
Reporting Title: CMA Familial Testing, FISH**Performing Location:** Rochester**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 Requirements:

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.

Specimen Minimum Volume:

1 mL

Forms:

Family Member Phenotype Information for Genomic Testing

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

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
CMAFF	CG781	Reason for Referral	Plain Text	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
52405	Result Summary	Alphanumeric		50397-9
52406	Result	Alphanumeric		62356-1
54644	Nomenclature	Alphanumeric		62378-5
52407	Interpretation	Alphanumeric		69965-2
CG781	Reason For Referral	Alphanumeric		42349-1
52408	Specimen	Alphanumeric		31208-2
52409	Source	Alphanumeric		31208-2
52410	Method	Alphanumeric		85069-3
55129	Additional Information	Alphanumeric		48767-8
53403	Disclaimer	Alphanumeric		62364-5
52411	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

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)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
_PBCT	Probe, +2			No	No (Bill Only)
_PADD	Probe, +1			No	No (Bill Only)
_PB02	Probe, +2			No	No (Bill Only)
_PB03	Probe, +3			No	No (Bill Only)
_ML10	Metaphases, 1-9			No	No (Bill Only)
_M30	Metaphases, >=10			No	No (Bill Only)
_IL25	Interphases,			No	No (Bill Only)
_I099	Interphases, 25-99			No	No (Bill Only)
_I300	Interphases, >=100			No	No (Bill Only)

Reference Values:

An interpretive report will be provided.