

FOR LAB USE ONLY
ACCESSION NO:
DATE & TIME RECEIVED:
TECHNICIAN:

**UAB MEDICAL GENOMICS LABORATORY**  
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[www.genetics.uab.edu/medgenomics](http://www.genetics.uab.edu/medgenomics)

**SPRED1 (NF1-like or Legius syndrome) TEST REQUEST FORM**

**THIS FORM AND PHENOTYPIC CHECKLIST MUST BE FILLED OUT COMPLETELY**

PATIENT NAME:		BIRTH DATE:	DAYTIME PHONE:	SEX:	SOC. SEC. NUMBER:
PATIENT'S ADDRESS:		CITY:	STATE:	ZIP CODE:	MED REC NUMBER:
EMAIL ADDRESS:			PARENT OR GUARDIANS NAME (IF MINOR):		
<b>SPRED1 MUTATION ANALYSIS TESTING</b>			Physician's Name:		
<input type="checkbox"/> Comprehensive Test (Test 1- Sequencing and Copy Number Analysis) <u>after</u> a negative <i>NF1</i> test <input type="checkbox"/> Comprehensive Test (Test 1- Sequencing and Copy Number Analysis)- <i>NF1</i> testing <u>not</u> previously done <input type="checkbox"/> Copy Number Analysis by MLPA <i>only</i> <input type="checkbox"/> Targeted Mutation Analysis (Test 2) Proband _____			Physician's Address:		
Facility where specimen obtained:			Phone:		
Date:			NPI Number:		
<b>REQUIRED</b> Diagnosis (ICD-9) Code (only in US):			Fax:		
Is Patient Pregnant?		Please check if applicable:		Mailing Address:	
<input type="checkbox"/> No <input type="checkbox"/> Yes If yes, date of LMP: _____		<input type="checkbox"/> Infectious diseases (AIDS, Hepatitis, etc)		_____	
Specimen type:			Phone No:		
<input type="checkbox"/> Cheek Swabs; # Swabs: _____			Fax No:		
<input type="checkbox"/> Peripheral Blood (EDTA); # Tubes: _____			<input type="checkbox"/> Other: _____		

**BILLING INFORMATION**

BILL INSTITUTION: (Please, provide name, address, and telephone number of entity responsible for payment)

Purchase Order Number: _____	Contact Name: _____
Billing address: _____	Phone #: _____
	Fax #: _____
	Email: _____

PAYMENT ENCLOSED:

- Cashier's Check
- VISA®  MasterCard®  Discover®  American Express®

Card Number: _____	Expiration Date: _____
Name as it appears on card: _____	3-digit Security Code: _____
Cardholder Signature: _____	
Cardholder Email Address: _____	

BILL CONTRACTED INSURANCE COMPANY:

Please include a copy of patient's insurance card. For a list of our contracted insurance companies, please visit our website at [www.genetics.uab.edu/medgenomics](http://www.genetics.uab.edu/medgenomics), under "Billing". Please include a copy of pre-approval statement if payment has been authorized. **We also need the patient's credit card information so that any balance left after insurance pays may be applied to it.**

FILE INSURANCE CLAIM WITH NON CONTRACTED COMPANY

Patient must pay full payment for test up front, (credit card or cashier's check) but UAB will file a claim for reimbursement with the patient's insurance company. Please send a copy of patient's insurance card, front and back.

**Informed Consent for SPRED1 Testing (NF1-like syndrome)**

I, \_\_\_\_\_, hereby agree to participate in testing for Neurofibromatosis type 1-like syndrome using a DNA-based test. I understand that biological samples (blood, cheek cells) will be removed from me using standard techniques which carry very little risk. In addition, if prenatal diagnosis is being performed, fetal cells obtained by chorionic villus sampling or amniocentesis will be used. I understand that the blood, cheek cells or fetal samples will be used for the purpose of attempting to determine if I and/or members of my family are carriers of the disease gene. In addition I hereby give permission to collect biological samples from my minor children, named below, to be used for RNA/DNA-testing for the disease listed above.

