Reporting Title: 15q11.2 Duplication, 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 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 (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: 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
DUP15	G_478	Source: • Whole blood Na Hep • Amniotic Fluid • Chorionic Villi • Skin Biopsy	Answer List	Yes
DUP15	G_481	Reason For Referral	Plain Text	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
30106	Specimen	Alphanumeric		N/A
30130	Specimen ID	Alphanumeric		N/A
G_478	Source	Alphanumeric		31208-2
30134	Order Date	Alphanumeric		N/A
G_481	Reason For Referral	Alphanumeric		42349-1
30136	Method	Alphanumeric		49549-9
30137	Result	Alphanumeric		In Process
30138	Interpretation	Alphanumeric		69965-2

Result ID	Reporting Name	Type	Unit	LOINC®
30139	Amendment	Alphanumeric		In Process
30147	Reveiwed By	Alphanumeric		In Process
30148	Release Date	Alphanumeric		N/A

Components:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
Billing only	Interp and report	1	88291		
Billing only	Chromosomal in situ hybridization	1	88273		
Billing only	DNA Probe, each	2	88271		

Reference Values:

An interpretative report will be provided.