Reporting Title: Williams Syn, 7q11.23 Del, 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: 20-30 mg
Collection Instructions:
1. Collect specimen by the transabdominal or transcervical method.
2. Transfer chorionic villi to 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
WSCR	CG389	Source: • Whole blood Na Hep • Amniotic Fluid • Chorionic Villi • Skin Biopsy	Answer List	Yes
WSCR	CG392	Reason For Referral	Plain Text	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
16263	Specimen	Alphanumeric		N/A
16347	Specimen ID	Alphanumeric		N/A
CG389	Source	Alphanumeric		31208-2
16264	Order Date	Alphanumeric		N/A
CG392	Reason For Referral	Alphanumeric		42349-1
16265	Method	Alphanumeric		49549-9
16267	Results	Alphanumeric		48970-8
16268	Interpretation	Alphanumeric		69965-2

Result ID	Reporting Name	Type	Unit	LOINC®
16269	Amendment	Alphanumeric		48970-8
16270	Consultant	Alphanumeric		N/A
16271	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	Molecular Gene,10-30 cell	1	88273		
Billing only	Molecular Cyto, DNA Probe	2	88271		

Reference Values:

An interpretive report will be provided.