Reporting Title: Miller-Dieker, 17p13.3 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:

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.
Specimen Stability Information: Refrigerated (preferred)/Ambient
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: Autopsy
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. Biopsy specimens are best taken by punch biopsy to include full thickness of dermis.
Specimen Stability Information: Refrigerated (preferred)/Ambient

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.
Specimen Stability Information: Ambient (preferred)/Refrigerated

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 the CVS 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 Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Fixed cell pellet
Container/Tube: Sterile container with a 3:1 fixative (methanol:glacial acetic acid)
Specimen Volume: Entire specimen
Specimen Stability Information: Ambient (preferred)/Refrigerated

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 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.
Specimen Stability Information: Refrigerated (preferred)/Ambient
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 Stability Information: Refrigerated (preferred)/Ambient

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Refrigerated	

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
FMDL	CG169	Source: • Amniotic Fluid • Autopsy • Blood • Chorionic Villi • Fixed Cell Pellet • Products of Conception or Stillbirth • Skin Biopsy	Answer List	Yes
FMDL	CG171	Reason For Referral	Plain Text	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
23321	Specimen	Alphanumeric		N/A
23322	Specimen ID	Alphanumeric		N/A
CG169	Source	Alphanumeric		31208-2
23324	Order Date	Alphanumeric		N/A
CG171	Reason For Referral	Alphanumeric		42349-1
23326	Method	Alphanumeric		49549-9
23327	Results	Alphanumeric		48974-0
23328	Interpretation	Alphanumeric		69965-2
23329	Amendment	Alphanumeric		In Process
23330	Reviewd By:	Alphanumeric		N/A
23331	Release 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 Cyto, DNA Probe	2	88271		
Billing only	Molecular Gene,10-30 cell	1	88273		

Reference Values:

An interpretive report will be provided.