

SAMPLE TYPES	REQUIREMENTS
GENOMIC DNA	<p>Next Generation Sequencing (NGS)*: Send 1000 ng total gDNA @ > 15ng/μL. Please ship the samples in 10 mM TRIS. Do not use EDTA.</p> <p>*Please be aware that copy number assessment from genomic DNA extracted externally might be compromised. Please speak with a laboratory genetic counselor for more information.</p> <p>Sanger Sequencing: Send > 500 ng total gDNA @ > 15ng/μL (varies by size of the gene and the variants requested).</p> <p>Non-Sanger Sequencing Tests: Send > 500 ng total gDNA @ > 15ng/μL.</p> <p>**Required DNA quality: High molecular weight DNA (> 12 kb). A260/A280 reading should be ≥ 1.8. A260/230 should be a ratio range of 1.8 to 2.2. Contact the laboratory for specific amounts if total ng cannot be met.</p> <p>Shipping Instructions: Ship overnight at ambient temperature.</p>
CULTURED CELL (AMNIOCYTES OR CHORIONIC VILLUS SAMPLING [CVS])	<p>Collection Container(s): Two T-25 flasks</p> <p>Collection and Processing Instructions:</p> <ul style="list-style-type: none"> • Transfer cultured amniocytes or cultured CVS to two T-25 flasks at ≥80% confluence. • Cultures must be topped off with sterile medium immediately before shipping and shipped immediately at ambient temperature to the laboratory by overnight express with arrival Monday-Friday only. • For CVS and AF prenatal specimen, maternal cell contamination (MCC) testing is mandatory. • <u>Direct CVS and AF will not be accepted. If direct CVS and AF are received, the direct CVS and AF samples will be rejected and returned to provider. PerkinElmer will provide the tracking number for the returning samples.</u>
DIRECT PRODUCT OF CONCEPTION (POC) / FETAL TISSUE	<p>Collection Container(s): Sterile, screw-top container filled with tissue culture transport medium.</p> <p>Collection and Processing Instructions:</p> <ul style="list-style-type: none"> • If autopsy is performed: Facia lata, diaphragm, tendon, skin, tissue from internal organs (if fresh), chest wall cartilage (particularly if macerated) or placenta from fetal side • If no autopsy is performed: Placenta from fetal side is preferred (e.g., villi). Umbilical cord is also acceptable. • Please state clearly on the test requisition form (TRF) whether the tissue was fetal or placental in origin. • Transport POC (min: 10mg) in sterile, screw-top container filled with tissue transport medium. Do not place in formalin. We will NOT accept samples in formalin. • If specimen size is too large for a normal collection tube, a larger sterile container can be used such as a sterile urine cup and can be flooded with several tubes of cytogenetic tissue media. • Shipped immediately at ambient temperature or with cold pack especially under hot weather to the laboratory by overnight express with arrival Monday-Friday only. • Alternatively, fresh POC and/or tissue samples can be snap frozen and kept at - 80°C. Frozen tissue sample should be shipped overnight on dry ice with arrival Monday - Friday only. • For POC/fetal tissue, maternal cell contamination (MCC) testing is recommended but not required.
Sample Requirements for MCC Samples	
WHOLE BLOOD (EDTA)	<ul style="list-style-type: none"> • Collection Container(s): EDTA (purple top) • Collection and Processing Instructions: 5 to 10 mL. • Preferred Sample Condition: Store at ambient temperature. Do not refrigerate or freeze. • Shipping Instructions: Ship overnight at ambient temperature ensuring receipt within 5 days of collection. • Comments: Clotted or hemolyzed samples are not accepted.

Notes for sample preparation and shipping:

- PerkinElmer will provide a shipping label to use for shipping the sample to our lab.
 - Please call 1 (866) 354-2910 to arrange this. At this time, you will also be connected to a laboratory genetic counselor to answer any questions about the testing.
- Label specimen legibly with the following information, ensuring the identifiers on the specimen match the same information on the requisition:
 - Gestational carrier name (first and last)
 - Unique identifying number
 - This number may be the gestational carrier's date of birth or medical record number
- Fill out the TRF completely, including gestational carrier, facility, clinician demographics, specimen and clinical information. Please include ALL relevant clinical information, the clinician's NPI number and the corresponding ICD-10 codes. Please be sure to provide accurate contact information on the TRF as it will be used for communication of results.
- Place the specimen, requisition forms, and any additional information in the shipping container. Be sure to include labeled familial samples if applicable. Failure to perform this step will likely result in delay of testing.

For general questions on the collection and return of sample results, please call: PerkinElmer Genomics at +1 (866) 354-2910 (Monday-Friday, 8:00AM - 5:00PM EST) or by emailing genomics@perkinelmer.com