



INDIANA UNIVERSITY

DEPARTMENT OF MEDICAL AND MOLECULAR GENETICS
School of Medicine

Specimen Criteria for Clients – Cytogenetics Laboratory

- Cytogenetics Laboratory requisition forms for cytogenetic analysis, chromosomal microarray (CMA), and fluorescence *in situ* hybridization (FISH) tests are available from the laboratory by fax, email, or on the cytogenetics website at <http://geneticslab.medicine.iu.edu/Requisition-Forms.html>. The appropriate requisition sheet should be completed by the physician, nurse, or physician's representative and accompany each sample.
- Please fill in all lines on the request form. Clinical information is necessary to help direct the analysis and interpretation.
- Send specimens in a plastic ziplock biohazard bag, and place paperwork in a separate bag or envelope. **Do not place paperwork in the same bag as the specimen.**
- For media and supplies (amniocentesis kits and/or bone marrow or tissue transport media), please phone the laboratory at (317) 274-2243. Frozen transport media should be stored in the freezer and completely thawed before usage.
- If a specimen is: 1) received in the incorrect container; 2) mislabeled; 3) of an insufficient quantity; or 4) otherwise of poor quality, the laboratory assistant or other staff member will contact the referring physician and, if possible, the person responsible for collecting the specimen, to alert them of the problem. In instances of mislabeling, written documentation clarifying the identity of the specimen is required from the individual who collected the specimen. If written documentation is not provided, the specimen will be discarded.
- To change test orders (e.g., add FISH studies), a physician may do so over the phone. A written order requesting the additional test must follow. Signed FAX orders are permitted.

Overnight Shipping Information

Outside of Indianapolis, please ship by FedEx or other overnight courier to:

IU Cytogenetic Laboratories
Department of Medical and Molecular Genetics
975 W. Walnut St.
IB 350
Indianapolis, IN 46202

Please call the laboratory and alert us that a specimen is en route.

Data Manager/Office – (317) 274-2243
Laboratory Manager – (317) 274-5219
Laboratory Assistant – (317) 278-6528
All Laboratories – (317) 274-2246 or (317) 274-1053

Shipped samples should **not** arrive on Sunday. If a sample must arrive on a Saturday or on a designated holiday, please inform the laboratory personnel to ensure that someone is available for receipt and processing. If sending a sample via FedEx for a Saturday delivery, make sure "Saturday Delivery" is indicated on the airbill. Shipping guidelines can be found online at <http://phmsa.dot.gov/hazmat/standards-rulemaking/regulations>. Click "Electronic Code of Federal Regulations (e-CFR: Title 49 CFR Parts 100-185)." From that page, click "100-177," then click "173.1 to 173.477," and then "§173.199 Category B infectious substances."

Delivery Information

There is a loading dock behind the Medical Research & Library Building (IB) for short-term parking during delivery.

Laboratory Hours

The laboratory is open from 7:30 a.m. to 5:30 p.m., Monday – Friday, and 8:00 a.m. to 4:00 p.m. on Saturday. The laboratory office is open from 8:30 a.m. to 5:00 p.m., Monday - Friday. Holidays are covered by “on call” staff.

