

## **COLLECTION AND STORAGE OF A SKIN BIOPSY FOR FURTHER NF1-RELATED GENETIC STUDIES**

1. **Important:** The medium and the appropriate container for shipment can be obtained from the lab upon request. Please e-mail us at least 3 days prior to the surgery date at [medgenomics@uab.edu](mailto:medgenomics@uab.edu) so we can ship a package with the appropriate medium. The medium should be kept refrigerated until the surgery date. The morning of the surgery, the media should be placed at room temperature for use. If a surgery is rescheduled after we ship the media, please contact us so we can ship out fresh media since the media expires within 1-2 weeks of receipt.
2. The **skin biopsy** should be performed under sterile conditions. The skin should be disinfected with a 0.5% Chloramine solution or similar disinfectant. A local anesthesia that does not contain adrenaline should be used such as 2% Xylocaine.
3. The skin biopsy (**minimum** 2-3 “punch” biopsies, diameter 0.5 cm) is surgically removed from the affected **pigmented area** of the patient and immediately immersed in the sterile pre-incubation transport medium (provided by MGL). The location where the “punch” biopsies were taken needs to be clearly labeled on the specimen containers as well as on the phenotypic checklist, accompanying the specimen.
4. Parafilm squares are also provided in the collection kit and should be used to seal the medium vials once the biopsy samples have been placed inside. Please ensure that the vials are not leaking and have been appropriately labeled with the patient’s name, date of birth, and/or medical record number.
5. For patients whose blood has never been tested, please also submit 10 milliliters of **whole blood** collected in EDTA (purple top) tubes.
6. The skin biopsy must be kept at room temperature at all times. We need to culture melanocytes from the biopsy for the analyses and hence, living, viable cells are needed. The skin biopsy must be sent to the Medical Genomics Laboratory **using priority service shipment** (next day delivery). The viability of melanocytes declines quickly 24 hours after the biopsy is removed. Please ALWAYS inform us ([medgenomics@uab.edu](mailto:medgenomics@uab.edu) or 205-934-5562) prior to shipping the biopsy. Always provide tracking number of the shipment and company as well. Always include a form for customs, if appropriate.
7. Address:  
Medical Genomics Laboratory  
Attn. Dr. Ludwine Messiaen  
330 KAUL Human Genetics Building  
720 20th Street S  
Birmingham, AL 35294-0024