
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:
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: Sunday – Thursday

Turnaround Time: 5 – 6 weeks

Special Instructions: Restricted draw times, see Test availability. 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.

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: Do Not centrifuge. Specimen must remain in original collection container. Ship specimen ASAP, with proper forms, at ambient temperature via overnight courier. Blood must reach the U of Alabama within 60 – 72 hours of draw.
Patient Preparation:	None
Sample Rejection:	Samples greater than 72 hours old, mislabeled or unlabeled specimens, specimens missing paperwork

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 January 2017 (800) 499-4363 Fax (205) 996-2929
Updates:	1/11/2017: Update CPT coding, volume update.