Urine sample collection for mitochondrial studies

Fresh/frozen muscle is the preferred sample type for mitochondrial testing; however, testing for the common mitochondrial variants and full sequencing of the mitochondrial genome is available on urine samples. Deletion/Duplication testing cannot be performed on urine as the DNA quantity is not sufficient.

1) A urine container (with patient label) should be provided to the patient with a Molecular Diagnostic Requisition requesting the MNM or LHON Variant Panel OR Mitochondrial full sequencing.

2) Have the patient collect 50 ml of “First morning” urine in the sterile container provided. Samples must be collected on a Sunday and/or Monday morning.

3) Request that an initial urine sample is collected- NOT “mid stream”

4) Have the patient write the time and date of collection on the Molecular Diagnostic Requisition.

5) The urine sample can be stored at room temperature until the patient is able to take it to the collection lab. Please ensure that the sample is taken to the lab within 24 hrs of collection.

6) The sample will then be transported from the collection lab to the Molecular Diagnostic Lab. Note that samples must arrive in the Molecular Diagnostic Lab within 7 days of collection.

7) The laboratory should ship the urine at room temperature to:

   Attention: Dr. Stacey Hume
   Edmonton Molecular Diagnostic Laboratory
   826 Medical Sciences Building
   University of Alberta Hospital
   Edmonton, AB T6G 2H7