
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 – SPRED1 Known family variant, DNA sequence analysis, each variant exon (Molecular Pathology Level 4)

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: 10 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 [Spred1Req form.pdf](#). 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.

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 holiday 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 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: <ol style="list-style-type: none">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;

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](#) June 2018
(205) 934-5562 Fax (205) 996-2929

Updates: 6/13/2018: Updated collection and shipping information