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

UAB MGL
Accession

For MGL Lab Use Only		Received:	Reviewed:	Accession:	Billing:	Other:
	Initials:					
	Date:					
	Comment:					

Important Notes

-This form must accompany all specimens received
-Billing information (page 5) must be included
-All specimens received must include **two** patient identifiers and **collection date**
-Testing must be ordered by a qualified clinician
Additional information is available online at www.uab.edu/medicine/genetics/medical-genomics-laboratory

Test Requisition Form

Patient Information:

Sample Collected: (MM/DD/YY)

Name: (First) (MI) (Last)

DOB: (MM/DD/YY) | MRN:

Address:

City: | State: | Zip:

Gender: | SSN:

Parent or Guardian name (if minor):

Ordering Physician:

☐ Please check box if physician should receive report directly

Name: | NPI:

Institution:

Address:

City: | State: | Zip:

Country: | Phone:

Please check preferred result delivery: ☐ Fax:

☐ Email:

Referring Lab/Hospital:

☐ Please check box if lab/hospital should receive report directly

Name:

Institution:

Address:

City: | State: | Zip:

Country: | Phone:

Please check preferred result delivery: ☐ Fax:

☐ Email:

Additional Reports to:

Name:

Institution:

Address:

City: | State: | Zip:

Country: | Phone:

Please check preferred result delivery: ☐ Fax:

☐ Email:

Previous Testing History

Check all that apply:

☐ Patient or family member is pregnant. LMP:
☐ Patient has had a bone marrow transplant
☐ Patient has had chemotherapy in the past 6 months
☐ Infectious diseases (AIDS, Hepatitis, etc.)

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

Name/Relationship to patient: | Test/Variant/Lab:

Name/Relationship to patient: | Test/Variant/Lab:

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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UAB MGL Accession

Name: (First) (MI) (Last) **DOB:** (MM/DD/YY)

Lymphocyte/White Blood Cell-based Comprehensive Testing via Next-Gen Sequencing

☐ **RUSH Analysis:** Testing completed within **15 working days** of receipt of sample
(Additional **$600 RUSH fee** applied; only available for tests on this page)

NF1/Legius syndrome and Other RASopathy Related Conditions

☐ **NF1-NG: NGS** and **Del/Dup:** *NF1* only

☐ **NFSP-NG: NGS** and **Del/Dup:** *NF1* and *SPRED1*

☐ **NNP-NG: NGS:** 17 genes (no ***NF1***): *BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2,* and *SPRED1*; and **Del/Dup:** *SPRED1* and *LZTR1*

☐ **RAS-NG: NGS:** 18 genes: *BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2,* and *SPRED1*; and **Del/Dup:** *NF1, SPRED1,* and *LZTR1*

☐ **CST-NG: NGS:** *HRAS* only

NF2/Schwannomatosis/Meningiomatosis

☐ **NF2-NG: NGS** and **Del/Dup**: *NF2* only

☐ **SCH-NG: NGS:** 3 genes: *LZTR1, NF2,* and *SMARCB1*; and **Del/Dup**: *NF2, LZTR1,* and *SMARCB1*

☐ **MEN-NG: NGS:** 4 genes: *NF2, SMARCB1, SMARCE1,* and *SUFU*; and **Del/Dup**: *NF2* and *SMARCB1*

Peripheral Nerve Sheath Tumor Testing

☐ **PNT-NG: NGS:** 6 genes: *NF1, NF2, KRAS, LZTR1, PTPN11* and *SMARCB1*; and **Del/Dup**: *NF1, NF2, LZTR1, and SMARCB1*

McCune-Albright Syndrome

☐ **GNAS-NG: NGS**: *GNAS* exons 8 and 9 only

Rhabdoid Tumor Predisposition Syndrome

☐ **RT-NG: NGS:** *SMARCB1* and *SMARCA4*; and **Del/Dup**: *SMARCB1* only

Tuberous Sclerosis Complex

☐ **TSCP-NG: NGS** and **Del/Dup**: *TSC1* and *TSC2*

Capillary Malformation Arteriovenous Malformation Syndrome

☐ **RASA-NG: NGS:** and **Del/Dup**: *RASA1* and *EPHB4*

Additional Information

Test Description Key:
Next Generation Sequencing **(NGS)**
Deletion/Duplication analysis **(Del/Dup)**

