

Collection/storage of a skin biopsy for further NF1-related genetic studies.

1. 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.
2. 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.
3. Please also include **buccal swab samples** from the patient. Instructions and sterile swabs will be included with the biopsy collection kit.
4. The skin biopsy should 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 should be sent to the Medical Genomics Laboratory **using priority service shipment** (next day delivery). Please ALWAYS inform us (mgl@genetics.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.
5. Address:
Medical Genomics Laboratory
Attn. Dr. Ludwine Messiaen
330 KAUL Human Genetics Building
720 20th Street S
Birmingham, AL 35294-0024
6. 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 data at mgl@genetics.uab.edu and we will ship a package with the appropriate medium. 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 one week of receipt.