

Instructions for Sampling Tissue for Cytogenetic Analysis

General

Each specimen must be clearly labeled with patient name and birth date. Requisition must supply name, birth date, gender, physician, originating lab or clinic, and clinical indication. Samples should be sent promptly to ICDL. Overnight shipping is acceptable where necessary. Courier service is available in some areas. Call ICDL for shipping information: (208) 381-5060. Sample requirements for FISH and chromosomal microarray correspond to the tissue type being analyzed (e.g. blood, bone marrow, amniotic fluid).

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 lithium heparin or ammonium heparin. Sample should be 1 – 5cc for newborns (if a heel stick is necessary, cleanse area with alcohol and allow to air dry). Collect blood in sterile capillary tubes and place into tube of pre-warmed (37°) transport medium obtained from ICDL). For testing on older children and adults, draw 5-10 cc; for Fragile X analysis, draw an additional 8-10cc in EDTA. Minimally, approximately 0.5cc of blood should be collected for cytogenetic analysis and a minimum of 1cc is required for microarray analysis. ***Please note, however, that if less than the minimum sample volume is received for microarray analysis and an aberration is identified, a redraw may be necessary in order to allow ancillary testing to establish chromosomal mechanism.

Metaphase cytogenetic results are normally available in 3 – 5 days; verbal preliminary stat results on newborns (metaphase chromosome based only) are usually available within 24 – 48 hours. Chromosome microarray results are typically available in 7-14 days.

In cases of fetal demise or stillbirth, aseptically draw peripheral heart puncture or cord blood into a sodium heparin tube.

Bone Marrow

Add marrow (0.5cc minimum) immediately into pre-warmed (37°) bone marrow transport medium obtained from ICDL. If transport medium is not available, a sodium heparin tube is acceptable. Preliminary results are available on request in 24 – 48 hours; complete analyses are normally available in within 5 days.

Solid Tissue for Fibroblast Culture

These samples MUST be taken before fixative (formalin) is used. Samples should never be frozen or placed on ice

Tissue requirements for spontaneous abortion or fetal demise characterization are 1 – 2mm³ of skin or placenta if autopsy is not ordered; chest wall cartilage (particularly if macerated), gonad, spleen, kidney, or other internal organs if autopsy is performed. Placenta tissue, including chorionic villi, is also preferred. Place each tissue in a separate tube with pre-warmed (37°) transport medium obtained from ICDL. Please include with clinical information the approximate gestational age, and fetal gender if known. Keep sample at room temperature or refrigerated and send to ICDL as soon as possible. Refrigerate if sample is not to be shipped immediately. Analysis may require 12 – 14 days or longer, depending on cell viability and if no cell growth is obtained, reflex to chromosome microarray is possible. Tissue requirements for skin or other body tissues from children or adults include 1 – 2mm full thickness punch biopsy placed in pre-warmed sterile (37°) tissue culture medium.

Amniotic Fluid

15 – 20cc sterile amniotic fluid in sterile screw-capped tubes (centrifuge tubes, Corning 430052 or equivalent). First few cc's drawn should be discarded to reduce chance of maternal cell contamination. Indicate on form if AFP and/or AChE are requested. Metaphase cytogenetic results are usually available within 7-8 days; those from chromosomal microarray within 7-14 days. If prenatal interphase FISH (Aneuvysion™) is also desired, a minimum of 20ccs of amniotic fluid is required.