For additional testing options via **tumor/biopsy**, please see **page 3** of this order form.
Please contact the lab via phone (205) 934-5562 or via email at medgenomics@uabmc.edu if you have any questions when completing this form.
For additional information, visit our website at www.uab.edu/medicine/genetics/medical-genomics-laboratory

Important points of consideration for testing

The average coverage for all of our panels is >1600x.Specifically for the *NF1* gene, the NGS approach covers >98% of the *NF1* coding region at ≥350X and 99% ≥200X, allowing detection of very low level mosaicism, down to 3-5% variant allele fraction respectively. For all other genes on our panels, the NGS approach covers an average of 99% at ≥200X. Remaining regions are covered at ≥30X. However, for patients with segmental/mosaic presentation, deep coverage in lymphocyte cells may be insufficient to identify the underlying gene change. Testing the affected tissue(s) may be necessary to confirm a diagnosis. Please see page 3 for our tumor/biopsy-based testing options.

Please note: For patients with an ongoing pregnancy who require comprehensive NF1 testing, "NF1-R" is recommended due to the sensitivity and fast turnaround time of this test (please see page 4 for this option).

Specimen Requirements

Accepted Specimens

Specimen requirements vary based on test requested; please see our website for more details.

-**Blood**: 3-6ml EDTA (receipt within one week of collection)
-**Saliva**: OGR-575 DNA Genotek (kits are provided upon request)
-**DNA**: extracted from lymphocyte cells, a minimum of 25ul at 3µg, O.D. value at 260:280nm ≥1.6 (must be extracted in a CLIA or equivalent certified lab)
-**Fibroblast cells**

Specimen Information:

□ Peripheral Blood (EDTA); # Tubes: _____

□ Extracted DNA; Source: _____

□ Saliva (kit must be provided by MGL)

□ Other, please describe: _____

Please note: failure to provide a date of collection can delay release of results

Sample Collected Date (required): _____

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UAB MGL Accession

Name: (First) (MI) (Last) **DOB:** (MM/DD/YY)

Tumor/Biopsy-based Comprehensive Testing

☐ Please check here if blood or DNA is provided for confirmation testing. Blood Collected: (MM/DD/YY)

NF1/SPRED1 on biopsied CALs and Neurofibromas

Please contact the laboratory at least one week in advance of the biopsy before ordering this test as media must be provided in advance and special shipping instructions apply. Biopsies must arrive *within 60 hours of collection***

☐ **NF14C: Sanger (RNA)** and **Del/Dup:** *NF1* (with automatic reflex to *SPRED1*) on biopsied CALs

☐ **NF14N: Sanger (RNA)** and **Del/Dup:** *NF1* on biopsied neurofibromas

NF2/Schwannomatosis

☐ **NF2-NG: Fresh/Frozen Tumor** for **NGS** and **Del/Dup:** *NF2* only

☐ **SCH-NG: Fresh/Frozen Tumor** for **NGS** and **Del/Dup**: *NF2, LZTR1,* and *SMARCB1*

☐ **NF24: Tumor Block** for **Sanger** and **Del/Dup**: *NF2* only

☐ **SCHP: Tumor Block** for **Sanger** and **Del/Dup**: *NF2, LZTR1,* and *SMARCB1*

Rhabdoid Tumor Predisposition Syndrome

☐ **RT-NG: Fresh/Frozen Tumor** for **NGS** *SMARCB1* and *SMARCA4*; and **Del/Dup**: *SMARCB1* only

☐ **SB14RT: Tumor Block** for **Sanger** and **Del/Dup**: *SMARCB1* only

RASopathy Related Conditions

☐ **NNP-NG: Fresh/Frozen Tumor** for **NGS** (no *NF1*): *BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2,* and *SPRED1*; and **Del/Dup:** *SPRED1* and *LZTR1*

