorescence in situ hybridization (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.

Low levels of mosaicism involving chromosomes 13, 18, 21, X, or Y may not be detected by this procedure.
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. Committee opinion No.682 summary: Microarrays and next-generation sequencing technology: The use of advanced genetic diagnostic tools in obstetrics and gynecology. Obstet Gynecol. 2016;128(6):1462-1463
2. American College of Obstetricians and Gynecologists. (2007). ACOG Practice Bulletin No. 88, December 2007. Invasive prenatal testing for aneuploidy. Obstet Gynecol. 2007;110(6):1459-1467
3. 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(5):854-865

4. 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):432-444

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 independently analyze 50 interphase nuclei (100 total). Aneuploidy of chromosomes 13, 18, 21, X, and Y is reported.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
3 to 4 days

Specimen Retention Time
Amniotic fluid: Discarded 14 days after results reported; Chorionic villi: Not retained

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

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
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 ID	Test Order Name	Order LOINC® Value
PADF	Prenatal Aneuploidy Detection, FISH	57317-0

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	85069-3
51938	Additional Information	48767-8
51942	Released By	18771-6
53861	Disclaimer	62364-5