Child's name	Date of Birth	Gender (F/M)

I understand that:

1. When direct DNA analysis is performed and a truncating mutation is identified, the test is >99% accurate. Rare variations in the DNA of individuals can sometimes be found and can cause uncertainty in predicting the carrier status. Thus the test is not 100% accurate and the results will be reported as a probability.
2. In other cases, the DNA test is unable to identify an abnormality although the abnormality may still exist. This event may be due to our current lack of knowledge of the complete gene structure or an inability of the current technology to identify certain types of mutations in the gene. The mutation detection system employed by the Medical Genomics laboratory for identifying SPRED1 mutations is the most sensitive developed at this point in time. I have been informed of the likelihood of finding a mutation in the gene for which I am being tested. \_\_\_\_\_(Initials)
3. I understand that the SPRED1 DNA analysis performed by the Medical Genomics Laboratory is specific for this disease and in no way guarantees my health or the health of my living or unborn children. The Medical Genomics Laboratory cannot be responsible for erroneous clinical diagnosis made elsewhere.
4. This test is relatively new and is being expanded and improved continuously. The test is not considered research, but is considered the best and newest laboratory service that can be offered. This testing is complex and utilizes specialized materials so that there is always some very small possibility that the test will not work properly or that an error will occur. There is a low error rate (perhaps 1 in 1000 samples) even in the best laboratories. My signature below acknowledges my voluntary participation in this test, but in no way releases the laboratory and staff from the Medical Genomics Laboratory from their professional and ethical responsibility to me.
5. I understand that my sample is not being banked. The laboratory does not return DNA samples to individuals or physicians. However, in some cases it may be possible for the laboratory to reanalyze my remaining DNA upon request. The request for additional studies must be ordered by my referring physician/counselor and there will be an additional fee.
6. Once my test result is completed, an aliquot of my DNA may be made anonymous (name and all other identifiers removed) and used for research purposes. Any results obtained could not be related to the original source, so no results would be reported. I understand declining to participate in this research will in no way negatively impact my clinical testing or treatment.
  - By checking this box, I indicate my desire to opt out of participation in anonymized research studies using my DNA sample.
7. Because of the complexity of DNA based testing and the important implications of the test results, results will only be reported to me through a physician, genetic counselor or certified genetics professional. The result reports are confidential and will only be released to other medical professionals or other parties with my express written consent. All laboratory data and personal information are strictly confidential and will not be released from the Medical Genomics Laboratory in full compliance with HIPPA. Participation in DNA testing is completely voluntary.
8. I will receive a copy of this consent form.

Signature: \_\_\_\_\_  
 Witnessed by: \_\_\_\_\_  
 Date: \_\_\_\_\_

Physician's/Counselor's statement: I have explained RNA/DNA testing to this individual. I have addressed the limitations outlined above and have answered person's questions.

Signature: \_\_\_\_\_

**MEDICAL GENOMICS LABORATORY: NF1/SPRED1 PHENOTYPIC CHECKLIST FORM**



Patient ID: \_\_\_\_\_

Referring Physician: \_\_\_\_\_ Date of Exam \_\_\_/\_\_\_/\_\_\_

**DEMOGRAPHIC INFORMATION**

Gender :  Male  Female

Date of Birth: \_\_\_/\_\_\_/\_\_\_

Ethnicity: Mother:  White  Black  Native American  Hispanic  Asian  Other:  
Father:  White  Black  Native American  Hispanic  Asian  Other:

**DIAGNOSIS**

NIH criteria:  >6 CAL spots >5mm, postpubertal >15mm  Optic glioma  
 >2 neurofibromas or 1 plexiform NF  >2 Lisch nodules  
 Axillary or inguinal freckling  A distinct osseous lesion  
 First degree relative diagnosed with NF1 by above criteria  
Does patient fulfill NIH diagnostic criteria for NF1?  Yes  No

Clinical diagnosis:  NF1  Multiple CAL spots  Familial multiple CAL spots  
 Spinal NF  Isolated neurofibromas  Segmental NF1  
 NF Noonan  Single NF1 feature  Watson Syndrome  
 Unknown

Family history:  Sporadic  Familial  Unknown Consanguinity:  Yes  No  Unknown

**Familial cases:** Please provide pedigree and details on the affection status of family members on a separate page

**GENERAL INFORMATION**

Height: \_\_\_cm Head circumference: \_\_\_cm Weight: \_\_\_kg

**NF SIGNS AND SYMPTOMS**

1) CAL spots:  0  1-5  ≥6 to 100  >100  
General impression on the borders of the CAL-spots:  
 typical well-defined smooth borders diameter:  
 irregular margins, ragged borders diameter:  
Please provide detail on size and location of the CAL-spots and other hyper/hypopigmentation areas on the figure provided on page 3. A digital picture of the skin findings would be very helpful.