☐ **RAS-NG: Fresh/Frozen Tumor** for **NGS**: *BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2,* and *SPRED1*; and **Del/Dup:** *NF1, SPRED1,* and *LZTR1*

Meningiomatosis

☐ **MEN-NG: Fresh/Frozen Tumor** for **NGS**: *NF2, SMARCB1, SMARCE1,* and *SUFU;* and **Del/Dup**: *NF2* and *SMARCB1*

Peripheral Nerve Sheath Tumor Testing

☐ **PNT-NG: Fresh/Frozen Tumor** for **NGS**: *NF1, NF2, KRAS, LZTR1, PTPN11* and *SMARCB1;* and **Del/Dup**: *NF1, NF2, LZTR1, and SMARCB1*

Tuberous Sclerosis Complex

☐ **TSC-NG: Fresh/Frozen Tumor** for **NGS** and **Del/Dup**: *TSC1* and *TSC2*

Additional Information

Test Description Key:
Next Generation Sequencing **(NGS)**
Sanger Sequencing **(Sanger)**
Deletion/Duplication analysis **(Del/Dup)**

Please contact the lab via phone (205) 934-5562 or via email at medgenomics@uabmc.edu if you have any questions when completing this form.
For additional information, visit our website at www.uab.edu/medicine/genetics/medical-genomics-laboratory

Important points of consideration for testing

- The MGL offers next generation sequencing testing options that provide the ability to identify variants (indels and substitutions) as low as 3% of the alleles, depending on coverage in the regions of interest.
- NF1/SPRED1 biopsy-based testing is considered the "gold standard" approach for confirming a diagnosis of mosaic/segmental NF1 or Legius Syndrome.
- A minimum of two biopsies is required for NF1 testing. Two or more tumors are suggested for our other testing options. There are no additional fees associated with testing on additional biopsy specimens.
- When proceeding with tumor-based testing for NF2, test code "SCH-NG" or "SCHP" (*NF2, SMARCB1, and LZTR1*) is suggested unless the patient has findings unique to NF2.

Specimen Requirements

Accepted Specimens

Specimen requirements vary based on test requested; please see our website for more details.

-**CALs or Neurofibromas**: require special media transport (kits are provided upon request, to be arranged *at least* one week in advance of procedure)
-**Fresh/Frozen Tumors**: please submit a pathology report; for additional requirements, see tumor submission checklist
-**Formalin-Fixed Paraffin-Embedded Tumors (Tumor Block):** please submit a pathology report; blocks are preferred to curls, when available; for additional requirements, see tumor submission checklist

Specimen Information:

□ Frozen □ Fresh

□ Paraffin Curls □ Paraffin Block

□ Extracted DNA; Source: _____

□ Biopsy-CAL-spot; # biopsies: _____

□ Biopsy-Neurofibroma; # biopsies: _____

Please note: failure to provide a date of collection can delay release of results

Tumor Collection Date (required): _____

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Name: (First) (MI) (Last) **DOB:** (MM/DD/YY)

Sanger Testing from Blood/Saliva/DNA

NF1/Legius syndrome and Other RASopathy Related Conditions

☐ **NF1-R: Sanger** and **Del/Dup:** *NF1* ***(RNA)***

☐ **NFSP-R: Sanger** and **Del/Dup:** *NF1* ***(RNA)*** and *SPRED1* ***(gDNA)***

Von Hippel-Lindau

☐ **VHL1: Sanger** and **Del/Dup**: *VHL*

PTEN-Related Disorders

☐ **PTEN1: Sanger** and **Del/Dup**: *PTEN*

Fragile X syndrome

☐ **FRX: PCR** and, if needed, reflexive confirmatory testing by **Southern blot** analysis: *FMR1*

Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)

☐ **MCD1: Targeted analysis of exon 11** and, if needed, reflexive full gene sequencing by **Sanger:** *ACADM*

Autosomal Recessive Polycystic Kidney Disease

☐ **PKDL: Linkage** Analysis for informativity

☐ **PKDPL: Prenatal Linkage** (see Prenatal Specimen Requirements)

