MAYO CLINIC LABORATORIES

Test Definition: CMAFF

Chromosomal Microarray (CMA) Familial Testing, FISH

Overview

Useful For

Determining the inheritance pattern of copy number changes previously identified by chromosomal microarray analysis in a patient and aiding in the clinical interpretation of the pathogenicity of the copy number change

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
_ML10	Metaphases, 1-9	No, (Bill Only)	No
_M30	Metaphases, >=10	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

If the copy number change identified in a patient is below the level of resolution of FISH analysis, CMA studies will be required. In this circumstance, this test will be cancelled and CMACB / Chromosomal Microarray, Congenital, Blood will be performed.

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 application of 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

- Family Member Phenotype Information for Genomic Testing

Method Name

Fluorescence In Situ Hybridization (FISH)

NY State Available

Yes

Specimen

Specimen Type
Whole blood

Ordering Guidance
This test is used to confirm the presence of a specific copy number change in a family member after it has been identified by chromosomal microarray (CMA) testing in a patient previously tested at Mayo Clinic Laboratories. All family member studies will be charged unless otherwise specified in the proband report.

Consultation with the laboratory is required prior to submitting a specimen when the initial patient (proband) was tested elsewhere. Whenever possible, family member testing should be performed by the original testing laboratory. If this is not possible, call 800-533-1710 and ask to speak with a laboratory genetic counselor to determine if testing will be accepted. Failure to contact the laboratory prior to ordering may result in test cancellation.

Shipping Instructions
Advise Express Mail or equivalent if not on courier service.

Necessary Information
Completion of the Family Member Phenotype Information for Genomic Testing form is required. The use of parental testing for the evaluation of uncertain copy number variants requires parental phenotypic information.

Clinical information on the family member being tested is essential for appropriate test interpretation and must be provided by the ordering clinician.

Specimen Required
Specimen Type: Whole blood
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.

Additional Information: Provide the name of the child (originally tested family member) on the request form. If testing was performed outside of Mayo Clinic Laboratories, consultation with the laboratory is required prior to ordering this test.

Forms
Family Member Phenotype Information for Genomic Testing

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

Chromosomal microarray (CMA) is a method for detecting copy number changes (gains or losses) across the entire genome. When copy number changes are identified in a patient, parental studies are sometimes necessary to assess their clinical significance. Changes that are inherited from clinically normal parents are less likely to be clinically significant in the patient and *de novo* changes are more likely to be pathogenic.

To identify familial copy number changes in parents of previously tested patients, fluorescence in situ hybridization testing is utilized. the parental results will provide the context for interpretation of the patient's CMA results.

Reference Values

An interpretive report will be provided.

Interpretation

An interpretive report will be provided.

Cautions

The results of this test may be of uncertain clinical significance.

Clinical Reference

1. Shaffer LG, Kashork CD, Saleki R, et al: Targeted genomic microarray analysis for identification of chromosome abnormalities in 1500 consecutive clinical cases. J Pediatr. 2006 Jul;149(1):98-102
2. Baldwin EL, Lee JY, Blake DM, et al: Enhanced detection of clinically relevant genomic imbalances using a targeted plus whole genome oligonucleotide microarray. Genet Med. 2008 May;10:415-429

Performance

Method Description

Fluorescence in situ hybridization using locus-specific probes targeted to the region of copy number gain or loss identified by the patient's chromosomal microarray testing. Ten metaphases and 200 interphase nuclei are analyzed to determine if the region is duplicated and 15 metaphases to determine if the region is deleted.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available
9 to 28 days

Specimen Retention Time
Three months

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 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)
88273 w/modifier 52-Chromosomal in situ hybridization, less than 10 cells (if appropriate)
88273-Chromosomal in situ hybridization, 10-30 cells (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
CMAFF	CMA Familial Testing, FISH	In Process

Result ID	Test Result Name	Result LOINC® Value
52405	Result Summary	50397-9
52406	Result	62356-1
54644	Nomenclature	62378-5
52407	Interpretation	69965-2

CG781	Reason For Referral	42349-1
52408	Specimen	31208-2
52409	Source	31208-2
52410	Method	85069-3
52411	Released By	18771-6
55129	Additional Information	48767-8
53403	Disclaimer	62364-5