MAYO CLINIC LABORATORIES

Test Definition: NADF

Newborn Aneuploidy Detection, FISH

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 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

- 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

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: 4 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.
3. Advise Express Mail or equivalent if not on courier service.
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. The following documents are available in Special Instructions:
-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

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 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 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
An interpretive report will be provided.

Cautions
The use of these probes has been approved by the FDA 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 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
- 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;72:705-710
2. Cassidy SB, Allanson JE: Management of Genetic Syndromes. Second edition. Hoboken, NJ, John Wiley and Sons, 2005, p 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;20:432-444

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 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 Fl. (remaining supernatant/whole fluid aliquots): Discarded 14 days after report. Blood: 4 weeks. Products of Conception (identifiable fetal tissue): Cremated quarterly after results reported. All Other Specimens: Discarded when results reported.

Performing Laboratory Location
Rochester

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 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 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