

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

UAB MEDICAL GENOMICS LABORATORY
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NEUROFIBROMATOSIS TYPE 1 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):		
NF1 MUTATION ANALYSIS TESTING		Physician's Name:		
<input type="checkbox"/> Comprehensive NF1 Test (Test 1) <input type="checkbox"/> RUSH? <input type="checkbox"/> Add SPRED1 testing when NF1 testing is negative <input type="checkbox"/> Copy Number Analysis by MLPA <i>only</i> <input type="checkbox"/> Targeted Mutation Analysis (Test 2) Proband _____ <input type="checkbox"/> Prenatal Targeted Analysis (Test 3) Proband _____ <input type="checkbox"/> Comprehensive test on biopsies (Test 4)		Physician's Address: _____ _____ _____		
Facility where specimen obtained:		Phone: _____ Fax: _____		
Date:		NPI Number: _____ Email: _____		
REQUIRED Diagnosis (ICD-9) Code (only in US):		ADDITIONAL REPORTS TO:		
Is Patient Pregnant? <input type="checkbox"/> No <input type="checkbox"/> Yes If yes, date of LMP: _____		Name: _____		
Please check if applicable: <input type="checkbox"/> Infectious diseases (AIDS, Hepatitis, etc)		Mailing Address: _____ _____		
Specimen type:		Phone No: _____ Fax No: _____		
<input type="checkbox"/> Cheek Swabs; # Swabs: <input type="checkbox"/> Peripheral Blood (EDTA); # Tubes:		<input type="checkbox"/> Amniotic Fluid <input type="checkbox"/> Direct CVS <input type="checkbox"/> Cultured CVS <input type="checkbox"/> Cultured Amniocytes		
		<input type="checkbox"/> biopsy CAL-spot: # biopsies: <input type="checkbox"/> biopsy neurofibroma: # biopsies: <input type="checkbox"/> Other biopsies: <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, 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. The RUSH fee must be paid up front by the patient.**

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. **The RUSH fee must be paid up front by the patient.**

Informed Consent for NF 1 Testing

I, _____, hereby agree to participate in testing for Neurofibromatosis type 1 using a RNA/DNA-based cascade of tests. 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. In >95% of the NF1 patients fulfilling the NIH diagnostic criteria, the RNA/DNA cascade of tests detects an abnormality, called a mutation, in the NF1 gene, and 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.
2. In other cases, the RNA-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 NF1 mutations is the most sensitive yet developed. 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 RNA/DNA NF1 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. In order to perform accurate prenatal diagnosis, biological samples are required from the fetus as well as from the affected individual in the family and from the biological mother.
5. 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.
6. 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.
7. A. Once my test result is completed, an aliquot of my DNA/RNA 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.
7. B. I indicate my desire to opt out of participation in anonymized research studies using my DNA/RNA sample by checking this box
8. Because of the complexity of RNA/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 are confidential and will not be released from the Medical Genomics Laboratory. Participation in RNA/DNA testing is completely voluntary.
9. 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	<input type="checkbox"/>	<input type="checkbox"/>	
Axilla	<input type="checkbox"/>	<input type="checkbox"/>	
Submammary	<input type="checkbox"/>	<input type="checkbox"/>	

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 Hypothalamic glioma Brainstem glioma Other glioma
 MPNST JMML Rhabdomyosarcoma
 Pheochromocytoma Colonic polyps Lipoma
 Other, specify: _____

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

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 _____.

