Lab Dept: Anatomic Pathology

Test Name: NEUROFIBROMATOSIS TYPE 1-LIKE (SPRED1) KNOWN MUTATION

General Information

Lab Order Codes:	SPREK
Synonyms:	Legius syndrome; Neurofibromatosis Type 1-like testing; SPRED1 Testing, NFLS
CPT Codes:	81403 – Molecular pathology procedure, Level 4 (analysis of single exon by DNA sequence analysis, analysis of >10 amplicons using multiplex PCR in 2 or more independent reactions, mutation scanning or duplication/deletion variants of 2-5 exons)
Test Includes:	A targeted mutation of a previously characterized SPRED1 mutation within the family.

Logistics

Test Indications:	Testing is for relative of patients with a known SPRED1 mutation.
Lab Testing Sections:	Anatomic Pathology - Sendouts
Referred to:	University of Alabama Medical Genomics (UAL Test: KT2)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	15 working days
Special Instructions:	Requests must include request form with referring or genetic counselor's name and address, billing information, completed informed consent and **phenotypic checklist**. All forms can be found at Forms. Targeted testing to all relevant relatives of a proband in whom a novel missense variant was identified is performed free of charge. Free of charge targeted testing will only be provided if the necessary phenotypic information on the proband and relatives filled out by a healthcare professional accompanies the samples. If no phenotypic information is provided, there will be charges to the test. Samples collected on Friday before 1400 can be shipped for Saturday delivery with special arrangements. Friday after 1400, Saturday/Sunday and

holiday collections, will be held in the lab and shipped on Monday, or next business day.
NOTE: Detailed and accurate completion of the requisition 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 top (EDTA) tube

Draw Volume: 6 mL (Minimum: 3 mL) whole blood **must be** in EDTA (Lavender) tubes

Processed Volume: Same as Draw Volume

Collection: Routine blood collection

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-Thursday).

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

Patient Preparation: None

Sample Rejection: Requests for Molecular Genetic testing 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:	Interpretive report
Critical Values:	N/A
Limitations:	Analysis of 86 unrelated individuals who presented with multiple CAL-spots only with or without a family history of CAL-spots, revealed a mutation in 7 of them (~ 8%). All were minor-lesion mutations (nonsense, frameshift and 1 missense mutation); no dosage alterations (total gene deletion or one/multi-exon copy number change) were found in Brems et al, but we have since identified them in our cohort of patients (Messiaen L, unpublished results).
Methodology:	A targeted mutation of a previously characterized SPRED1 mutation within the family. Targeted testing involves direct sequencing of a specific region or copy number analysis by MLPA and quantitative PCR.
References:	University of Alabama Medical Genomics Laboratory December 2023
Updates:	6/13/2018: Updated collection and shipping information 12/18/2023: Updated turnaround time.