






Whole Genome Sequencing Family Member

	Test Code	D2999
	Test Summary	Family member test for WGS TRIO analysis
	Turn-Around-Time (TAT)*	6 - 8 weeks
	Acceptable Sample Types	Whole Blood (EDTA) (Preferred sample type) DNA, Isolated Dried Blood Spots Saliva
	Acceptable Billing Types	

Detailed Sample Requirements

Whole Blood (EDTA) (Preferred sample type)

Collection Container(s):

EDTA (purple top)

Collection:

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.

Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.

Shipping: Ship overnight at ambient temperature ensuring receipt within 5-days of collection.

SPECIAL INSTRUCTIONS: Clotted or hemolyzed samples are not accepted.

DNA, Isolated

Collection:

Required DNA Quantity by Test Type*:

- **Next Generation Sequencing (NGS):** Send >1000 ng total gDNA @ >15 ng/?L. Please ship samples in 10mM Tris. Do not use EDTA.
- **Sanger Sequencing:** Send >500 ng total gDNA @ >15 ng/?L (varies by the size of the gene and the variants requested).
- **Non-Sanger Sequencing Tests:** Send >500 ng total gDNA @ >15 ng/?L.

Sample Condition: * 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.

Shipping: Ship overnight at ambient temperature.

SPECIAL INSTRUCTIONS:

- **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.

Dried Blood Spots

Collection Container(s):

Dried blood spot card

Collection:

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.

- **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.
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.
Shipping: Follow kit instructions. Double bag and ship overnight at ambient temperature.

Saliva

Collection Container(s):

Oragene™ Saliva Collection Kit or ORAcollect-Dx kit

Collection:

Collect saliva on an Oragene™ Saliva Collection Kit ORAcollect-Dx kit according to the manufacturer's instructions.

Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.

Shipping: Ship overnight at ambient temperature.

SPECIAL INSTRUCTIONS: Please contact PerkinElmer to request the saliva collection kit for patients that cannot provide a blood sample as whole blood is the preferred sample.