MAYO CLINIC LABORATORIES

Test Definition: NADF

Newborn Aneuploidy Detection, FISH, Blood

Overview

Useful For

Screening for chromosomal aneuploidies of chromosomes 13, 18, 21, X, and Y in newborn peripheral blood specimens

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
_I099	Interphases, 25-99	No, (Bill Only)	No
_I300	Interphases, >=100	No, (Bill Only)	No
_IL25	Interphases, <25	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
_PBCT	Probe, +2	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

Whole blood

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. FISH testing should be ordered in conjunction with additional cytogenetic testing (CHRCB / Chromosome Analysis, Congenital Disorders, Blood; or CMACB / Chromosomal Microarray, Congenital, Blood), as it does not substitute for complete cytogenetic analysis.

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

Container/Tube:

Preferred: Green top (sodium heparin)

Acceptable: Lavender top (EDTA) or yellow top (ACD)

Specimen Volume: 4 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send specimen in original tube. **Do not aliquot**.
3. Other anticoagulants are not recommended and are harmful to the viability of the cells.
4. Cord blood is acceptable.

Forms

New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file.

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

Specimen Minimum Volume

1 mL

Reject Due To

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

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Ambient (preferred)		

	Refrigerated		

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 in 1000 have a major chromosome anomaly, of which 6.5 in 1000 involve aneuploidy of 1 of these 5 chromosomes.

Diagnosis of chromosomal disorders can be performed by chromosome analysis of uncultured blood, standard chromosome study, and the technique utilizing fluorescence in situ hybridization (FISH) based on interphase cells. Standard chromosome analysis takes 3 to 10 days and analysis from uncultured newborn blood is often unsatisfactory and labor-intensive. FISH-based methods facilitate rapid diagnosis of aneuploidy and may be helpful in medically urgent evaluations of newborn infants suspected to have aneuploidy of any of these chromosomes.

This test does not detect chromosomal aneuploidies other than 13, 18, 21, X, and Y or any structural anomaly that does not result in gain of these chromosomes.

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

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.

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 a complete chromosome analysis (CHRCB / Chromosome Analysis, Congenital Disorders, Blood) or chromosomal microarray (CMACB / Chromosomal Microarray, Congenital, Blood) be performed in conjunction with this test. In cases where the fluorescence in situ hybridization (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:
-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 specimen may not permit adequate analysis
-Improper packaging may result in broken, leaky, and contaminated specimen during transport

Clinical Reference

1. Jalal SM, Law ME. Detection of newborn aneuploidy by interphase fluorescence in situ hybridization. Mayo Clin Proc. 1997;72(8):705-710
2. Cassidy SB, Allanson JE: Management of Genetic Syndromes. 2nd ed. John Wiley and Sons; 2005:557
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(5):432-444
4. Carroll J, Wigby K, Murray S. Genetic testing strategies in the newborn. J Perinatol. 2020:40(7):1007-1016

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

4 weeks

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

88271x2, 88291-DNA probe, each (first probe set), Interpretation and report
88271x2-DNA probe, each; each additional probe set (if appropriate)
88271x1-DNA probe, each; coverage for sets containing 3 probes (if appropriate)
88271x2-DNA probe, each; coverage for sets containing 4 probes (if appropriate)
88271x3-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

Test ID	Test Order Name	Order LOINC® Value
NADF	Newborn Aneuploidy Detection, FISH	57318-8

Result ID	Test Result Name	Result LOINC® Value
51930	Result Summary	50397-9
51932	Interpretation	69965-2
54552	Result	57318-8
CG694	Reason for Referral	42349-1
51933	Specimen	31208-2
51934	Source	31208-2
51935	Method	85069-3
51931	Additional Information	48767-8
51936	Released By	18771-6
53862	Disclaimer	62364-5