Yale Molecular Cytogenetics Laboratory

Lab Location:
Yale University School of Medicine
Department of Genetics
Cytogenetics Laboratory
333 Cedar Street, WWW335/338
P.O. 208005
New Haven, CT 06520-8005

Specimen Mailing Address:
Core Laboratory
55 Park Street
5th Floor
New Haven, CT 06510

Hours of Operation:
Monday-Friday, 8:00am-5:30pm

Late hour and weekend delivery:
Samples can be directed to the Core Laboratory at 55 Park Street, which is located on the fifth floor and is open 24 hours, 7 days a week. For STAT samples outside of routine hours, please e-mail both the Lab Manager and Lab Director.

Contact Persons:
Audrey Meusel, Secretary, 203-785-2146, Audrey.Meusel@yale.edu
Autumn DiAdamo, Laboratory Manager, 203-785-5001, Autumn.DiAdamo@yale.edu
Peining Li, Laboratory Director, 203-785-6317, Peining.Li@yale.edu

Requisition forms
Available at: http://medicine.yale.edu/labs/cytogenetics/test_requisitions.html

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

- the patient's name, date of birth, and gender
- 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 collection or incomplete requisition could cause processing delay or unsuccessful analysis

Form-4100

TEST & SPECIMEN TYPE	CLINICAL INDICATIONS	TAT (STAT)	COLLECTION	PROCESSING	SHIPPING
Prenatal Chromosome Analysis					
Amniotic Fluid (AF) *Note the gestational age in weeks and if AFP testing is needed.	AMA, ultrasound anomalies, abnormal screening, family history, etc.	10-14 days	Discard first 2mL of amniotic fluid. Collect 15-25mL in a sterile syringe.	Use sterile technique to transfer to sterile 15mL screw-top plastic conical tubes for transport. Uniquely identify twins. Do **not** centrifuge.	Maintain at room temperature. Send to Lab ASAP
Chorionic villi sampling (CVS) *Note the gestational age in weeks.	AMA, ultrasound anomalies, abnormal screening, family history, etc.	7-10 days	Collect 20-35mg of budding villi in sterile 15mL screw-top plastic conical tubes containing RPMI.	Contact lab for tubes containing sterile transport media. Do **not** centrifuge.	Maintain at room temperature. Send to Lab ASAP
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 sodium heparin (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. Send to lab ASAP

Products of conception (POC) *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^2 biopsy (about the size of a pencil eraser) by sterile procedure. (For POCs, villi are specimen of choice)	Contact lab for tubes containing sterile transport media OR transfer specimen to 15ml or 50ml conical tube containing RPMI or HEPES. Uniquely identify twins.	Maintain at room temperature. If transit is to be delayed for > 1 day, refrigerate it. Do **not** freeze.
Constitutional Cytogenomic Analysis					
Peripheral blood (PB) (aCGH as first tier test, and karyotype for chromosome syndrome or structural abnormality)	Developmental delay (DD), mental retardation (MR), dysmorphic features, congenital defects, heart defects, autism (ASD), etc.	7-10 days STAT: 3-5 days,	Infant, 1-2mL; Child/Adult, 3-5mL Collected into a sterile sodium heparin (green-top) vacutainer tube.	Invert tube immediately upon completion of blood collection to prevent formation of clots.	Maintain at room temperature. Send to laboratory within 24 hours.
Cancer Cytogenetic Analysis					
Spicule-rich bone marrow aspirate; peripheral blood; bone core biopsy; lymph node; effusion. (Bone marrow is specimen of choice for leukemia. Peripheral blood for CLL.)	Leukemias, lymphomas, multiple myeloma, myelodysplastic syndromes, etc.	5-7 days	Collect 1-2mL bone marrow; 5-10mL blood; 0.5-1cm^2 biopsy (bone core; lymph node). Collect bone marrow or blood in a sterile sodium heparin (green-top) vacutainer tube.	Invert tube immediately upon completion of sample 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. Send to lab ASAP.
Solid tumor, neoplastic tissue	Solid tumors.	14-28 days	Collect 3-4mm^2 biopsy (about the size of a pencil eraser) in a sterile, screw-top container filled with sterile transport media (RPMI or HEPES).	Ensure the container is tightly sealed to prevent leakage.	Maintain at room temperature. If transit is to be delayed for >1 day, refrigerate it. Do **not** freeze.
Fluorescence In Situ Hybridization (FISH) Analysis					
AneuVysion screen, (AF, CVS, POC, PUBS) *Note the gestational age in weeks.	Rapid prenatal screen of aneuploidy in chromosomes X/Y/18 and 13/21.	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.	3-10 days STAT: 3-5 days	Follow specimen requirements for tissue type.		
Cancer FISH: Bone marrow; solid tumor; leukemic blood	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.		