UAB MEDICAL GENOMICS LABORATORY
720 South Twentieth Street, Suite 330
Birmingham, Alabama 35294-0005
www.genetics.uab.edu/medgenomics
Tel: (205) 934-5562
Fax: (205) 996-2929

Accession:

For MGL Laboratory Use only

Test Requisiton Form: Accurate completion of this document is necessary for reporting purposes (see full policy on website)

Patient Information:

Patient Name: (First)	(MI)	(Last)	
MRN:		DOB (mm/dd/yyy):	
Gender:	Social Security#:		
Address:			
City:	State:	Zip:	
Phone:	Email:		
Parent or Guardian name (if minor):			

Please place additional patient identification stickers here

Ordering Physician:

☐**Please check if physician should receive report directly**		
Name:		NPI:
Address:		
City:	State:	Zip:
Institution:	Email:	
Phone:	Fax:	

Additional Reports to:

Name:		
Address:		
City:	State:	Zip:
Institution:	Email:	
Phone:	Fax:	

For MGL Lab Use only:

	Initials:	Date:	Comment:
Received:			
Reviewed:			
Accession:			
Billing:			
Other:			

Lab/Hospital Information:

☐**Please check if Lab/Hosptial should receive report directly**		
Name:		
Address:		
City:	State:	Zip:
Phone:	Fax:	

Billing Information:

Please Note: Out of State Medicaid is not accepted under any circumstances

It is the referring physician's responsibility to discuss pricing and billing with the patient. Full information on the billing policies is available at **www.genetics.uab.edu/medgenomics**. Credit card information MUST be provided at time of sample submission for insurance and self pay clients.

By completing this form, you agree that you have discussed the MGL's billing policies with your patient. As insurance prices are not listed on the internet, please call the billing coordinator at 205-934-5523 to request a quote, if needed, and pass this information along to the client.

☐ Institutional Bill:

☐Please check if billing institution should receive report

Institution:		
Address:		
City:	State:	Zip:
Contact:	Email:	
Phone:	Fax:	

☐ Payment Enclosed:

☐Cashier's Check ☐Visa ☐MasterCard ☐Discover ☐American Express

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

☐Bill Contracted Insurance Company

Please include a copy of patient's insurance card, front and back. For a list of contracted insurance companies, please visit website at www.genetics.uab.edu/medgenomics. Please include pre-approval statement if payment has been authorized. **RUSH fee must be paid up front**

☐File Insurance Claim with Non-Contracted Company

Patient must pay full payment for test up front via credit card or cashier's check. UAB will file a claim for reimbursement with the patient's insurance company. Please send a copy of the patient's insurance card, front and back. The RUSH fee must be paid up front by the patient. Credit card information must be provided at the same time as

Informed Consent:

Provider's statement: I acknowledge the risks, benefits, limitations, and implications of genetic testing as outlined on the complete informed consent handout; and I have discussed the test(s) requested with the patient/guardian and I have answered his/her questions regarding testing. Informed consent has been obtained from the patient/guardian and the hard copy will be maintained.

Provider's Signature: _____.

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Patient Name: (First) (MI) (Last)

DOB:

Test Request Information

If multiple tests are requested, please specify order in which testing should be performed.

□ Please Check if RUSH Testing is Requested

-RUSH testing is avaliable for: NFSP1, NF11, NF21, SB11, and PKDS

Please call prior to ordering tests marked with (*)

Order	Code	NF1 and Legius
	NFSP1	NF1/SPRED1 Seq and Del/Dup on Blood
	NF11	NF1 Seq and Del/Dup
	NF14C	NF1/SPRED1 Seq and Del/Dup on CALs*
	NF14N	NF1 seq and Del/Dup on neurofibromas*
	SPD1	SPRED1 Seq and Del/Dup on Blood

Order	Code	Neurofibromatosis Type 2
	NF21	NF2 Seq and Del/Dup on Blood
	NF24	NF2 Seq and Del/Dup on Tumor

Order	Code	Schwannomatosis and RT
	SB11	SMARCB1 Seq and Del/Dup on Blood
	SB14RT	SMARCB1 Seq and Del/Dup on RT
	SB14Sch	SMARCB1 Seq and Del/Dup on Tumor

Order	Code	PTEN Related Disorders
	PTEN1	PTEN Seq and Del/Dup on Blood
	PTENS	PTEN Sequencing only
	PTENM	PTEN Del/Dup only

Order	Code	MCADD
	MCD1	ACADM Sequencing
	MCD2	ACADM Exon 11 only

Order	Code	Von Hippel Lindau
	VHL1	VHL Seq and Del/Dup on Blood

Order	Code	Rasopathy Testing
	NNP	Noonan Panel: 12 genes
	CFCP	CFC Panel: 4 genes
	LPDP	LEOPARD Panel: 3 genes
	CST1	Costello (HRAS sequencing)
	MTM1	Metachondromatosis (PTPN11 seq)

