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**Reporting Title:** Prenatal Aneuploidy Detection, FISH**Performing Location:** Rochester**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 Requirements:**

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.

**Specimen Minimum Volume:**

Amniotic fluid: 2 mL; Chorionic villi: 2 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 Type	Temperature	Time	Special Container
Varies	Refrigerated (preferred)		
	Ambient		

**Ask at Order Entry (AOE) Questions:**

Test ID	Question ID	Description	Type	Reportable
PADF	CG695	Reason for Referral	Plain Text	Yes

Test ID	Question ID	Description	Type	Reportable
PADF	CG696	Specimen: • Amniotic Fluid • Chorionic Villi	Answer List	Yes

**Result Codes:**

Result ID	Reporting Name	Type	Unit	LOINC®
51937	Result Summary	Alphanumeric		50397-9
51939	Interpretation	Alphanumeric		69965-2
54553	Result	Alphanumeric		57317-0
CG695	Reason for Referral	Alphanumeric		42349-1
CG696	Specimen	Alphanumeric		31208-2
51940	Source	Alphanumeric		31208-2
51941	Method	Alphanumeric		85069-3
51938	Additional Information	Alphanumeric		48767-8
53861	Disclaimer	Alphanumeric		62364-5
51942	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

**Supplemental Report:**

No

**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, &lt;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)
_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.