

Specimen Requirements for Cytogenetics

General

Each specimen must be clearly labeled with at least two patient identifiers such as patient name and birth date. Each specimen must be accompanied by a paper test requisition including the following: name, birth date, gender, physician, originating lab or clinic, **clinical indication, and tests ordered**. The clinical indication is required for appropriate cell culture parameters to be chosen. Samples should be sent as soon as possible to the Cytogenetics Laboratory with same-day or overnight transport preferable. Specimen requirements for FISH correspond to the tissue type being studied (e.g., blood, bone marrow, amniotic fluid). *Samples should never be frozen or placed on ice.*

Amniotic Fluid

15-20 mL sterile amniotic fluid in sterile screw-capped tubes (centrifuge tubes, Falcon 2037 or equivalent). First few mLs drawn should be discarded to reduce chance of maternal cell contamination. Indicate on test requisition form if AFP and/or ACHE is requested. If prenatal interphase FISH aneuploidy screening is also requested (Aneuvysion™), a minimum of 20 mL amniotic fluid is required. Specimens with visible red blood will be rejected for Aneuvysion™ screening due to the increased likelihood of maternal leukocyte contamination. Aneuvysion results are typically available within 24 hours after specimen receipt at the laboratory; final written cytogenetic reports are usually available in 6 to 8 days. If additional tests are required, please contact the laboratory for test-specific amniotic fluid volume requirements.

Peripheral Blood

Aseptically draw venous blood into a *sodium*-heparin tube and mix well (or draw blood into syringe lubricated with *sodium*-heparin for injection). **Do not use EDTA, lithium, or ammonium-heparin.** Sample size is 2-5 mL for routine, family, and mosaicism studies (1 mL minimum), and 5-10 mL is required for high-resolution chromosome analysis. For patients also having molecular fragile X studies, draw 2-3 mL in *sodium*-heparin *and* an additional 7-10 mL in EDTA. If heel stick is necessary, cleanse area with alcohol and allow to dry. Collect blood in sterile capillary tubes and place into tube of prewarmed (37°C) transport medium obtained from Cytogenetics. For newborns, a minimum of 0.5 mL of blood is required for a routine analysis; high-resolution chromosome analysis is not typically offered for newborns due to the larger volume of peripheral blood required. Final written reports are normally available in 4 to 15 days. Verbal preliminary results for newborns with congenital anomalies are usually available within 24 to 48 hours, and a final written report is issued within 7 days in most cases.

In cases of fetal demise or stillbirth

Blood (peripheral heart puncture, or cord blood) if time of death is 2 days or less. Aseptically draw into *sodium* heparin tube.

Tissue Requirements for Spontaneous Abortion or Fetal Demise

Solid Tissue for Fibroblast Culture (products of conception or skin biopsies) *These samples must be taken before fixative (formalin) is used! Samples should never be frozen or placed on ice!*

Tissues should be transported in cell culture media containing antibiotics. Kits with tissue transport media and a requisition form are available from the laboratory for convenient sampling and shipping. Less desirable but acceptable for shipping is the use of normal saline or Hank's Balanced Salt Solution. Unacceptable sample conditions include samples that have been frozen or placed in fixative of any kind or samples placed in viral transport media.

Products of Conception 5 mm³ of placenta from near the umbilical cord insertion site containing chorionic villi and! or 1-3 mm³ of skin if autopsy is not ordered. If autopsy is performed, chest wall cartilage (particularly if fetus is macerated), gonad, spleen, kidney, or other internal organs can be submitted in addition to placental tissue. Tendons and umbilical cord are typically not good specimens due to difficulty in cell dissociation for cell culture. *In cases of fetal demise, the placenta maintains cell viability longer due to maternal blood circulation; therefore, placental specimens are more likely to yield cell growth and therefore a cytogenetic result.*

Place each tissue type in a separate tube with warmed transport media containing antibiotics obtained from the Cytogenetics Laboratory. Please include with the clinical information, the *approximate gestational age* and *fetal gender* if known. Keep sample at room temperature or refrigerated and send to the Cytogenetics Laboratory as soon as possible. Refrigerate if sample cannot be shipped immediately. Analysis may require 12 to 14 days or longer depending on tissue! cell viability and cell growth rate.

Skin or Other Tissues from Children or Adults

1-2 mm full-thickness skin punch biopsy placed in warm tissue culture media. If a mosaicism analysis is desired, please clearly indicate so on the test requisition form; this may require submission of more than one biopsy from the patient.

Bone Marrow

Add marrow (0.5 mL minimum) immediately into prewarmed (37°C) bone marrow transport medium obtained from Cytogenetics. Peripheral blood slide (or ACD tube), bone marrow slide, and CBC should be sent with sample if hematological interpretation is also requested. If transport medium is not available, a sodium heparin tube is acceptable. Preliminary results are available on request in 24 to 48 hours. Complete analyses and a final written report are normally available in 3 to 10 days.

Solid Tumors / Lymph Nodes

Solid tumor and lymph node specimens should be submitted in tumor transport media, available from the Cytogenetics Laboratory. No minimum specimen size is requested, but submission of at least 1-3 mm³ is desirable and will increase the likelihood of obtaining a meaningful result. Please include the tumor excision site, differential diagnosis, tumor status (necrotic, mitotic index if known), and patient clinical history in addition to other patient specimen information. Please ship these specimens as quickly as possible to Cytogenetics as delays in transport affect tumor cell viability.

HER2, Glioma, EGFR

Paraffin-embedded breast, brain or lung tumor tissue. Submission of an intact block is preferred; if not available, submit 2 to 4 slides with 3- to 4-micron-thick sections cut using a distilled-water bath. Include H&E stained slide for review.