Reporting Title: Subtelomeric Region Anomalies, FISH
Performing Location: Rochester

Specimen Requirements:

Provide a reason for referral 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.

Forms:
1. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.
2. If not ordering electronically, submit a Cytogenetics/AFP Congenital Disorders Request Form (Supply T238) with the specimen.

Advise Express Mail or equivalent if not on courier service.

Submit only 1 of the following specimens:

Preferred:
Specimen Type: Blood
Container/Tube: Green top (sodium heparin)
Specimen Volume: 5 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.

Acceptable:
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. Place the tubes in a Styrofoam container (Supply T329).
2. Fill remaining space with packing material.
3. Unavoidably, about 1% to 2% of mailed-in specimens are not viable.
4. Bloody specimens are undesirable.
5. If the specimen does not grow in culture, you will be notified within 7 days of receipt.
6. Results will be reported and also telephoned or faxed, if requested.

Specimen Type: Chorionic villi
Container/Tube: 15-mL tube containing 15 mL of transport medium
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 (Supply T095).
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: Products of conception or stillbirth
Container/Tube: Sterile container with sterile Hank's balanced salt solution (Supply T132), Ringer's solution, or normal saline
Specimen Volume: 1 cm(3) of placenta (including 50-mg of chorionic villi) and a 1-cm(3) biopsy specimen of muscle/fascia from the thigh
Collection Instruction: 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.
Forms: Final Disposition of Fetal/Stillborn Remains (if fetal specimen is sent) in Special Instructions

Specimen Type: Skin biopsy
Container/Tube: Sterile container with sterile Hank's balanced salt solution (Supply T132), 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 Type	Temperature	Time
Varies	Ambient (preferred)	
	Refrigerated	

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
FRTEL	G_547	Source: • Whole blood Na Hep • Amniotic Fluid • Chorionic Villi • Products of Conception or Stillbirth • Skin Biopsy	Answer List	Yes
FRTEL	G_549	Reason For Referral	Plain Text	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
17184	Specimen	Alphanumeric		31208-2
17185	Specimen ID	Alphanumeric		N/A
G_547	Source	Alphanumeric		N/A
17187	Order Date	Alphanumeric		N/A
G_549	Reason For Referral	Alphanumeric		42349-1
17188	Method	Alphanumeric		In Process
17190	Results	Alphanumeric		In Process
17191	Interpretation	Alphanumeric		69965-2
17192	Amendment	Alphanumeric		In Process
17193	Consultant	Alphanumeric		N/A
17194	Report Date	Alphanumeric		N/A

Components:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
Billing only	Cytogenetic Interp/Report	1	88291		
Billing only	Chrom situ hybrid 10-30	2	88273		
Billing only	DNA probe, each	41	88271		

Reference Values:

An interpretive report will be provided.