Collection of Specimens for Cytogenetic Analysis

1. Peripheral Blood for Chromosome and/or FISH Analysis: **Dark Green-top Sodium heparin tubes.** Collect 2-10 mL of blood into dark green-top sodium heparinized tubes (2-4 mL for infants; 7-10 mL for adults). DO NOT FREEZE. Keep at room temperature. Final results should be available in 7-10 days for standard chromosome analysis. Preliminary results of RAPID specimens are provided within 3 days and final results within 7 days. The referring physician will be notified if results are abnormal or if cultures result in no growth or contamination.
2. Peripheral Blood for Chromosomal Microarray (CMA): **One (1) Dark Green-top Sodium heparin and One (1) EDTA tube.** Collect 3 mL whole blood per tube into one purple-top EDTA and one dark green-top sodium heparin tube. DO NOT FREEZE. Keep at room temperature, and deliver or ship specimen. Results should be available in 10-14 days. Specimens not meeting criteria listed in next section (#6) are subject to rejection.
3. Buccal Swab for Chromosomal Microarray (CMA): **ORAc collect•Dx OCD-100 (supplied by the laboratory).** Ensure the sponge tip does NOT come into contact with any surface prior to collection. Donor should NOT eat, drink, smoke or chew gum for 30 min prior to collection. Place sponge between cheek and lower gum (see package images) and gently rub in a back and forth motion, 10 times on each side. Holding the tube upright, unscrew the blue cap, turn the cap upside down, insert the sponge into the tube and close cap tightly. Invert the capped tube and shake vigorously 10 times.
4. Peripheral Blood for Fanconi Anemia Stress Test: **Two (2) Dark Green-top Sodium heparin tubes.** Collect 7-12 mL of blood into dark green-top sodium heparinized tubes (EDTA tubes will not be accepted). DO NOT FREEZE. Keep at room temperature, and deliver or ship specimen. Final results should be available in 7-10 days. The referring physician will be notified if cultures result in no growth or contamination.
5. Amniotic Fluid for Chromosomes, FISH, CMA: **Sterile tubes (provided by the laboratory).** Collect 20-25 mL of amniotic fluid at 16 weeks of gestation or greater in a sterile syringe. **Discard the first 2-3 mL of aspirated fluid, or place in a tube labeled “0” to avoid maternal cell contamination.** Transfer the remaining specimen to sterile centrifuge tubes in 3-4 sterile aliquots, labeled 1st, 2nd, etc., and transport to the laboratory within 24 hours. If MCC is also requested, one EDTA tube of maternal blood (3 mL) should accompany the specimen. The amniotic fluid should be refrigerated if there is a delay in transport. If the fluid sample is bloody, sterile centrifuge tubes containing sodium heparin should be used (these tubes are also available upon request). Results and final reports are faxed in 7-10 days. The referring physician and/or genetic counselor may be notified if the date of preliminary report exceeds 8 days.
6. Chorionic Villi Specimens (CVS) for Chromosomes, FISH, CMA: **Sterile container with transport media (provided by the laboratory).** Collect 20-30 mg of chorionic villi, either transabdominally or transcervically, place in CVS transport media, and transport at room temperature to the laboratory within 24 hours. The sample should be refrigerated if there is a delay in transport. If MCC is also requested, one EDTA tube of

maternal blood (3 mL) should accompany the specimen. Preliminary results should be available in 7-10 days, and the physician and/or genetic counselor may be contacted if the preliminary report exceeds 9 days.