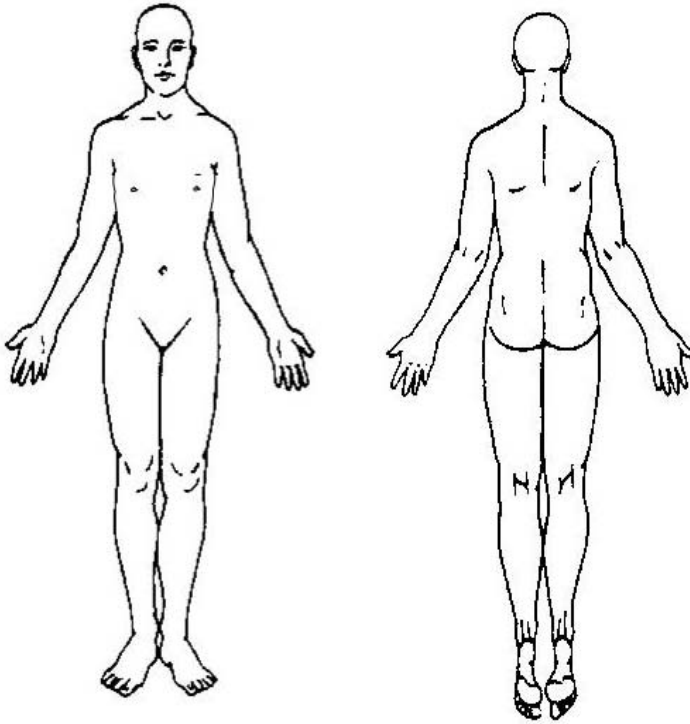
14) Education: Too young for school At or above age level Below age level
 HS completion College graduate Higher degree Unknown

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

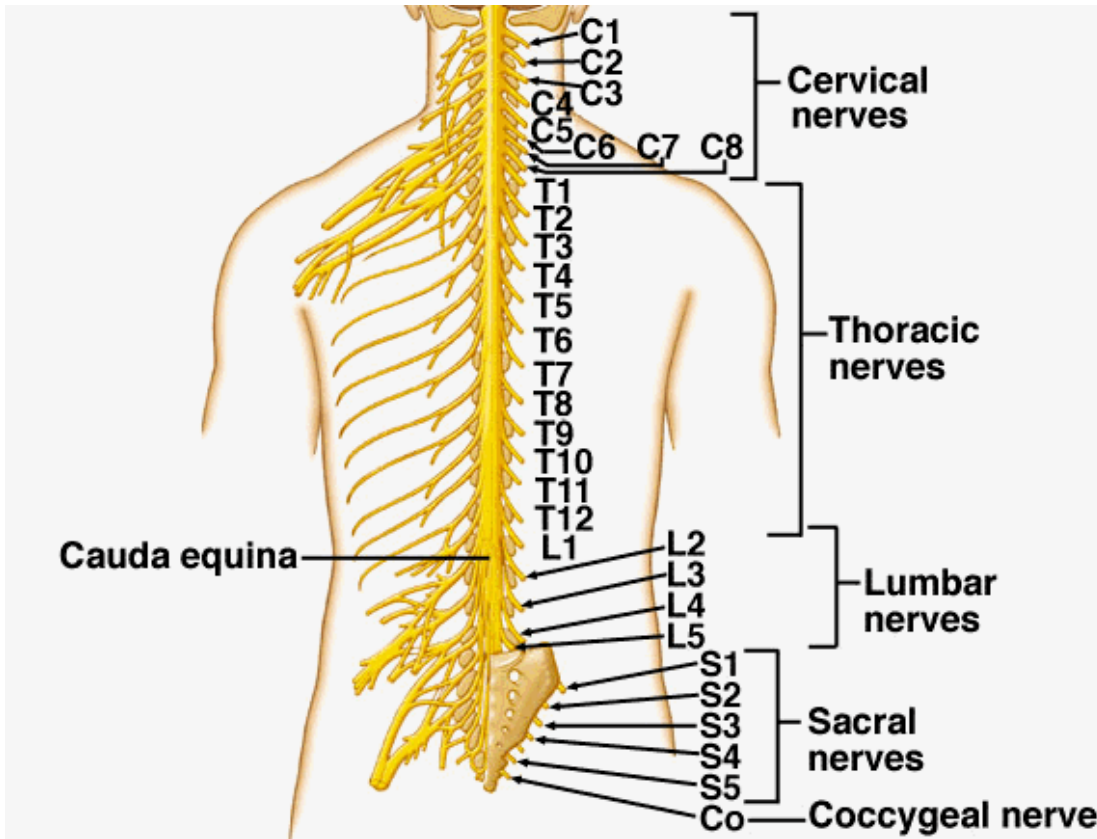
Neurofibromas 

CAL-spots 

Freckling 

Hyperpigmented region 

Detail on location of spinal tumors ↓



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