

COLLECTION/STORAGE OF NEUROFIBROMAS (PLEXIFORM/CUTANEOUS/INTRADERMAL/SPINAL) 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 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. Cutaneous neurofibromas should by preference be “shaved” off. Other types of tumors should be surgically removed from the patient. The biopsies should be a minimum of 5 mm in diameter and thickness and be **immediately** immersed in the sterile pre-incubation transportation medium:

DMEM + 10% FCS + Peni/Strepto (1x) + 2 μ M Forskolin.

The location where the biopsies were taken needs to be clearly labeled on the specimen containers as well as on the phenotypic checklist, accompanying the specimen.

3. 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.
4. For patients whose blood has never been tested, please also submit 10 milliliters of **whole blood** collected in EDTA (purple top) tubes.
5. The **tumor tissue** must be kept **at room temperature** at all times. We need to culture Schwann cells from the tumors for the analyses and hence, **living, viable cells are needed**. The tumor must be sent to the Medical Genomics Laboratory **using priority service shipment** (next day delivery). Please ALWAYS inform us (medgenomics@uab.edu or 205-934-5562) prior to shipping the biopsy, provide tracking number of the shipment + company and include a form for customs, if appropriate.
6. Address:
Medical Genomics Laboratory
Attn. Dr. Ludwine Messiaen
330 KAUL Human Genetics Building
720 20th Street S
Birmingham, AL 35294-0024