Order	Code	Single Rasopathy Genes
	SGS1	Single Gene Sequencing*

Disorder suspected:

Indicate Gene Desired:

□ PTPN11 □ SOS1 □ RAF1 □ HRAS
□ KRAS □ NRAS □ SHOC2 □ SPRED1
□ CBL □ BRAF □ MAP2K1 □ MAP2K2

Order	Code	Autosomal Recessive PKD
	PKD1	PKHD1 Seq and Del/Dup
	PKDS	PKHD1 Seq only
	PKDL	Linkage Analysis
	PKDPL	Prenatal Linkage

Gene	Code	Known Mutation Testing	
	KT2	Targeted	***Complete Previous Testing History***
	PT2	Prenatal	

Patient History

Specify indications for testing (ICD-9 codes required):

Please check if applicable: □ Infectious diseases (AIDS, Hepatitis, etc)

Has this patient received a bone marrow transplant? □ Yes □ No

Has this patient had chemotherapy in the past 6 months? □ Yes □ No

Is the patient pregnant? □ Yes, LMP: □ No

Previous Testing History

Has this patient or relatives had previous testing? □ Yes □ No

Name/Relationship to patient:

Test/Mutation/Lab:

Name/Relationship to patient:

Test/Mutation/Lab:

Specimen Type

Specimen requirements vary based on test requested; please see www.genetics.uab.edu/medgenomics for more details

Comprehensive Testing:

Date collected:

□ Peripheral Blood (EDTA); # Tubes:

□ Biopsy-CAL-spot (specify location on checklist); # biopsies:

□ Biopsy-neurofibroma (specify location on checklist); # biopsies:

□ Tumor (specify location on checklist):

Pathology: □ *Frozen* □ *Fresh* □ *Paraffin*

□ Extracted DNA (not preferred specimen for NF1); Source:

□ Other, please describe:

Targeted Mutation Analysis

Date collected:

□ Peripheral Blood (EDTA); # Tubes:

□ Extracted DNA | □ Cheek Swabs; # Swabs:

□ Other, please describe:

Prenatal Testing

□ Amniotic Fluid | □ Direct CVS (cleaned)

□ Cultured Amniocytes | □ Cultured villus cells

Back-up Culture Facility (required):

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Informed Consent for Genetic Testing

****This form does not need to be returned to the Medical Genomics Laboratory if Informed Consent portion of the Test Request form has been signed.*****

I hereby consent for:

Name:	DOB:	Gender:

To participate in genetic testing for the following RNA/DNA-based cascade of tests ordered by my physician at the University of Alabama at Birmingham (UAB) Medical Genomics Laboratory (MGL):

Genetic Tests:

I understand that:

1. Any biological samples submitted for genetic testing (e.g., blood, cheek cells, saliva, amniotic fluid, chorionic villi, tumor, and/or tissue) will be removed from me and/or my minor child(ren) using standard techniques which carry their associated risks.

2. Any samples obtained will be used for the purpose of attempting to determine if I and/or members of my family carry genetic changes in the disease genes ordered by my physician.

3. The genetic tests performed at the MGL are the most sensitive developed and are highly specific. However, sensitivity and specificity are test-dependent. Additional testing details and the specific detection rates of each test can be found at www.genetics.uab.edu/medgenomics.

4. The following are possible outcomes for the specific tests listed above:

Positive	Unknown Significance	Negative
This is an indication that I may be predisposed to or have the specific disease, or condition tested. Further testing may be needed to confirm the diagnosis.	There may be a possibility that the laboratory findings will be ambiguous or of unknown significance. This may require additional testing from me or my family members. In rare circumstances, findings may be suggestive of a condition different than the diagnosis that was originally made.	There is a chance that I will still have this genetic condition even though the genetic test results are negative. Due to limitations in technology and incomplete knowledge of genes, some changes in RNA/DNA or protein products that cause disease may not be detected by this test.

5. 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 incomplete knowledge of the gene structure or an inability of current technology to identify certain types of mutations in the gene. When clinically necessary, the MGL may use a method called linkage analysis. This method is not a direct test, but will report the probability that you and/or family members have an inherited disease or disorder. In some families, the markers used in linkage analysis may be uninformative. If so, linkage testing cannot provide results for the family members in question.

6. The RNA/DNA analysis performed by the MGL is specific for the genetic test listed above and in no way guarantees my health or the health of my living or unborn children. The MGL cannot be responsible for an erroneous clinic diagnosis made elsewhere.

7. The tests performed at the MGL are expanded and improved continuously. The tests offered are not considered research but are considered the best and newest laboratory service that can be offered. Genetic testing is complex and utilizes specialized materials so 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 acknowledges my voluntary participation in this test, but in no way releases the laboratory and staff from the MGL from their professional or ethical responsibility to me.