2) Skin fold freckling:  None Left Right Comments (e.g. very faint,.....):  
Groin    
Axilla    
Submammary

3) Lisch nodules:  None  Left  Right  Unknown

4) Cutaneous neurofibromas (soft nodules that project above the skin): histopathologically confirmed: Y / N  
 0  2-6  6-99  100-500  >500

5) Intradermal neurofibromas (soft depression within the skin with pink/purple overlying discoloration):  
 0  2-6  6-99  100-500  >500  
histopathologically confirmed: Y / N

6) Subdermal neurofibromas (firm nodules palpable underneath the skin):  
 0  2-6  6-99  100-500  >500  
histopathologically confirmed: Y / N

MEDICAL GENOMICS LABORATORY: NF1/SPRED1 PHENOTYPIC CHECKLIST FORM



- 7) Plexiform neurofibromas:  None  visible from outside  with hyperpigmentation  
 internal  without hyperpigmentation
- Head  Neck  Trunk  L Arm  L Hand  L Leg  L Foot  
 Abdomen  Pelvis  Genital Region  R Arm  R Hand  R Leg  R Foot
- histopathologically confirmed: Y / N

- 8) Spinal neurofibromas (neurofibromas arising from the spinal nerve root) :  
 Unknown  Absent by MRI  Present, asymptomatic  
 Present, symptomatic
- If present: please provide detail on **figure** page 4  
 unilateral or  bilateral;  
 C\_\_\_\_\_ T\_\_\_\_\_, L\_\_\_\_\_, S\_\_\_\_\_ regions.  
 histopathologically confirmed: Y / N

- 9) Optic glioma:  Unknown  Absent by MRI  
 Present by MRI, symptomatic  
 Nerve (L and/or R)  
 Chiasm  
 Present by MRI, asymptomatic  
 Nerve (L and/or R)  
 Chiasm

- 10) Other neoplasms:  None  Brainstem glioma  Other glioma  
 Hypothalamic glioma  JMML  Rhabdomyosarcoma  
 MPNST  Colonic polyps  Lipoma  
 Pheochromocytoma  
 Other, specify: \_\_\_\_\_

- 11) Skeletal Abnormalities:  None  Pseudoarthrosis  Sphenoid wing dysplasia  
 Long bone dysplasia  scoliosis  Dysplastic vertebrae  
 Bone cysts  Other: \_\_\_\_\_  
 pectus excavatum

- 12) Cardiovascular disease:  Absent  Unknown  
 Present:  Hypertension  Aortic stenosis  Renal artery stenosis  
 moya moya  Other \_\_\_\_\_

- 13) Development:  Normal  Abnormal  Exam not done  ADD  Hyperactivity  Learning disability  
 IQ: Full scale \_\_\_\_\_, Verbal \_\_\_\_\_, Performance \_\_\_\_\_.

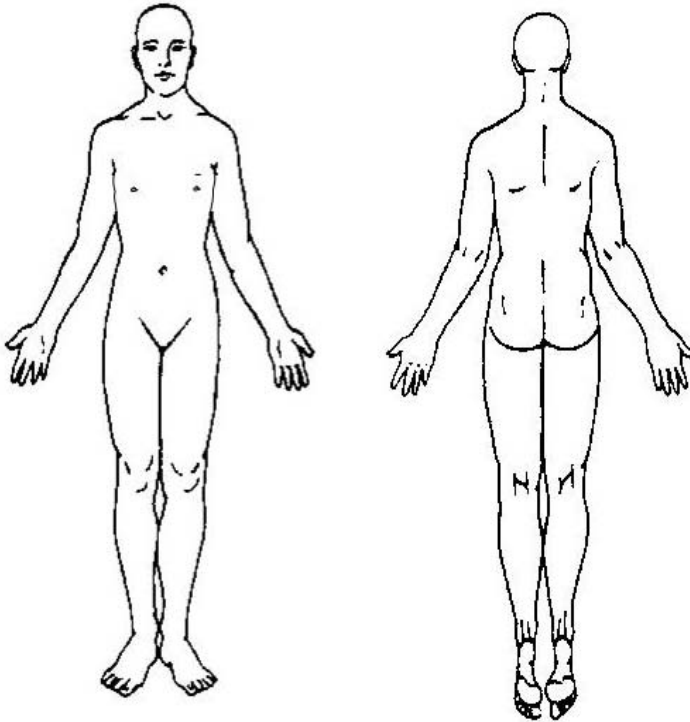
- 14) Education:  Too young for school  At or above age level  Below age level  
 HS completion  College graduate  Higher degree  Unknown