FATHER: Name and DOB (mm/dd/yyyy) _____

MOTHER: Name and DOB (mm/dd/yyyy) _____

Known Variant Testing

☐ **KT2:** Targeted detection of a specific, previously identified known variant in any gene that is available at our lab by Sanger sequence, MLPA, and/or FISH analysis (**Complete Previous Testing History: Page 1**)

☐ **KT2-NG:** Targeted testing for a known variant with deep coverage of the alleles and detection of mosaicism for a variant present in at least 3% of alleles (**Complete Previous Testing History: Page 1**)

☐ **RT2:** Targeted RNA-based testing for VOUS found during Next Generation Sequencing (**Complete Previous Testing History: Page 1**)

☐ **PT2:** Prenatal testing (see Prenatal Specimen Requirements; **Complete Previous Testing History: Page 1**)

☐ **MCC:** Blood specimen for mother provided for maternal cell contamination studies (required if not previously tested)

☐ Other (unlisted options, please indicate below)

Please contact lab before selecting this option

Additional Information

Test Description Key:
Next Generation Sequencing **(NG)**
Sanger Sequencing **(Sanger)**
Deletion/Duplication analysis **(Del/Dup)**

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For additional information, visit our website at www.uab.edu/medicine/genetics/medical-genomics-laboratory

Specimen Requirements

Accepted Prenatal Specimens

Specimen requirements vary based on test requested; please see our website for more details.
-**Direct CVS**: minimum 10 mg cleaned villi
-**Direct amniotic fluid:** minimum 10 ml fluid
-**Cultured CVS**: Two T25 flasks (>70% confluent)
-**Cultured amniocytes**: Two T25 flasks (>70% confluent)

Prenatal Specimen Information:

□ Amniotic fluid □ Direct CVS (cleaned)
□ Cultured amniocytes □ Cultured villus cells

Location of back-up culture (required): _____

Sample Collected Date (required): _____

Accepted Specimens

Specimen requirements vary based on test requested; please see our website for more details.
-**Blood**: 3-6ml EDTA (must arrive within **60-72 hours of collection** for **RNA-based** tests)
-**DNA**: extracted from lymphocyte cells, a minimum of 25ul at 3µg, O.D. value at 260:280nm ≥1.6 (must be extracted in a CLIA or equivalent certified lab)
-**Sperm (for KT2-NG only)**: Fresh, sterile semen collection using a local sperm bank/cryobank facility

Specimen Information:

□ Peripheral Blood (EDTA); # Tubes: _____

□ Extracted DNA; Source: _____

□ Other, please describe: _____

Please note: failure to provide a date of collection can delay release of results

Sample Collected Date (required): _____

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Name: (First) (MI) (Last)	DOB: (MM/DD/YY)

Billing

☐ Please hold sample until further notice from the ordering facility.

Important Information

By completing this form, you **agree** that you have discussed the MGL's billing policies with your patient.
Credit card information **MUST** be provided with sample submission for self-pay clients. **Please note**: If you are paying via self-payment or requesting a benefits investigation, there will be a **3-5 working day delay** on the initiation of your test. Requests for **cancellation** of ongoing testing must be submitted to the laboratory **within three working days of specimen arrival**. Individuals or institutions submitting requests after the three working day window may still incur charges for the cost of testing.
Full information on the billing policies is available at www.uab.edu/medicine/genetics/medical-genomics-laboratory

☐ Institutional Bill

☐ Please check box if billing institution should receive report directly

Institution:		PO# (if applicable):
Address:		
City:	State:	Zip:
Contact (Name and Title):		Preferred method of contact: ☐ Email ☐ Phone
Email:	Phone:	Fax:

☐ Self-Payment Enclosed

☐ Visa ☐ MasterCard ☐ Discover ☐ American Express

Name as it appears on card:		
Card Number:	Expiration: (MM/YY)	3-digit Security Code:
Cardholder's Signature:		Preferred method of contact: ☐ Email ☐ Phone
Email:		Phone:

☐ Bill Third Party Insurance Company

Please include a copy of the pre-approval statement or provide the approval number if payment has been pre-authorized in advance of shipment.
Insurance Carrier: _____

Insurance pre-verification/authorization previously performed? ☐ Yes ☐ No If yes, **approval number is required:** _____

☐ Please check box if you would not like insurance pre-verification/authorization to be performed by the MGL.

