

Collection and Storage of a Skin Biopsy for NF1-Related Genetic Studies (Melanocyte culture)

1. Shipping Medium and Container:

- **Request the shipping medium and container** from the lab at least **1 week prior to surgery** by emailing **medgenomics@uabmc.edu**.
- The lab will send the appropriate **medium** and **specimen package** for biopsy collection.
- **Keep the medium refrigerated** until the surgery date. On the day of surgery, bring the medium to **room temperature**.
- If surgery is rescheduled after the medium has been shipped, contact the lab for fresh media, as it expires within **1-2 weeks**.

2. Skin Biopsy Collection:

- Perform the skin biopsy under **sterile conditions**.
- Disinfect the skin with **0.5% Chloramine solution** (or similar disinfectant).
- Use a **local anesthesia** without adrenaline, such as **2% Xylocaine**.
- **Minimum 2-3 punch biopsies** (diameter 0.3 cm) should be taken from **3 anatomically different pigmented areas**.
- Immediately place biopsies in the sterile **pre-incubation transport medium** (provided by MGL).
- Clearly label the **location of each biopsy** on the specimen containers and the **phenotypic checklist**.

3. Specimen Sealing and Labeling:

- Use **Parafilm squares** (provided in the kit) to seal the medium vials after placing the biopsies inside.
- Ensure that the vials are **leak-proof** and labeled with the **patient's name, DOB, and/or medical record number**.

4. Whole Blood Submission (If Applicable):

- If **blood has never been tested**, submit **10 ml of whole blood** collected in **EDTA (purple top) tubes**.

5. Shipment of Skin Biopsy:

- Keep the biopsy at **room temperature** at all times.
- **Living, viable melanocytes** are needed for analysis, so the skin biopsy must be sent to the **Medical Genomics Laboratory** via **priority service shipment** (next-day delivery).
- **Melanocyte viability declines quickly** (within 24 hours) after biopsy removal.
- **ALWAYS inform the lab** at **medgenomics@uabmc.edu** or **205-934-5562** prior to shipment. Provide the **tracking number** and **shipment company**, and include a **customs form** if applicable.

6. Shipping Address:

UAB Medical Genomics Laboratory
Attn. Dr. Yunjia Chen
720 20th Street South
Suite 330
Birmingham, AL 35233-2032