
Lab Dept: **Anatomic Pathology**

Test Name: **NEUROFIBROMATOSIS TYPE 1 (NF1)
SEQUENCING (INCLUDES SPRED1)**

General Information

Lab Order Codes: NFMA

Synonyms: Von Recklinghausen disease; Legius Syndrome (SPRED1)

CPT Codes: Combinations of the following codes will be billed based on findings:
81405 – SPRED1 Sequencing
81408 – NF1 Sequencing
81479 – NF1 Deletion/Duplication
81479 – SPRED1 Deletion/Duplication

Test Includes: Sequencing and Deletion/Duplication of the NF1/SPRED1 genes

Logistics

Test Indications: Patients with multiple CALMs w/wo skinfold freckling and no other typical NF1 associated features (Lisch nodules, bone abnormalities, neurofibromas, optic pathway gliomas).

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: University of Alabama Medical Genomic Laboratory (NFSP-NG)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours (See [Special instructions](#))

Turnaround Time: 25 working days

Special Instructions: A [completed requisition form](#) and informed consent with a phenotypic checklist must accompany each sample. For questions regarding the forms, please call 1-800-499-4363.

Monday – Thursday collections are preferred. Samples collected on Friday before 1400 can be shipped for Saturday delivery with special arrangements. Friday after 1400, Saturday/Sunday and collections, will be held in the lab and shipped on Monday or next business day.

NOTE: Detailed and accurate completion of this document is necessary for reporting purposes. The Medical Genomics Laboratory issues its clinical

reports based on the demographic data provided by the referring institution on the lab requisition form. It is the responsibility of the referring institution to provide accurate information. If an amended report is necessary due to inaccurate or illegible documentation, additional reports will be drafted with charge.

Specimen

Specimen Type: Whole blood

Container: Lavender (EDTA) top tube

Draw Volume: 6 mL EDTA (Minimum pediatric: 3 mL) whole blood

Processed Volume: Same as Draw Volume

Collection: Routine venipuncture. Invert sample gently to mix. Forward immediately to the lab.

Special Processing: Lab Staff:

1. **Do Not** centrifuge. Send whole blood at room temperature.
2. **DO NOT SHIP ON ICE.**
3. Include completed forms and requisition.
4. Be sure the shipping air bill is marked "Priority", Domestic.
5. Specimens must be packaged to prevent breakage and absorbent material must be included in the package to absorb liquids in the event that breakage occurs. Also, the package must be shipped in double watertight containers

Shipping:

Monday- Thursday, ship specimen as priority with proper forms, at ambient temperature via overnight courier.

Friday before 1400 specimens can be shipped at ambient temperatures for Saturday delivery. Call the University of Alabama Genomics lab (205-934-5562) for special instructions.

Friday after 1400, Saturday or Sunday and holidays specimens should be held in the lab at ambient temperatures and shipped ambient on Monday or the next business day (Monday-Thurs)

Note: Blood collections are stable for 1 week after collection.

Patient Preparation: None

Sample Rejection: Requests for Molecular Genetic testing for NF1 will not be accepted for the following reasons: No label (patients full name and date of collection) on the specimens; No referring physician's or genetic counselor's names and addresses; No billing information; No Phenotypic checklist form; Mislabeled or unlabeled specimens; Incorrect specimen type; Specimen frozen; Mislabeled specimens

Interpretive

Reference Range: No mutations detected

Critical Values: N/A

Limitations: N/A

Methodology: Next Generation Sequencing and Deletion/Duplication

References: [University of Alabama Medical Genomics Laboratory](#) June 2018
(800) 499-4363 Fax (205) 996-2929

Updates: 1/11/2017: Update CPT coding, volume update.
6/13/2018: Updated CPT, collection and shipping information.