

Sex-Determining Region Y, Yp11.3 Deletion, FISH

Reporting Title: SRY, Yp11.3, FISH

Performing Location: Rochester

Additional Testing Requirements:

This test must be ordered in conjunction with conventional chromosome studies (CHRCB / Chromosome Analysis, Congenital Disorders, Blood).

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 balanced salt solution, Ringer's solution, or normal saline

Specimen Volume: 4 mm diameter

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:



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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 villus

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.

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

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 balanced salt solution, Ringer's solution, 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 balanced salt solution, Ringer's solution, or normal saline

Specimen Volume: 4 mm diameter

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.

Specimen Minimum Volume:

Amniotic fluid: 5 mL

Autopsy, skin biopsy: 4 mm

Blood: 2 mL

Chorionic villi: 5 mg Fixed cell pellet: 1 pellet

Products of conception: 1 cm(3)

Forms:

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- 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).

Specimen Type	Temperature	Time	Special Container
Varies	Refrigerated (preferred)		
	Ambient		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Туре	Reportable
SRYF	CG717	Reason for Referral	Plain Text	Yes
SRYF	CG718	Specimen:	Answer List	Yes

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
52003	Result Summary	Alphanumeric		50397-9
52005	Interpretation	Alphanumeric		69965-2
54565	Result	Alphanumeric		62356-1
CG717	Reason for Referral	Alphanumeric		42349-1
CG718	Specimen	Alphanumeric		31208-2
52006	Source	Alphanumeric		31208-2
52007	Method	Alphanumeric		85069-3
52004	Additional Information	Alphanumeric		48767-8
53850	Disclaimer	Alphanumeric		62364-5
52008	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.



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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
_1099	Interphases, 25-99			No	No (Bill Only)
_1300	Interphases, >=100			No	No (Bill Only)
_IL25	Interphases,			No	No (Bill Only)
_M30	Metaphases, >=10			No	No (Bill Only)
_ML10	Metaphases, 1-9			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)
_PB1	Probe Set, 1st			No	No (Bill Only)



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Reference Values:

An interpretive report will be provided.