
Reporting Title: 22q11.2 Deletion/Duplication, FISH**Performing Location:** Rochester**Shipping Instructions:**

Advise Express Mail or equivalent if not on courier service.

Necessary Information:

[Provide a reason for testing 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.](#)

Specimen Requirements:**Submit only 1 of the following specimens:****Specimen Type:** Amniotic fluid**Container/Tube:** Amniotic fluid container**Specimen Volume:** 20-25 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.

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.
4. Results will be reported and also telephoned or faxed, if requested.

Specimen Type: Autopsy**Supplies:** Hank's Solution (T132)**Container/Tube:** Sterile container with sterile Hank's solution, Ringer's solution, or normal saline**Specimen Volume:** 1 cm(3) biopsy specimen of muscle/fascia from the thigh**Collection Instructions:**

1. Wash biopsy site with an antiseptic soap.
2. Thoroughly rinse area with sterile water.
3. **Do not** use alcohol or iodine preparations.
4. Biopsy specimens are best taken by punch biopsy to include full thickness of dermis.

Specimen Type: Blood**Container/Tube:** Green top (sodium heparin)**Specimen Volume:** 4 mL**Collection Instructions**

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

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: 20-25 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.

Specimen Type: Fixed cell pellet

Supplies: Hank's Solution (T132)

Container/Tube: Sterile container with a 3:1 fixative (methanol:glacial acetic acid)

Specimen Volume: Entire specimen

Specimen Type: Products of conception or stillbirth

Supplies: Hank's Solution (T132)

Container/Tube: Sterile container with sterile Hank's solution, Ringer's solution, sterile RPMI transport media, or normal saline

Specimen Volume: 1 cm(3) of placenta (including 20 mg of chorionic villi) **and** a 1-cm(3) biopsy specimen of muscle/fascia from the thigh

Collection Instructions If a fetus cannot be specifically identified, collect villus material or tissue that appears to be of fetal origin.

Additional Information: Do not send entire fetus.

Specimen Type: Skin biopsy

Supplies: Hank's Solution (T132)

Container/Tube: Sterile container with sterile Hank's solution, Ringer's solution, or normal saline

Specimen Volume: 1 cm(3) biopsy specimen of muscle/fascia from the thigh

Collection Instructions:

- 1. Wash biopsy site with an antiseptic soap.
- 2. Thoroughly rinse area with sterile water.
- 3. Do not use alcohol or iodine preparations.
- 4. A local anesthetic may be used.
- 5. Biopsy specimens are best taken by punch biopsy to include full thickness of dermis.

Forms:

1. **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)

2. [Final Disposition of Fetal/Stillborn Remains](#) (if fetal specimen is sent) in Special Instructions (Only for products of conception or stillbirth specimen).

3. If not ordering electronically, complete, print, and send a [Cardiovascular Test Request Form](#) (T724) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Refrigerated (preferred)		

	Ambient		
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Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
DD22F	CG669	Reason for Referral	Plain Text	Yes
DD22F	CG670	Specimen	Plain Text	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
51851	Result Summary	Alphanumeric		50397-9
51853	Interpretation	Alphanumeric		69965-2
54538	Result	Alphanumeric		62356-1
CG669	Reason For Referral	Alphanumeric		42349-1
CG670	Specimen	Alphanumeric		31208-2
51854	Source	Alphanumeric		31208-2
51855	Method	Alphanumeric		85069-3
51852	Additional Information	Alphanumeric		48767-8
51856	Released By	Alphanumeric		18771-6
53875	Disclaimer	Alphanumeric		62364-5

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)
- 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	Available Separately
_PBCT	Probe, +2	1	88271	No	No, (Bill Only)

_PADD	Probe, +1	1	88271	No	No, (Bill Only)
_PB02	Probe, +2	1	88271	No	No, (Bill Only)
_PB03	Probe, +3	1	88271	No	No, (Bill Only)
_ML10	Metaphases, 1-9	1	88273	No	No, (Bill Only)
_M30	Metaphases, >=10	1	88273	No	No, (Bill Only)
_IL25	Interphases, <25	1	88274	No	No, (Bill Only)
_I099	Interphases, 25-99	1	88274	No	No, (Bill Only)
_I300	Interphases, >=100	1	88275	No	No, (Bill Only)

Reference Values:

An interpretive report will be provided.