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 workind 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.

Specimen

Specimen Type:	Whole blood

Container:	Lavender top (EDTA) tube
Draw Volume:	3 – 6 mL
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection
Special Processing:	Lab Staff: Do Not centrifuge. Specimen should remain in original collection container. Specimens can be stored at refrigerated temperatures if collected Friday-Sunday. Send Monday-Thursday via UPS or Federal Express marked “priority”. 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.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens; frozen specimens, contaminated specimens, absence of referring physician and address, absence of billing information, absence of informed consent, absence of phenotypic checklist

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 March 2018 (205) 934-5562 Fax (205) 996-2929