- 15) Noonan phenotype:  Absent  Possible  Unknown  
 Present:  Short stature  Low set ears  Midface hypoplasia  
 Hypertelorism  Webbed neck  Pulmonic Stenosis


- 16) Segmental NF phenotype:  Absent  Possible  
 Please provide detail on size and localization of neurofibromas and/or CAL-spots and/or freckling and/or hyperpigmented region using the **figure** on page 3.




location/size of pigmentary lesions and/or neurofibromas ↓




Indicate size and location of

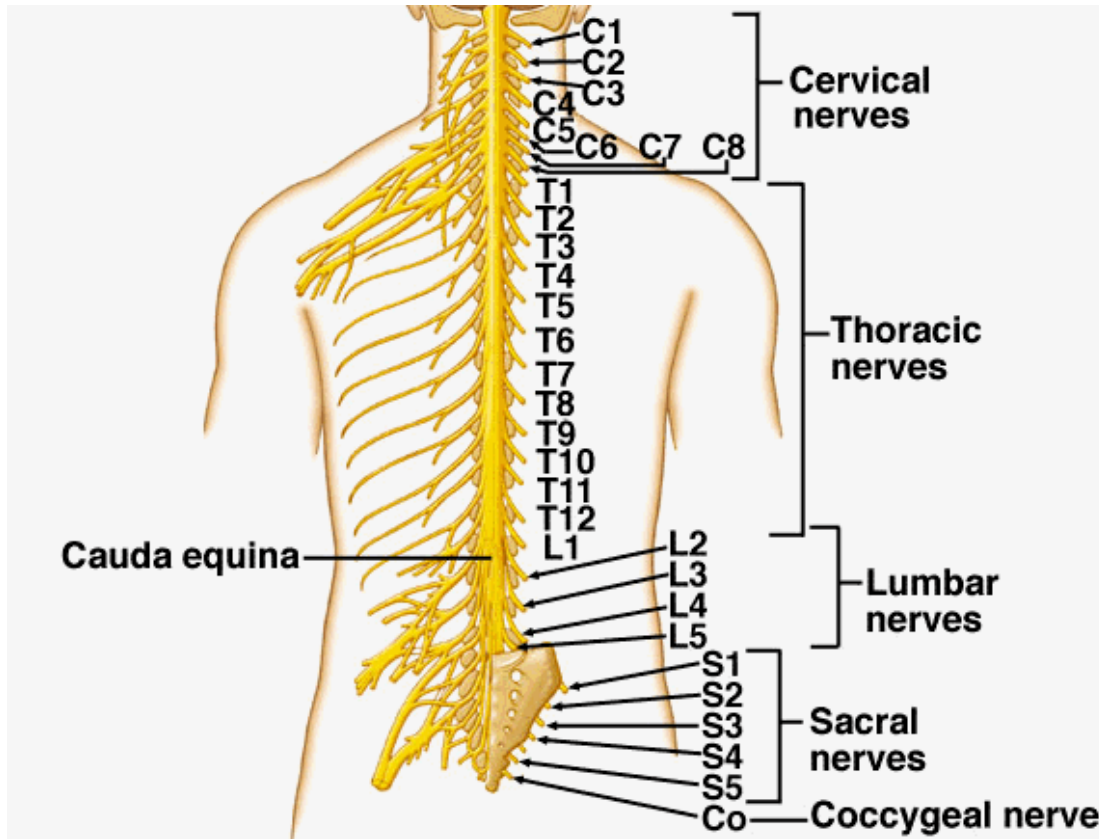
Neurofibromas 

CAL-spots 

Freckling 

Hyperpigmented region 

Detail on location of spinal tumors ↓



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