7. Bone Marrow Specimens for Chromosome and/or FISH Analysis of Hematologic Disorders: **Sodium heparin tube, heparinized syringe, or media in flasks (provided by the laboratory).** Collect 2 mL of bone marrow aspirate into a heparinized syringe using the first draw or a repositioned needle, and place in containers of medium supplied by this laboratory, or into sodium heparinized tube. DO NOT FREEZE. Keep at room temperature. Final reports are sent within 7-10 days. The referring physician may be notified if the date of final report exceeds 10 days.
8. Rapid Bone Marrow Specimens (for urgent diagnosis in newborns): **Sodium heparin tube, heparinized syringe, or media in flasks (provided by the laboratory).** Collect a bone marrow aspirate (~1 mL) into a heparinized syringe and placed in containers of medium supplied by this laboratory. DO NOT FREEZE. Keep at room temperature. Preliminary results are available in 3-4 hours. The physician will also be notified immediately if cultures result in no dividing cells for analysis.
9. Solid Tumors: **Sterile container with transport media (supplied by the laboratory).** Collect solid tumor specimens aseptically, place in medium supplied by the laboratory, and immediately transport to the laboratory. If there is a delay in shipping, refrigerate the specimen. Time for analysis varies with rate of tumor growth, but most results are available in 7-10 days.
10. Paraffin-embedded Tissue (PET): Tissue Sections. Use 4-micron tissue sections from formalin-fixed, paraffin-embedded blocks on positively-charged slides. 2-3 slides per target area are sufficient. The laboratory does not accept blocks. **The tissue must be fixed in 10% buffered formalin, and the cold ischemia time and total time of exposure to formalin should be recorded when available.** A specimen should be accompanied by an H & E stained slide with the tumor area marked. A pathology report of the tissue to be studied must also be sent. **Decalcified bone specimens will not be accepted.**
11. Tissue from Spontaneous Abortions, Stillborns, Terminations or Autopsy Specimens: **Sterile container with transport media (supplied by the laboratory).** Clean fetal skin with 70% alcohol, not betadine or any antiseptics with metal ions. Collect 3-10 mm³ of tissue aseptically, and place in tissue transport media. If tissue transport media is unavailable, use any type of sterile media in a sterile container. Villi is preferred, although fetal cartilage, membrane, or tendon are acceptable. Typically, tissue from fetal organs (lung, liver, etc.) and skin is not viable. Do not send the entire fetus. Place each tissue type in a separate collection media. Transport the specimen at room temperature to the laboratory within 24 hours of collection, and refrigerate if there is a delay in transport. If MCC is also requested, one EDTA tube of maternal blood (3 mL) should accompany the specimen. Time for analysis varies with rate of tissue growth, but most results are ready in 12-14 days. **SINCE LIVING CELLS ARE REQUIRED, THE SPECIMEN SHOULD NOT BE FROZEN, PLACED ON ICE, OR DROPPED INTO FIXATIVE. DO NOT PLACE TISSUE IN FORMALIN OR SALINE.**
12. Skin Biopsies or Surgical Specimens: **Sterile container with transport media (supplied by the laboratory).** Clean the skin with 70% ethanol, not betadine or any antiseptics with metal ions. Collect a skin punch or surgery skin specimen aseptically, and place in tissue transport media (available upon request). If tissue transport media is unavailable, use any type of sterile media in a sterile container. Transport the sample at room temperature to the laboratory within 24 hours of collection, and refrigerate if there is a delay in transport. If the skin sample is for culturing for a send-out test, a completed requisition form for the send-out laboratory must also accompany the sample. If the sample is for cytogenetic analysis, a result should be available in approximately 21 days. The physician or

genetic counselor will be contacted if the report exceeds 25 days. If the sample is for Fanconi anemia breakage studies, a result should be available in 6 weeks. These specimens grow slowly, and usually require 4 weeks for results. The physician will be notified immediately if cultures result in no growth or contamination. **SINCE LIVING CELLS ARE REQUIRED, THE SPECIMEN SHOULD NOT BE FROZEN, PLACED ON ICE, OR DROPPED INTO FIXATIVE. DO NOT PLACE TISSUE IN FORMALIN OR SALINE.**

13. **Urine for UroVysion FISH Analysis: Sterile container.** These specimens may only be utilized for UroVysion FISH studies for cases of hematuria, bladder cancer, or suspicion of bladder cancer. For urine, collect a minimum of 50 mL in centrifuge tubes or other tightly-capped plastic container. First morning void is preferred. **DO NOT FREEZE.** Keep at room temperature. Transport the sample to the laboratory within 24 hours of collection, and refrigerate if there is a delay in transport. Specimens received in the laboratory later than 4 days after collection will be subject to rejection. A pathology report of the specimen to be studied must also be received if available. Final results should be available within 2 days of sample receipt.
14. **Biliary Strictures: Sterile container with ThinPrep PreservCyt® Solution from Hologic.** These specimens are collected for FISH studies utilizing centromere probes from chromosomes 3, 7, and 17 and a locus-specific probe for CDKN2A on 9p21. Transport biliary brushings or fluid collected via an Endoscopic Retrograde Cholangiopancreatography (ERCP) procedure to the laboratory at room temperature within 24 hours of collection; refrigerate if there is a delay in transport. **DO NOT FREEZE.** Specimens received in the laboratory later than 3 days after collection will be subject to rejection. Final results should be available within 2 days of sample receipt unless received on Friday afternoon.

