Instructions for Ultrarapid Genomic Testing

This package contains:  
• Two (2) Dried Blood Spot (DBS) test strips  
• Two saliva swab packs  
• Return shipping for all samples

Collect two DBS test strips from the proband. Collect one saliva swab from each parent.

DBS Sample Collection and Preparation for Proband  
1. For detailed collection instructions and requirements, please refer to the DRIED BLOOD SPOT SAMPLE COLLECTION INSTRUCTIONS card included within the pack.  
2. Fill five (5) spots on EACH of the two (2) DBS test strips. Refer to the pictures included on the instructions card to ensure acceptable application.  
3. Allow DBS test strips to dry for three (3) hours at ambient temperature of 18° to 25°C (64° to 77°F).  
4. Place both cards in the provided return envelope (yellow envelope with urgent sticker affixed). Include test requisition form (TRF) in the envelope with the proband’s samples.

Saliva Collection and Preparation for Parent(s)  
1. For detailed collection instructions and requirements, please refer to SALIVA COLLECTION INSTRUCTIONS included within the box.  
2. Place each saliva sample into its own provided clear biohazard bag and place in the foam slot in the box. Place the box into the provided return envelope.  
3. If parent samples are shipping separately from the proband samples, please photocopy the TRF for each shipping envelope – if photocopying is easily available.

Before Shipping Ensure:  
✓ All specimens are labeled legibly with the following information, ensuring the identifiers on the specimen match the same information on the test requisition form (TRF).  
  • Proband or Parent name (first and last)  
  • Unique identifying number (this number may be the proband or parent date of birth or medical record number)  
✓ The TRF is filled out completely, including patient, facility, clinician demographics, specimen, and clinical information. Please be sure to provide accurate contact information on the TRF as it will be used for communication of results.  
✓ The specimens, requisition form, and any additional information are included in the correct shipping containers. Failure to perform this step will likely result in delay of testing.  
✓ SHIP PROBAND DBS TEST STRIPS AS SOON AS POSSIBLE. IF PARENT SAMPLES CANNOT BE SHIPPED ON THE SAME DAY AS THE PROBAND SAMPLES, PROVIDER MUST ORDER “PROBAND ONLY” ON THE TRF.  
✓ If Proband and Parent samples can be shipped on the same day, Provider should order “TRIO” option. Please place all specimens inside the same yellow shipping envelope with the urgent sticker affixed.  
✓ If only one Parent sample is available to ship same day as Proband sample, and the second Parent sample is arriving later, please select “TRIO” option on the TRF and note that in the comments section.  
✓ If only one Parent sample is available to ship same day as Proband sample, and the second Parent sample is not available and will never arrive, please select “TRIO” option on the TRF and note that in the comments section.  
✓ If Parent samples cannot ship with Proband sample, but will arrive at a later date, Provider must order Test Code D2314 Ultrarapid Whole Genome Sequencing, Family Member 1 and D2315 Ultrarapid Whole Genome Sequencing, Family Member 2 (if applicable) - and the turnaround time for the Parent results will be 4 weeks. This is in addition to ordering Proband Only (D2312).

Samples may be shipped at any time to our laboratory, but samples shipped on Saturday will not arrive and be accessioned until Monday. The turnaround time of 5 days begins once sample has arrived in our laboratory.

For general questions on the collection and return of sample results, please call PerkinElmer Genomics at +1 (866) 354-2910.