

SAMPLE TYPES	CODE	REQUIREMENTS
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: Minimum 5mL. The blood tube should be inverted several times immediately after blood collection to prevent coagulation.</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>
SALIVA	SV	<p>Collection Container(s): Oragene™ Saliva Collection Kit or ORAcollect-Dx kit</p> <p>Collection and Processing Instructions: Collect saliva on an Oragene™ Saliva Collection Kit ORAcollect-Dx 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>
DRIED BLOOD SPOTS	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. • For pre-punched DBS: The required minimum 6 punches with 3.2 mm or 4 punches 4.75 mm. <p>Preferred Sample Condition: Follow the instructions provided with the collection set. Store the dried blood at ambient temperature for up to two days. If the specimen cannot be sent as soon as it is dry; the filter paper should be placed in a sealable plastic bag and stored in a refrigerator (≤ 8 °C), or preferably in a freezer.</p> <p>Shipping Instructions: Follow kit instructions. Double bag and ship overnight at ambient temperature.</p>
GENOMIC DNA	DNA	<p>Required DNA Quantity by Test Type*:</p> <ul style="list-style-type: none"> • Next Generation Sequencing (NGS): Send >1000ng total gDNA @ > 15ng/μL. Please ship samples in 10mM Tris. Do not use EDTA. • Sanger Sequencing: Send > 500 ng total gDNA @ > 15 ng/μL (varies by size of the gene and the variants requested). • Non-Sanger Sequencing Tests: Send > 500 ng total gDNA @ > 15 ng/μL <p>* Required DNA Quality: High molecular weight DNA (>12kb). A260/A280 reading should be ≥ 1.8. A260/230 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> <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.

SAMPLE TYPES	CODE	REQUIREMENTS
CULTURED CELL FROM A PRENATAL SAMPLE (INCLUDING CULTURED CVS, AF, AND POC)	CCV	<p>Collection Container(s): Two T-25 flasks</p> <p>Collection and Processing Instructions:</p> <ul style="list-style-type: none"> • All prenatal specimens will be tested for maternal cell contamination (MCC). Send a maternal blood (EDTA tube) for comparison. If blood is unavailable, we will accept genomic DNA and Saliva sample types as stated above. • 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.
DIRECT PRODUCTS OF CONCEPTION (POC)/TISSUE	POC	<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"> • All prenatal specimens will be tested for maternal cell contamination (MCC). Send a maternal blood (EDTA tube) for comparison. • If the 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. • State clearly in the TRF whether the tissue was of fetal or placental in origin. • Transport products of conception and/or fresh tissue (minimum: 10 mg) in sterile, screw-top container filled with tissue transport medium. Do not place in formalin. • If the 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 - NO WEEKEND SHIPMENTS!

For prenatal samples, 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.