Please send a **legible copy** of the patient's insurance card, front and back.

ICD-10 Codes **(required)**:

Important Considerations for Insurance Billing

For a list of contracted insurance companies, please visit our website or call our billing coordinator at 205-934-5523. As insurance prices are not listed, please call the billing coordinator to request a quote, if needed.
The MGL will contact the insurance provider to inquire regarding the CPT code coverage for all samples submitted for insurance payment. The healthcare provider will be contacted with the copay/deductible and also in cases where the insurance provider denies coverage of the requested codes or supporting documents are required from the provider to confirm coverage. **This service is not offered for prenatal samples.**
Please note: An insurance verification is not a guarantee of payment. Out of State Medicaid is not accepted under any circumstances. All RUSH fees must be paid up front. By completing this form, you agree that you have discussed the MGL's billing policies with your patient.

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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. Additionally, in very rare instances, this test may reveal an important genetic change that is not directly related to the clinical reason for ordering this test. This would be considered an incidental finding. The MGL reserves the right to report these incidental findings if they are clinically relevant to the patients and/or their families In such instances, these results will be discussed with my healthcare provider and additional testing may be recommended. 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* & RASOPATHIES 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

Clinical diagnosis:
- ☐ NF1
- ☐ Spinal NF
- ☐ NF Noonan
- ☐ Segmental NF1
- ☐ Noonan syndrome
- ☐ Noonan syndrome with multiple lentigines (LEOPARD) syndrome
- ☐ Cardio-facio-cutaneous syndrome (CFC)
- ☐ Costello syndrome
- ☐ Multiple CAL spots-only
- ☐ Familial multiple CAL spots-only
- ☐ Legius syndrome
- ☐ Isolated neurofibromas
- ☐ Single NF1 feature
- ☐ Unknown

NF1 NIH criteria:
- ☐ >6 CAL spots >5mm, postpubertal >15mm
- ☐ >2 neurofibromas or 1 plexiform NF
- ☐ Axillary or inguinal freckling
- ☐ First degree relative diagnosed with NF1 by above criteria
- ☐ Optic glioma
- ☐ >2 Lisch nodules
- ☐ A distinct osseous lesion

Does patient fulfill NIH diagnostic criteria for NF1? ☐ Yes ☐ No

Family history: ☐ Sporadic (proband is a "founder") ☐ Familial (proband is a "non-founder") ☐ Unknown
Consanguinity: ☐ Yes ☐ No ☐ Unknown

GENERAL INFORMATION

Height: ____ cm (☐ Short stature) Head circumference: ____ cm (☐ Macrocephaly) Weight: ____ kg

Clinical Features

Craniofacial:
- ☐ Absent
- ☐ Macrocephaly
- ☐ Palpebral ptosis
- ☐ Midface hypoplasia
- ☐ Other:_____
- ☐ Unknown
- ☐ Bitemporal narrowing
- ☐ Low posterior hairline
- ☐ Short / webbed neck
- ☐ Hypertelorism
- ☐ Low set / rotated ears
- ☐ Downslanting palpebral fissures

Ectodermal: Please provide detail on size/ location of the CAL-spots and other hyper/hypopigmentation areas on figure page 3
- ☐ Absent
- ☐ Deep palmar/plantar creases
- ☐ Multiple nevi / lentigines
- ☐ Unknown
- ☐ Dry/hyperkeratotic skin
- ☐ Abnormal/sparse eyebrows
- ☐ Hair abnormalities
- ☐ Other_____

Café-au-lait 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:

Skin fold freckling: ☐ None ☐ Unknown

	Left	Right	Comments (e.g. very faint, etc):
Groin	☐	☐	_____
Axilla	☐	☐	_____
Submammary	☐	☐	_____

Lisch nodules: ☐ None ☐ Unknown ☐ Left ☐ Right

Neurofibromas:

Cutaneous neurofibromas (soft nodules that project above the skin):
Histopathologically confirmed: Y / N Select One
☐ 0 ☐ 1 ☐ 2-6 ☐ 6-99 ☐ 100-500 ☐ >500

