

# SAMPLING INSTRUCTIONS AND TRANSPORT

## CYTOGENETIC SPECIMENS (FOR CHROMOSOMAL STUDIES)

- **Amniotic fluid: 20-30 cc** of fluid collected in one or two **sterile** 15ml conical centrifuge tubes (provided). Discard the first 2cc of fluid collected.

Amniocentesis is ideally performed **between the 15<sup>th</sup> and 18<sup>th</sup>** week of pregnancy, when the success rate for cultivating fetal cells is higher. Sampling of amniotic fluid is performed under conditions of **absolute sterility**.

The amniotic fluid may be stored at 4° C (refrigerator) for up to **48 hours** following sampling.

Ship via courier service at room temperature in the provided containers.

Do not heat or freeze.

- **Chorionic villi (CVS): 10-15 mg** in a 15ml **sterile** conical tube containing tissue culture medium (provided).

For CVS, it is recommended that sampling is performed around the **11<sup>th</sup> week** of gestation under conditions of absolute sterility.

The sample may be stored at 4° C (refrigerator) for up to 48 hours following sampling.

Ship via courier service at room temperature in the provided containers.

Do not heat or freeze.

- **Products of abortion:** chorionic villi (**CVS**) are preferred, but the analysis of other types of **fetal tissue** is also feasible. The tissue sample is placed in 15ml **sterile** conical tube containing tissue culture medium (provided). **We require a small amount of peripheral blood from the mother (in EDTA) to accompany the sample.**

Sampling must be performed under conditions of **absolute sterility**.

The sample may be stored at 4° C (refrigerator) for up to 48 hours following sampling.

Ship via courier service at room temperature in the provided containers.

Do not heat or freeze.

**Fetal blood:** Minimum 1 -2cc of fresh whole blood in sodium heparin tubes (green-top).

Sampling must be performed under conditions of **absolute sterility**.

The sample may be stored at 4° C (refrigerator) for up to 48 hours following sampling.

Ship via courier service at room temperature in the provided containers.

Do not heat or freeze.

- **Peripheral blood:** 3-5cc of fresh whole blood in sodium heparin tubes (green-top).

The sample may be stored at 4° C (refrigerator) for up to 48 hours following sampling.

Ship via courier service at room temperature in the provided containers.

Do not heat or freeze.

- **Bone marrow:** 2-3 cc of bone marrow aspirate in sodium heparin tubes (green-top). For samples requiring molecular genetic analysis (real time quantitative PCR), place a small aliquot (1-2 cc) into a separate EDTA tube (purple-top).

The sample may be stored at 4° C (refrigerator) for up to 48 hours following sampling.

Ship via courier service at room temperature in the provided containers.

Do not heat or freeze.

- **Tissue samples:** Biopsy samples should be at least 3mm X 5mm in area. Aseptically place biopsy in 15ml **sterile** conical tube containing tissue culture medium (sterile saline can be used if no media is available).

The sample may be stored at 4° C (refrigerator) for up to 48 hours following sampling.

Ship via courier service at room temperature in the provided containers.

Do not heat or freeze.

## **SPECIMENS FOR MOLECULAR GENETIC TESTING (DNA ANALYSIS)**

- **Peripheral blood:** adult: 10 cc, child: 6 cc, infant: 2-3 cc, in **EDTA** tube (purple-top).  
The sample may be stored at 4° C (refrigerator) for up to 48 hours following sampling.  
Ship via courier service at room temperature in the provided containers.  
Do not heat or freeze
- **Fetal Specimens:** Same sampling and shipment as for cytogenetic specimens.  
In case of cultures, please send two 90% confluent cultures in T-25 flasks.  
Ship via courier service at room temperature in the provided containers.  
Do not heat or freeze.
- **Tissue samples:** At least 2mg of tissue samples should be placed into a 2ml screw-cap tube containing sterile saline.  
The sample may be stored at 4° C (refrigerator) for up to 24 hours following sampling.  
Ship via courier service at room temperature in the provided containers.  
Do not heat or freeze.

### **Special notes and requirements for fetal specimens (prenatal testing):**

We require that the referring physician/center consult with us regarding unusual prenatal cases prior to submitting any samples. For the most accurate fetal results, parallel analysis of family members is often required. For all types of disorders, **prior knowledge of the parental/familial mutations is necessary** or blood specimens should be submitted from both parents and the affected sibling for molecular analysis. Certain disorders may require testing of additional family members for precise interpretation of fetal results. **Maternal blood is required for all prenatal tests to rule out maternal cell contamination.**

### **GENERAL NOTES**

All specimens should be clearly labeled with the patient's name. Fetal specimens should be clearly labeled with mother's full name and type of fetal sample (CVS or amnio). Always accompany the sample with the completed test requisition form. If in doubt, ALWAYS CONSULT WITH US before sampling and shipment.

**PLEASE COMPLETE THE PROVIDED TEST REQUISITION FORMS BY PROVIDING ALL THE NECESSARY INFORMATION INDICATED ON THE FORM. INCOMPLETE INFORMATION MAY RESULT IN TEST DELAY AND IS A POTENTIAL SOURCE OF ERROR. FURTHERMORE, CLINICAL AND OTHER USEFUL INFORMATION WILL INCREASE TESTING SENSITIVITY AND WILL PROVIDE BETTER INTERPRETATION OF TEST RESULTS.**