
Reporting Title: Williams Syn, 7q11.23 Del, FISH**Performing Location** Rochester**Shipping Instructions:**

Advise Express Mail or equivalent if not on courier service.

Necessary Information:

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.

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.

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 (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 (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 (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.

Forms:

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 in Special Instructions:

[-Informed Consent for Genetic Testing \(T576\)](#)

[-Informed Consent for Genetic Testing-Spanish \(T826\)](#)

| Specimen Type | Temperature | Time | Special Container |
|---------------|--------------------------|---------|-------------------|
| Varies | Refrigerated (preferred) | 0 hours | |
| | Ambient | 0 hours | |

Ask at Order Entry (AOE) Questions:

| Test ID | Question ID | Description | Type | Reportable |
|---------|-------------|---------------------|------------|------------|
| WS7F | CG680 | Reason for Referral | Plain Text | No |
| WS7F | CG681 | Specimen | Plain Text | No |

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 | Available Separately | Always Performed |
|---------|----------------|-----------|----------|----------------------|------------------|
| _PBCT | Probe, +2 | | 88271 x2 | No | No |
| _PADD | Probe, +1 | | 88271 x1 | No | No |
| _PB02 | Probe, +2 | | 88271 x2 | No | No |
| _PB03 | Probe, +3 | | 88271 x3 | No | No |

| | | | | | |
|-------|-----------------------|--|-------------------------|----|----|
| _ML10 | Metaphases, 1-9 | | 88273 w/ modifier 52 | No | No |
| _M30 | Metaphases, >=10 | | 88273 | No | No |
| _IL25 | Interphases, <25 | | 88274 w/modifier 52 | No | No |
| _I099 | Interphases, 25-99 | | 88274 | No | No |
| _I300 | Interphases, >=100 | | 88275 | No | No |

Result Codes for Reflex Tests:

| Test ID | Result ID | Reporting Name | Type | Unit | LOINC |
|---------|-----------|----------------|------|------|-------|
|---------|-----------|----------------|------|------|-------|

Reference Values:

An interpretive report will be provided.