

## **Protocol for the collection and shipment of DNA, blood for DNA extraction, and serum samples**

### **Shipping Arrangements**

1. Samples should be collected and dispatched to us in batches of at least 5-10.
2. Notify MYO-SEQ sample collection by e-mail ([lauren.phillips@ncl.ac.uk](mailto:lauren.phillips@ncl.ac.uk) or [katherine.johnson@ncl.ac.uk](mailto:katherine.johnson@ncl.ac.uk)) or phone (+44 (0) 191 241 8834) at least 5 working days prior to any planned shipment.
3. We will confirm that sufficient bar code labels, a sample dispatch form and packaging labels are already with you, and dispatch more if necessary.
4. Arrangements for a courier to collect the samples should be made by you. Shipments can be charged to our DHL account (# communicated upon request) or billed to us direct.
5. For each patient you should have a set of barcoded labels. Affix the relevant barcode to the sample tubes prepared as described below and fill in the sample dispatch form (preferably electronically). Place the samples in the packaging and add the form before collection by the courier.

### **DNA Samples**

1. MYO-SEQ requires **good quality** DNA samples (>10kb genomic DNA) at least 1ug of DNA in 40ul aqueous solution. The concentration should be in the range of 50ng/ul.
2. DNA samples can be sent at ambient temperature, but we would prefer to receive them frozen or refrigerated.
3. DNA samples should be labelled with the barcodes provided before freezing.

### **Blood Collection Protocol for DNA extraction**

1. Collect 4-10 ml blood in an EDTA tube (*we strongly recommend BD Safety-LOK TM*).
2. Invert the tube 5 times to ensure adequate mixing with the EDTA.
3. Label tubes with patient ID number, collection centre (study site) ID and date.
4. Store at -80°C.
5. Ship the frozen samples (in batches) on dry ice (at least 5 kg) to the MRC BioBank Newcastle (the address is given at the end of this document).

***Note: The process from blood draw to storage at -80°C should be completed within 3 hours.***

***Note: For DNA extraction, whole blood samples must be sent in original tubes without centrifugation.***

### **Serum Collection Protocol**

1. Collect (up to) 6 ml blood in a serum tube with clot activator (*we strongly recommend BD Vacutainer Serum Tube ref 368815*).
2. Invert the tube 5 times to ensure adequate mixing with the clot activator.
3. Allow to stand for 30 minutes at room temperature for clotting.

4. Centrifuge tubes at 1500g (rpm dependent on rotor) for 10 minutes.
5. Aliquot serum in 200 µl aliquots and transfer to 2 ml screw cap micro or cryo tubes (*we strongly recommend Sarstedt freezer screw cap micro tubes or Nunc sterile screw cap cryo tubes*).  
***Note: This process should be completed within one hour after centrifugation.***
6. Label tubes with patient ID number, collection centre (study site) ID and date.
7. Store at -80°C.  
***Note: The process from blood draw to storage at -80°C should be completed within 3 hours.***
8. Ship the frozen samples (in batches) on dry ice (at least 5 kg) to the MRC BioBank Newcastle (the address is given at the end of this document).

#### **General Notes:**

- It is very important for each centre to follow these protocols of standardised serum and blood collection.
- The first tube used for drawing blood should NOT be used for the serum sample, because the first tube could contain contaminating skin cells which could affect the analysis to be carried out.
- **Centres are kindly requested to send either DNA or whole blood for DNA extraction (although both is also okay).**
- Advance notice of the arrival of the serum/DNA/blood samples should be given either by email (preferred) or by phone to Dan Cox (contact details below).
- The serum and blood samples for DNA extraction must remain frozen throughout transport; transporting the samples on ≥5 kg of dry ice means that they remain frozen for up to 4 days.
- A reputable courier service (e.g. FedEx) should be used to ensure the package with the serum/DNA/blood sample batches arrives at the above address within 4 days from taking the samples out of the freezer.
- It must be ensured that the package arrives at the above address Mon-Fri, so the blood samples need to be taken on appropriate days.

Address for Shipment and further questions:

**Lauren Phillips or Dan Cox  
MRC Centre for Neuromuscular Diseases Biobank  
Institute of Genetic Medicine  
Newcastle University  
International Centre for Life  
Central Parkway  
Newcastle upon Tyne  
NE1 3BZ  
United Kingdom**

**Contact: Lauren Phillips + 44 191 241 8834 [lauren.phillips@ncl.ac.uk](mailto:lauren.phillips@ncl.ac.uk)  
Dan Cox + 44 191 241 8834 [dan.cox@ncl.ac.uk](mailto:dan.cox@ncl.ac.uk)**