8. The MGL does not return DNA samples to individuals or physicians. While the MGL is not a specimen banking facility, 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.

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9. An aliquot of my DNA/RNA may be used for validation, educational and/or research purposes. For some molecular genetic tests, a synopsis of clinical information and test results may be included in HIPAA-compliant, de-identified public databases as part of the National Institute of Health's effort to improve diagnostic testing and our understanding of the relationships between genetic changes and clinical symptoms. If I would like to opt out of participation, I can contact the MGL via email at medgenomics@uab.edu or calling the laboratory at 205-934-5562.

10. Because of the complexity of genetic testing and the important implications of the test results, results will only be reported to me through a physician, genetic counselor, or a certified genetics professional. The results are confidential to the extent allowed by law. They will only be released to other medical professionals or other parties with my written consent or as otherwise allowed by law. Participation in testing is completely voluntary.

11. **For Prenatal Testing:** If prenatal diagnosis is being performed, fetal cells obtained by chorionic villus sampling or amniocentesis will be used. In order to perform accurate prenatal diagnosis, biological samples are required for the fetus as well as from the affected individual in the family and from the biological mother.

12. I and/or my physician/counselor have signed the informed consent portion of the test order form indicating that we have discussed the items on this document and I will also receive a copy of this consent form.

Subject's Signature	Date	Physician's Signature	Date
Please Print Subject's Name		Please Print Physician's Name	
Assent of Parent	Date	Genetic Counselor's Signature	Date
Assent of Child	Date	Please Print Genetic Counselor's Name	

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

7) Plexiform neurofibromas: ☐ None ☐ visible from outside ☐ with hyperpigmentation ☐ without hyperpigmentation ☐ internal

☐ 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 _____.

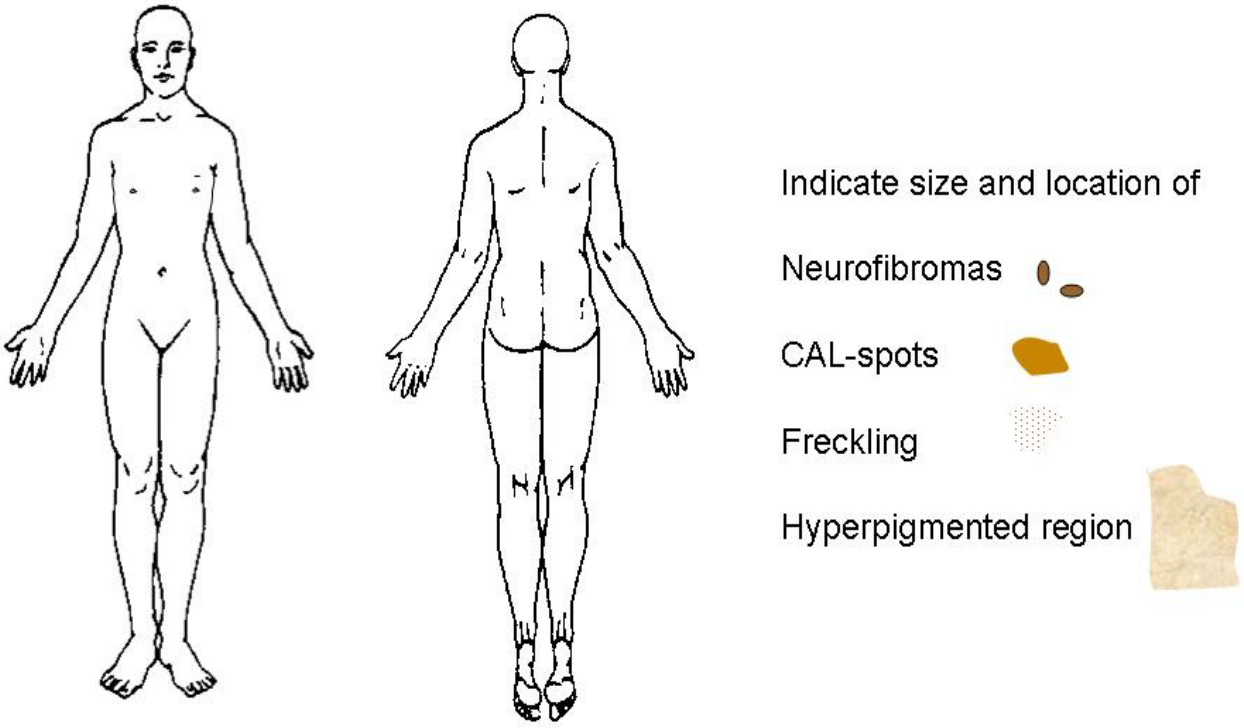
14) Education: ☐ Too young for school ☐ At or above age level ☐ Below age level ☐ HS completion ☐ College graduate ☐ Higher degree ☐ Unknown

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 ↓



Detail on location of spinal tumors ↓