Intradermal neurofibromas (soft depression within the skin w/ pinkish overlying discoloration):
Histopathologically confirmed: Y / N Select One
☐ 0 ☐ 1 ☐ 2-6 ☐ 6-99 ☐ 100-500 ☐ >500

Subdermal neurofibromas (firm nodules palpable underneath the skin):
Histopathologically confirmed: Y / N Select One
☐ 0 ☐ 1 ☐ 2-6 ☐ 6-99 ☐ 100-500 ☐ >500

Plexiform neurofibromas:
Histopathologically confirmed: Y / N Select One
☐ None ☐ Visible from outside ☐ Internal
☐ With hyperpigmentation ☐ Without hyperpigmentation
☐ Head ☐ Neck ☐ Trunk ☐ L Arm ☐ L Hand ☐ L Leg ☐ L Foot
☐ Abdomen ☐ Pelvis ☐ Genital area ☐ R Arm ☐ R Hand ☐ R Leg ☐ R Foot

Spinal neurofibromas (arising from the spinal dorsal nerve root): If present, please provide detail on figure page 3
Histopathologically confirmed: Y / N Select One
☐ Unknown ☐ Absent by MRI ☐ Present, asymptomatic ☐ Present, symptomatic
☐ unilateral ☐ bilateral;
C_____ T_____, L_____, S_____ regions.

Other neoplasms: ☐ Absent ☐ Unknown

Optic glioma:
☐ Absent by MRI ☐ Present by MRI, **symptomatic** ☐ Present by MRI, **asymptomatic**
☐ Nerve (L and/or R) ☐ Chiasm

☐ Hypothalamic glioma ☐ Brainstem glioma ☐ Other glioma
☐ MPNST ☐ JMML ☐ Rhabdomyosarcoma
☐ Pheochromocytoma ☐ Colonic polyps ☐ Lipoma
☐ schwannoma ☐ meningioma ☐ juvenile xanthogranuloma
☐ breast cancer ☐ Other, specify:_____

Skeletal: ☐ Absent ☐ Unknown
☐ Long bone dysplasia ☐ Pseudarthrosis ☐ Sphenoid wing dysplasia
☐ Bone cysts ☐ scoliosis ☐ Dysplastic vertebrae
☐ pectus excavatum ☐ pectus carinatum ☐ Cubitus valgus
☐ Broad chest / telethelia ☐ Other:_____

Cardiovascular: ☐ Absent ☐ Unknown
☐ Present: ☐ Hypertension ☐ Aortic stenosis ☐ Renal artery stenosis
☐ Moya moya ☐ Pulmonary valve stenosis
☐ Arrhythmia ☐ Hypertrophic cardiomyopathy
☐ Atrial septal defect ☐ Ventricular septal defect
☐ ECG anomalies ☐ Mitral valve anomaly
☐ Unknown ☐ Other_____

