

# Yale Molecular Cytogenetics Laboratory

## Lab Location:

WWW335, 333 Cedar Street, Department of Genetics, Yale Medical School  
P.O.Box 3333, New Haven, CT 06510

## Hours of Operation:

Monday-Friday, 8:00am-5:00pm

## Late Hour and Weekend Delivery:

Samples can be directed to Laboratory Central on the 5th floor of the Clinic Building, 789 Howard Avenue. Laboratory Central is located to the left of the elevator exit and is open 24 hours, 7 days a week. Parking is available at the entrance of 789 Howard Ave.

## Contact Persons:

Audrey Meusel, Laboratory Administrator, 203-785-2646

Joan Samuelson, Supervisor, 203-785-5140

Peining Li, Laboratory Director, 203-785-6317

## Specimen Requirements:

Each specimen must be accompanied with a completed REQUISITION FORM that provides:

- the patient's name, date of birth, gender, address, and insurance information
- specimen type, time/date of specimen collection, and test desired
- clinical indications, history, and therapy
- referring physician's name (not resident's), phone/fax/pager numbers, and address

\*NOTE: improper specimen and incomplete requisition form could cause delay in process or unsuccessful in analysis.

**Form-4100**

TEST & SPECIMEN TYPE	CLINICAL INDICATIONS	TAT (STAT)	COLLECTION	PROCESSING	SHIPPING
<b>Prenatal Chromosome Analysis</b>					
Amniotic Fluid (amniocentesis)  *Note the gestational age in weeks	AMA, ultrasound anomalies, abnormal screening, family history, etc.	10-14 days  (STAT or prelim available upon request)	Discard first 2mL of amniotic fluid. Collect 15-25mL in a sterile syringe.	Use sterile technique to transfer to 15mL sterile screw-top plastic conical tubes for transport. Uniquely identify twins. <b>Do not centrifuge.</b>	Maintain at room temperature.  <b>Send to Lab ASAP</b>
Chorionic villi sampling (CVS)  *Note the gestational age in weeks.	AMA, ultrasound anomalies, abnormal screening, family history, etc.	4-10 days  (STAT or prelim available upon request)	Collect 15-40mg of budding villi in 15mL screw-top plastic conical tubes.	Our lab will provide the screw-top tubes containing sterile transport media. <b>Do not centrifuge.</b>	Maintain at room temperature.  <b>Send to Lab ASAP</b>
Percutaneous umbilical blood sampling (PUBS) (Cordocentesis; periumbilical blood sampling; or fetal blood)	AMA, ultrasound anomalies, abnormal screening, family history, confirmatory study etc.	3-7 days	Collect 1-3mL in sterile, 15mL <b>sodium heparin</b> (green-top) vacutainer tube. Collect under ultrasound guidance.	Invert tube immediately upon completion of blood collection to prevent formation of clots.	Maintain at room temperature.  <b>Send to lab ASAP</b>
Products of conception (POC)  *Villi are specimen of choice, Prefer dual specimens: villi (or AF) and fetal tissues (skin). Must specify the type of specimen submitted	Spontaneous abortion, miscarriage, fetal demise, etc.	14-28 days	Collect 3-4mm <sup>2</sup> biopsy (about the size of a pencil eraser) by sterile procedure, and place in 15ml tube with transport media (provided by the cytogenetic lab).	including placental villi or skin biopsy, and other non-neoplastic organ tissue. (In case of fetal demise, a skin or organ specimen might be appropriate). Uniquely identify twins.	Maintain at room temperature. If transit is to be delayed for > 1 day, refrigerate it. <b>Do not freeze.</b>

<b>Constitutional Chromosome Analysis</b>					
Peripheral blood	Developmental delay, mental retardation, dysmorphic features, congenital defects, heart defects, autism, etc.	7-10 days STAT: 4-5 days,  Prelim available.	Infants, 1-2mL; Children, 3-5mL; Adults, 5-10mL; Collected into a sterile 15mL <b>sodium heparin</b> (green-top) vacutainer tube.	Invert tube immediately upon completion of blood collection to prevent formation of clots.	Maintain at room temperature. Send to laboratory the same day it is collected.
<b>Cancer Cytogenetic Analysis</b>					
Spicule-rich bone marrow aspirate; peripheral blood; bone core biopsy; lymph node; or effusion.  (Bone marrow is specimen of choice for leukemia. Peripheral blood could be the choice for CLL )	Leukemias, lymphomas, Multiple myeloma, myelodysplastic syndromes, etc	7-10 days	Collect 1-2mL bone marrow; 5-10mL blood; 0.5-1cm <sup>2</sup> biopsy (bone core; lymph node). Collect bone marrow or blood in sterile 15ml centrifuge tube with media (provided by cytogenetics lab).	Invert tube immediately upon completion of blood collection to prevent formation of clots. For bone marrow it is important that a first draw, spicule-rich sample be collected.	Maintain at room temperature.  <b>Send to lab ASAP.</b>
Solid tumor, neoplastic tissue  (inform the lab prior to surgery or needle aspiration to facilitate optimal specimen collections and culture set up)	Solid tumors	10-21 days	Collect 3-4mm <sup>2</sup> biopsy (about the size of a pencil eraser) in a sterile, screw-top container filled with transport media (provided by cytogenetics lab).	Ensure the container is tightly sealed to prevent leakage.	Maintain at room temperature. If transit is to be delayed for >1 day, refrigerate it. <b>Do not freeze.</b>
<b>Fluorescent In Situ Hybridization (FISH) Analysis</b>					
AneuVysion screen, Microdeletion testing, (amniotic fluid, CVS, POC, PUBS)  *Note the gestational age in weeks.	Rapid prenatal screen of aneuploidy in chromosomes X/Y/18 and 13/21; Rapid prenatal screen of microdeletions in 15q, 22q, etc	1-2 days	Follow specimen requirements for tissue type.		
Constitutional FISH: peripheral blood; or prenatal cases (AF, CVS, and POC)	Identify specific chromosomal abnormalities: aneuploidies, microdeletions, deletions, translocations, inversions, etc	1-7 days STAT: 1-2 days	Follow specimen requirements for tissue type.		
Cancer FISH test: Bone marrow; solid tumor; paraffin blocks	deletions, translocations, inversions, etc	1-7 days STAT: 1-2 days	Follow specimen requirements for tissue type.		
Gender determination	Ambiguous genitalia (SRY), XX/XY sex-mismatch transplant.	1-7 days STAT: 1-2 days	Follow specimen requirements for tissue type.		
Subtelomeric study  Peripheral blood (Check with lab for other tissues).	Identify cryptic subtelomeric imbalances.	7-14 days	See constitutional blood section.		