Criteria for Rejection of Specimens

Fresh specimens [non-Paraffin Embedded Tissue (PET)] received that are older than **4 days** are subject to rejection. In this case, the physician or physician's representative will be contacted to determine the suitability of testing. After the laboratory staff explains the likelihood of successful testing, if the physician indicates that testing is still warranted, the patient will be billed full charge, and testing will be initiated. Clotted specimens or specimens that may be otherwise rejected may be accepted if another sample cannot be obtained.

Specimens are subject to rejection if:

1. Unlabeled, frozen, or grossly contaminated.
2. Packaged in improper containers.
3. The incorrect tissue type is sent.
4. Exposed to extreme temperatures.
5. Inadequate amount or volume.
6. Non-PET specimens have been placed in formalin.
7. Received in the laboratory after an extended period of time (4 days or more).
8. Decalcified bone specimens.

Specimens that are mislabeled should be re-collected if possible. If it is not possible to obtain a second specimen that is correctly labeled, a record should accompany the requisition form detailing the incident and which corrective actions were taken. We must receive a signed document regarding mislabeled specimen if a second specimen is not obtained.

The physician will be notified immediately if a specimen is considered unacceptable.

Specific rejection criteria for the different specimen types:

1. Amniotic Fluid Specimens for Chromosomes, CMA, FISH: Amniotic fluid may be rejected if it is frozen, clotted, more than 4 days old, or collected in non-sterile or improper containers.
2. Chorionic Villus Specimens for Chromosomes, CMA, FISH: Chorionic villus specimens may be rejected if there is less than 1 mg of tissue, or if the tissue is dehydrated, more than 48 hours old, sent in an open container, shipped in saline rather than medium, frozen, or placed in formalin.
3. Other Tissue Specimens: Other tissue specimens that have been placed in formalin (prior to culture) are rejected. Tissue specimens that are collected in saline or in an incorrect (non-sterile) container, frozen, more than 4 days old, or grossly contaminated may be rejected.
4. Bone Marrow and Tumor Specimens: Bone marrow and tumor specimens that are more than 96 hours old, collected into the wrong tube (e.g., lithium heparin, EDTA), frozen, clotted, collected in saline, or inadequate in amount (<0.5 mL of bone marrow) are subject to possible rejection.
5. Peripheral Blood Specimens for Chromosomes and/or FISH Analysis: Peripheral blood samples for cytogenetic analysis that are more than 4 days old, clotted, or collected in EDTA or lithium heparin tubes (for 24 hour or longer), frozen or on ice, or lacking identifying information on the tube may be rejected. Specimens from patients recently transfused (<1 week) with whole blood are discouraged.
6. Peripheral Blood Specimens for Chromosomal Microarray (CMA): Peripheral blood samples for CMA analysis that are more than 3-4 days old, do not meet volume requirements, are clotted, collected in lithium gel tubes (for 24 hour or longer), frozen or on ice, or lacking identifying information on the tube may be rejected.
7. Paraffin-Embedded Tissue (PET) FISH Analysis: Analysis can only be completed on tissue preserved in 10% buffered formalin. Do not send blocks. HER2 testing should only be performed on cases of invasive breast cancer or gastric cancer. **DECALCIFIED BONE SPECIMENS ARE NOT ACCEPTED FOR FISH ANALYSIS.**
8. Urine Specimens for UroVysion FISH Analysis: Urine samples for UroVysion FISH analysis that are more than 24 hours old, do not meet volume requirements, are frozen, lacking identifying information on container, grossly contaminated, or collected in non-sterile or improper containers may be rejected.
9. Biliary Strictures for FISH Analysis: Biliary brushings or fluid samples for FISH analysis that are kept at room temperature for more than 48 hours, lacking identifying information on the specimen container, grossly contaminated, or collected in non-sterile or improper containers may be rejected.
10. Buccal Swab Specimens for CMA: Samples may be rejected if there is insufficient volume (<0.5 ml), visible particulate matter, or collected in improper container.