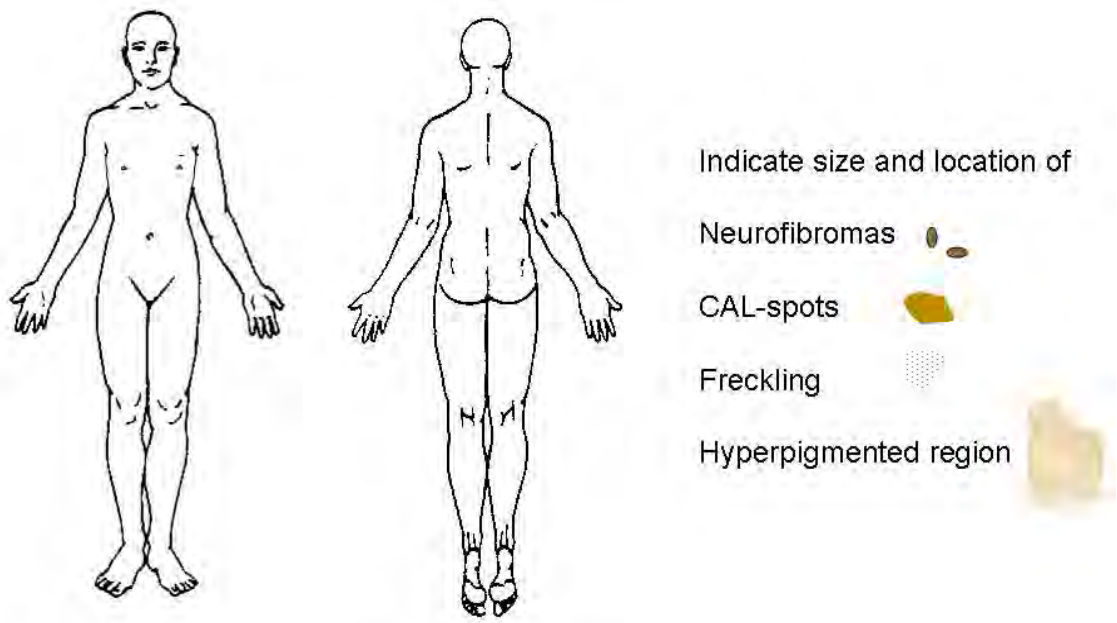
Development: ☐ Normal for age ☐ Delayed for age ☐ Hypotonic ☐ Hypertonic
☐ Gross Motor Delays ☐ Fine Motor Delays ☐ ADD ☐ Speech Delays
☐ Hyperactivity ☐ Learning disability ☐ Unknown ☐ Exam not done
☐ Other: _____ **IQ: Full scale** _____, **Verbal** _____, **Performance**_____.

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

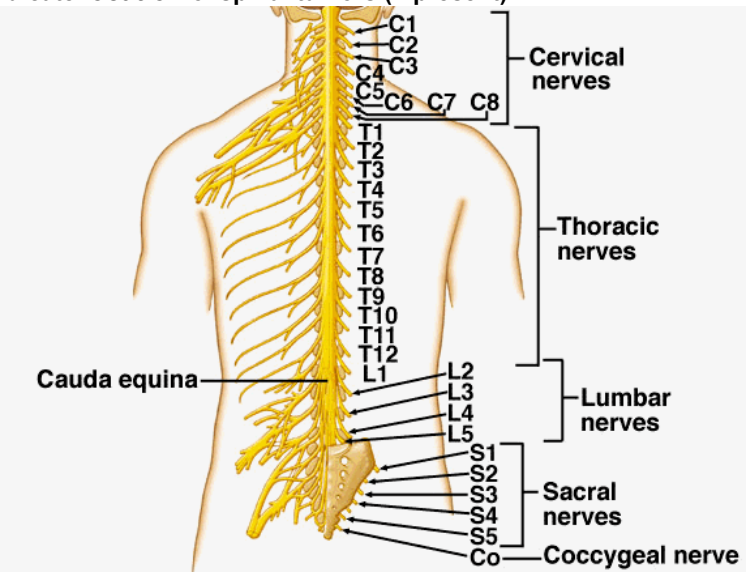
Hematological: ☐ abnormal hemostasis ☐ Factor XI deficiency ☐ Other:_____ ☐ Unknown

Segmental NF phenotype: ☐ Absent ☐ Possible

Please indicate location/size of pigmentary lesions and/or neurofibromas



Please indicate location of spinal tumors (if present)



Additional comments/remarks:

MEDICAL GENOMICS LABORATORY: NF2 & SCHWANNOMATOSIS 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

Does the patient have a clinical diagnosis of NF2? ☐ Yes ☐ No If Yes, age at diagnosis:_____

Family history: ☐ Sporadic ☐ Familial ☐ Unknown

SIGNS AND SYMPTOMS

Ear: ☐ Absent ☐ Unknown
☐ Present: ☐ Bilateral Deafness ☐ Unilateral Deafness **Age of symptoms:_____**
☐ Balance Dysfunction ☐ Tinnitus
☐ Audiometric Abnormality, please describe:_____
☐ Other, please describe:_____

Eyes ☐ Absent ☐ Unknown
☐ Present: ☐ Blindness **Age of Symptoms:_____**
☐ Lenticular opacities
☐ Lisch nodules
☐ Left ☐ Right
☐ Bilateral ☐ Unknown

