

SAMPLE TYPES	CODE	REQUIREMENTS
DRIED BLOOD SPOTS <i>PREFERRED SAMPLE TYPE</i>	DBS	<p>Collection Container(s): Dried blood spot card</p> <p>Collection and Processing Instructions: Follow kit instructions. Briefly, allow blood to saturate card until indicated areas are filled and blood has soaked through card. Air dry card at ambient temperature for at least 3 hours.</p> <ul style="list-style-type: none"> • NBS: Please contact PKIG to request the StepOne® kit. • Gene Sequencing: Please contact PKIG to request the DBS collection kit. <p>Preferred Sample Condition: Follow kit instructions. Store at ambient temperature.</p> <p>Shipping Instructions: Follow kit instructions. Double bag and ship overnight at ambient temperature.</p>
SALIVA <i>PREFERRED SAMPLE TYPE</i>	SV	<p>Collection Container(s): Oragene™ Saliva Collection Kit</p> <p>Collection and Processing Instructions: Collect saliva on an Oragene™ Saliva Collection Kit according to the manufacturer's instructions. Please contact PKIG to request the saliva collection kit for patients that cannot provide a blood sample as whole blood is the preferred sample.</p> <p>Preferred Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.</p> <p>Shipping Instructions: Ship overnight at ambient temperature.</p>
WHOLE BLOOD (EDTA) <i>PREFERRED SAMPLE TYPE</i>	WB	<p>Collection Container(s): EDTA (purple top)</p> <p>Collection and Processing Instructions: Infants (< 2-years): 2 to 3 mL; Children (>2-years): 3 to 5 mL; Older children and adults: 5 to 10 mL Southern Blot Analysis requires 3 mL blood.</p> <p>Preferred Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.</p> <p>Shipping Instructions: Ship overnight at ambient temperature ensuring receipt within 5-days of collection.</p> <p>Comments: Clotted or hemolyzed samples are not accepted.</p>
AMNIOTIC FLUID	AF	Please contact a PKIG Laboratory genetic counselor for these requests.
BONE MARROW	BM	<p>Collection Container(s): EDTA (Purple Top) or ACD (Yellow Top)</p> <p>Collection and Processing Instructions: Collect 2 to 3 mL of bone marrow.</p> <p>Preferred Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.</p> <p>Shipping Instructions: Ship overnight at ambient temperature ensuring receipt within 5-days of collection.</p>
CHORIONIC VILLI	CV	Please contact a PKIG Laboratory genetic counselor for these requests.
CORD BLOOD	CB	<p>Collection Container(s): EDTA (Purple Top) or ACD (Yellow Top)</p> <p>Collection and Processing Instructions: Collect 1 to 3 mL of fetal blood.</p> <p>Preferred Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.</p> <p>Shipping Instructions: Ship overnight at ambient temperature ensuring receipt within 5-days of collection.</p>
CULTURED AMNIOCYTES	CAF	Please contact a PKIG Laboratory genetic counselor for these requests.

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CULTURED CHORIONIC VILLI	CCV	<i>Please contact a PKIG Laboratory genetic counselor for these requests.</i>
CULTURED FIBROBLASTS	CF	<p>Collection Container(s): T25 or T75 flask</p> <p>Collection and Processing Instructions: Collect at least 1-T25 (or T75) with a confluency of 75 to 80%.</p> <p>Preferred Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.</p> <p>Shipping Instructions: Ship overnight at ambient temperature.</p>
DNA, ISOLATED	DNA	<p>Required DNA Quantity by Test Type*:</p> <p>Next Generation Sequencing (NGS): Send >500ng total gDNA @ > 15ng/μL. Please ship samples in 10mM Tris. No EDTA.</p> <ul style="list-style-type: none"> • Sanger Sequencing: Send 5 to 25 μg (varies by size of gene). Please contact the laboratory for specific amounts. • Non-Sequencing Tests: Send 20 μg • Array-based Tests: Send 10 μg <p>*Required DNA Quality: High molecular weight DNA (>12kb). A260/A280 reading should be ≥ 1.8.</p> <p>Shipping Instructions: Ship overnight at ambient temperature.</p> <p>Comments:</p> <ul style="list-style-type: none"> • Research Laboratories: DNA extracted in research laboratories is not acceptable. Only under exceptional circumstances (e.g. proband not available) will DNA extracted in a research laboratory be accepted for clinical testing. Additional testing (e.g. of other family members) may be required to confirm results. • Laboratories outside the United States: Non-US laboratories are not subject to CLIA regulations and will be reviewed on a case-by-case basis. Please call to speak with a laboratory genetic counselor prior to submitting a DNA sample from any non-CLIA certified laboratory. • Special Notes: If extracted DNA is submitted, information regarding the method used for extraction should be sent along with the sample.
FORMULIN-FIXED PARAFFIN EMBEDDED (FFPE) TISSUE	PE	<p>Collection Container(s): Block or slides</p> <p>Collection and Processing Instructions: Submit FFPE block and H&E stained slide <u>OR</u> submit two 15μm scrolls and H&E stained slide <u>OR</u> submit FFPE unstained slides (15 μm x 3) and H&E stained slide.</p> <p>Preferred Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.</p> <p>Shipping Instructions: Ship overnight at ambient temperature in an insulated container. Do not heat or freeze.</p> <p>Comments: Full strength liquid formaldehyde fixed tissue is NOT acceptable.</p>
Muscle Biopsy	MB	<p>Collection Container(s): Sterile container</p> <p>Collection and Processing Instructions: 1 to 2 mm in length or 100 mg is acceptable. Flash freeze sample upon collection using liquid nitrogen.</p> <p>Preferred Sample Condition: Store flash frozen sample at -20°C or colder.</p> <p>Shipping Instructions: Ship frozen sample overnight on dry ice. Not accepted on Saturdays.</p>

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PRODUCTS OF CONCEPTION	POC	<p>Collection and Processing Instructions: <i>Specific tissue of origin; Contact laboratory genetic counselor for further instructions.</i></p> <p>Preferred Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.</p> <p>Shipping Instructions: Ship same day or next day at ambient temperature.</p> <p>Comments: Please speak with a laboratory genetic counselor for these requests, as director approval is required. Specimen requirements are similar to cytogenetics requirements.</p>
SKIN BIOPSY	SK	<p>Collection Container(s): Sterile container with PKIG transport media or other sterile culture media (RPMI, Hanks Solution).</p> <p>Collection and Processing Instructions: Collect 1 to 2 cm pieces of tissue from a central location rather than a distal location to enhance cell viability and growth. Place in a sterile container with PKIG transport media or other sterile culture media. Use sterile dissection (no prep) for internal tissue. Tissue fixed in formalin cannot be used.</p> <p>Preferred Sample Condition: Store at ambient temperature. Do not refrigerate or freeze. In the absence of media, place in a sterile container with a small amount of sterile saline.</p> <p>Shipping Instructions: Ship same day or next day at ambient temperature.</p>