Schwannomas

Vestibular schwannomas **Age of Symptoms:_____**
☐ Bilateral
☐ Unilateral
☐ No evidence of vestibular nerve tumor by MRI (Age:___yrs)
☐ Lack of symptoms of vestibular nerve tumor, but no MRI done (Age:___yrs)
☐ Unknown

Spinal schwannomas **Age of Symptoms:_____**
☐ Present
☐ No evidence by MRI (Age:___yrs)
☐ Lack of symptoms, but no MRI done (Age:___yrs)
☐ Unknown
Provide location of spinal tumors: C___ to C___, T___ to T___, L___ to L___

Other schwannomas **Age of Symptoms:_____**

☐ Absent ☐ Unknown
☐ Head ☐ Neck ☐ Trunk ☐ L Arm ☐ L Hand ☐ L Leg ☐ L Foot
☐ Abdomen ☐ Pelvis ☐ Genital area ☐ R Arm ☐ R Hand ☐ R Leg ☐ R Foot

Present only in an anatomically limited distribution(single limb or segment of the spine): ☐yes or ☐no

Result *SMARCB1*-staining on the tumor specimen: ☐Abnormal (no *SMARCB1*-staining)
☐Normal (*SMARCB1* staining)
☐Not performed

MEDICAL GENOMICS LABORATORY: NF2 & SCHWANNOMATOSIS PHENOTYPIC CHECKLIST FORM

Meningiomas Age of Symptoms:_____

- ☐ Present, Location: _____
- ☐ No evidence by MRI (Age:___yrs)
- ☐ Unknown

Other spinal tumors Age of Symptoms:_____

☐ Absent by MRI ☐ Present, asymptomatic ☐ Present, symptomatic ☐ Unknown

Pathology Known: ☐Yes, please specifiy:_____ ☐No

Provide location of spinal tumors: C___ to C___, T___ to T___, L___ to L___

Cranial nerve involvement Age of Symptoms:_____

- ☐ Present, Location: _____
- ☐ No evidence by MRI (Age:___yrs)
- ☐ Unknown

Skin

CAL spots

☐ 1 ☐ 2-3 ☐ 4-5 ☐ >5-10 ☐ >10 Age of Symptoms:_____

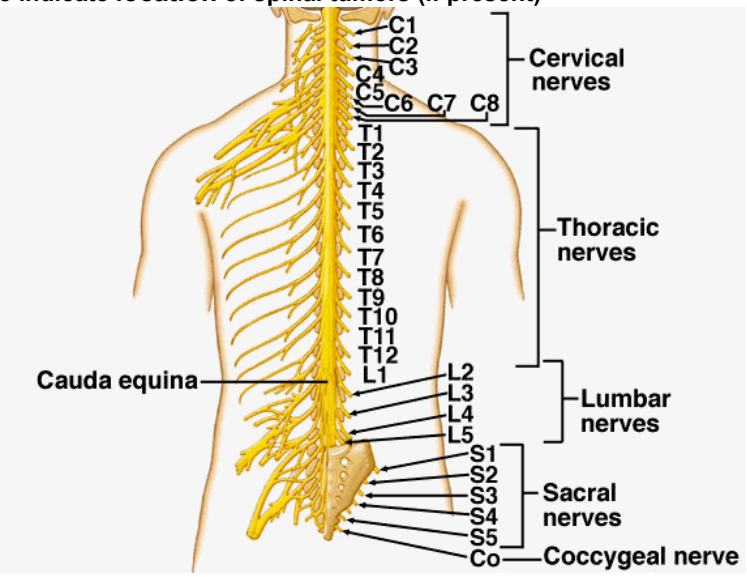
Neurofibromas

☐ 1-5 ☐ ≥6-99 ☐ ≥100 Age of Symptoms:_____

Skin fold freckling	Left	Right
Groin	☐	☐
Axilla	☐	☐
Submammary	☐	☐

Age of Symptoms:_____

Please indicate location of spinal tumors (if present)



Additional comments/remarks: