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

Accession:
For MGL Laboratory Use only

Test Requisition Form

- This form must accompany all specimens received
- Billing instructions are available on page 5
- All specimens received must include two patient identifiers
- Testing must be ordered by a qualified clinician

Additional testing information is available at www.genetics.uab.edu/medgenomics

Patient Information:		**Ordering Physician:**	
Date of specimen collection:		☐ **Please check box if physician should receive report directly**	
Patient Name: (First) (MI) (Last)		Name:	NPI:
DOB: (MM/DD/YY)	MRN:	Institution:	
Address:		Address:	
City: State: Zip:		City: State: Zip:	
Gender:	SSN:	Email:	
Phone:	Email:	Phone:	Fax:
Parent or Guardian Name (if minor):		**Additional Reports to**	
Please list other information here:		Name	
		Address:	
		City: State: Zip:	
		Institution:	Email:
		Phone:	Fax:

For MGL Lab Use only:				**Lab/Hospital Information:**	
	Initials:	Date:	Comment:	☐ **Please check box if Lab/Hospital should receive report directly**	
Received:				Name:	
Reviewed:				Address:	
Accession:				City: State: Zip:	
Billing:				Email:	
Other:				Phone:	Fax:

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:_____

Patient History (Please check all that apply)

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

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:

Updated 2/21/2018

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DOB: (MM/DD/YY)

Comprehensive Testing for Constitutional/Mosaic Mutations with Deep Coverage via NGS

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

Acceptable Specimen Types
- **Blood,** (3-6ml EDTA; no time limitations associated with receipt)
- **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)
- **Fibroblasts**

Key used below:
Next Generation Sequencing (NGS)
Sanger Sequencing (Sanger)
Deletion/Duplication analysis (Del/Dup)

☐ **RUSH Analysis: Testing completed within 15 working days of receipt of sample**
(Additional $600 RUSH fee applied; only available for tests listed on page 2)

NF1/SPRED1 and Other RASopathy Related Conditions

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

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

☐ **NNP-NG: NGS** 16 genes (no *NF1*): *BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2,*and *SPRED1* and **Del/Dup**: *SPRED1*

☐ **RAS-NG**: **NGS** 17 genes: *BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2,* and *SPRED1* and **Del/Dup**: *NF1* and *SPRED1*

☐ **SPD1-NG: NGS** and **Del/Dup**: *SPRED1* only

☐ **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*
Del/Dup: *NF2*, *LZTR1*, and *SMARCB1*

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

Peripheral Nerve Sheath Tumor Testing

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

Rhabdoid Tumor Predisposition Syndrome

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

Tuberous Sclerosis Complex

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

Capillary Malformation Arteriovenous Malformation Syndrome

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

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.

The average coverage of our panel is >1800x. Specifically for the *NF1* gene, the NGS array covers >99.8% of the *NF1* coding region at ≥350X and 100% ≥200X, allowing detection of very low level mosaicism, down to 3-5% MAF respectively (regions covered by ≥350X respectively ≥200X).

For all remaining genes on our panels, the NGS array covers >99.5% of the coding region at ≥350X and 99.2% covered at ≥200X. Remaining regions are covered at ≥30X.

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, please visit our website at www.genetics.uab.edu/medgenomics.

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

Date collected:

□ Peripheral Blood (EDTA); # Tubes:	□ Saliva (kit must be provided by MGL)
□ Extracted DNA; Source:	□ Other, please describe:

Updated 2/21/2018

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Tumor/Biopsy Based Comprehensive Testing

Key used below:
Next Generation Sequencing (NGS)
Sanger Sequencing (Sanger)
Deletion/Duplication analysis (Del/Dup)

NF1/SPRED1 on biopsied CALs and Neurofibromas

☐ **NF14C: Sanger** and **Del/Dup**: *NF1* and *SPRED1* on **biopsied CALs**

☐ **NF14N: Sanger** and **Del/Dup**: *NF1* on **biopsied 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.****

Peripheral Nerve Sheath Tumor Testing

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

NF2/Schwannomatosis/Meningiomatosis
(Please choose testing options based on correct specimen)

Fresh/Frozen Tumor

☐ **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***;** **Del/Dup**: *NF2 & SMARCB1*

Tumor Block

☐ **NF24: Sanger** and **Del/Dup**: *NF2*

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

Tuberous Sclerosis Complex

☐ **TSCP-NG: NGS** and **Del/Dup**: *TSC1* & *TSC2* on **Fresh/Frozen Tumor**

Rhabdoid Tumor Predisposition Syndrome

Fresh/Frozen Tumor

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

Tumor Block

☐ **SB14RT: Sanger** and **Del/Dup**: *SMARCB1*

☐ **Please check here if blood is provided for confirmation testing.**

Important points of consideration for testing

When proceeding with biopsy based testing for NF1, RNA-based tissue culture analysis would be the suggested starting point. Please contact the laboratory before ordering this test as media must be provided in advance.

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.

When proceeding with tumor based testing for NF2, test code "SCH-NG" (*NF2, SMARCB1, and LZTR1*) is suggested unless the patient also has additional findings unique to NF2.

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, please visit our website at www.genetics.uab.edu/medgenomics.

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

Date collected:

☐ Peripheral Blood (EDTA); # Tubes:	☐ Saliva (kit must be provided by MGL)
☐ Extracted DNA; Source:	☐ Other, please describe:
☐ Biopsy-CAL-spot; # biopsies:	☐ Biopsy-Neurofibroma; # biopsies:

☐ Tumor (specify location on checklist):
☐ Frozen ☐ Fresh ☐ Paraffin Block ☐ Paraffin Curls

Pathology:

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

DOB: (MM/DD/YY)

Sanger Testing from Blood/Saliva/DNA

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

Acceptable Specimen Types

- **Blood,** (3-6ml EDTA; no time limitations associated with receipt)
- **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)
- **Fibroblasts**

Key used below:
Sanger Sequencing (Sanger)
Deletion/Duplication analysis (Del/Dup)

NF1/SPRED1 and RASopathy Related Conditions

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

☐ **NFSP1-R: Sanger and Del/Dup:** *NF1**(RNA)*** & *SPRED1 **(gDNA)***

Von Hippel Lindau

☐ **VHL1: Sanger & Del/Dup:** *VHL*

PTEN Related Disorders

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

Autosomal Recessive Polycystic Kidney Disease

☐ **PKDL: Linkage** Analysis for informativity

☐ **PKDPL: Prenatal** Linkage

PARENT: Father's Name and DOB (mm/dd/yyyy):
_____

PARENT: Mother's Name and DOB (mm/dd/yyyy):
_____

Fragile X syndrome

☐ **FRX: PCR and Southern Blot analysis**: *FMR1*

Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)

☐ **MCD1: Targeted analysis of exon 11:** *ACADM*

☐ **MCD2: Sanger:** *ACADM*

Other (Please contact laboratory before selecting this testing option)

Known Mutation Testing

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

☐ **PT2:** Prenatal testing (**Complete Previous Testing History: Page 1)**

☐ **MCC:** Blood specimen for mother provided for maternal cell contamination studies (required)

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

☐ **KT2-NGS:** Targeted testing for a known mutation with deep coverage of the alleles and detection of mosaicism for a mutation present in <3% mutant allele fraction (MAF) (**Complete Previous Testing History: Page 1**)

Important points of consideration for testing

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, please visit our website at www.genetics.uab.edu/medgenomics.

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

Date collected:

☐ Peripheral Blood (EDTA); # Tubes:	☐ Saliva (kit must be provided by MGL)
☐ Extracted DNA; Source:	☐ Other, please describe:

Prenatal Testing

☐ Amniotic Fluid	☐ Cultured Amniocytes
☐ Direct CVS (cleaned)	☐ Cultured Villus Cells

Location of back-up culture (required):

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Billing

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

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. Credit card information MUST be provided with sample submission for self-pay clients. Full information on the billing policies is available at www.genetics.uab.edu/medgenomics.

Please note: If you are paying via self-payment or desiring a benefits investigation, there will be a 3-5 business day delay on the initiation of your test

☐ **Institutional Bill** Please check box if billing institution should receive report directly: ☐

Institution:	PO#	
Address:		
City:	State:	Zip:
Contact:	Contact Title:	
Email:	Phone:	Fax:

☐ **Payment Enclosed** ☐ 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 Third Party Insurance Company** Insurance pre-verification\authorization previously performed? ☐ Yes ☐ No

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

ICD-10 Diagnosis Codes (required):

Please send a legible copy of the patient's insurance card, front and back. All RUSH fees must be paid up front.

For a list of contracted insurance companies, please visit our website at www.genetics.uab.edu/medgenomics or call our billing coordinator at 205-934-5523.

The MGL will contact the insurance provider to inquire regarding the CPT code coverage for all samples submitted for insurance payment. The provider will be contacted if: a) the insurance provider denies coverage of the requested codes b) supporting documents are required from the provider to confirm coverage c) a copay/deductible is expected to exceed $500. **This service is not completed on prenatal samples**. **Please note: An insurance verification is not a guarantee of payment.**

☐ **Please check box if you would not like this service to be performed by the MGL.**

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

SMARCB1 (Alias: *INI1*) PHENOTYPIC CHECKLIST FORM – UAB MEDICAL GENOMICS LABORATORY

Referring Physician:_____ **Date of Exam** ___/___/___

DEMOGRAPHIC INFORMATION

Gender : ☐ **Male** ☐ **Female** **Date of Birth:** ___/___/___

Ethnicity: **Mother:** ☐ **White** ☐ **African-American** ☐ **Native American** ☐ **Hispanic** ☐ **Asian** ☐ **Other**
Father: ☐ **White** ☐ **African-American** ☐ **Native American** ☐ **Hispanic** ☐ **Asian** ☐ **Other**

AT/RT (Atypical Teratoid/Rhabdoid Tumor predisposition syndrome)-related phenotypic checklist To be completed if AT/RT is suspected

Clinical history relevant to tumor:

Date of surgery:

Treatment prior to surgery may impact the quality of genetic testing on the tumor specimen (e.g. chemotherapy). Please specify:

Location of the tumor:
- ☐ **Brain**
- ☐ **Kidney**
- ☐ **Spine**
- ☐ **Cerebral spinal fluid**
- ☐ **Other, please specify:**

Result *SMARCB1*-staining on the tumor specimen:
- ☐ **Abnormal (no *SMARCB1*-staining)**
- ☐ **Normal (*SMARCB1* staining)**
- ☐ **Unknown**

Family history: ☐ **Sporadic** ☐ **Familial** ☐ **Unknown**

If familial, specify location and type of tumor(s) in family members:

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Tumor Specimen Submission Checklist

The following requirements must be met in order to process tumor specimens for Neurofibromatosis type 2 and/or Schwannomatosis testing. The UAB Medical Genomics Lab now proudly offer Fresh/Frozen Tumor testing utilizing Next Generation Sequencing. Tumor blocks will still be completed using Sanger sequencing. Please confirm that the submitted tissue meets each requirement by placing a check mark next to each statement. If your pathology department is unable to confirm this information for you, please select the checkbox below. If Neurofibromatosis Type 1 is your primary concern, neurofibromas and biopsied CALs still require being collected in our media and are run via Sanger sequencing. Please contact the MGL at (205) 934-5562 to request collection media for these sample types.

For NGS Sequencing

Fresh Tumor Specimen Checklist	
	This tumor is a least 5mm-cubed
	This specimen contains at least 60% pure tumor content
	This specimen has been sent in basic, sterile culture media such as RPMI or PBS
Frozen Tumor Specimen Checklist	
	This tumor is at least 5mm-cubed
	This specimen contains at least 60% pure tumor content
	This specimen has been snap frozen and sent on dry ice

For Sanger Sequencing	
Formalin-fixed paraffin embedded block	
	This tumor block has a surface area of a least 5mm squared **or** This specimen contains at least 3-6 loose paraffin curls (no slides) that are 30-50 microns thick
	This tumor specimen contains greater than 70% nucleated cells
	This specimen contains at least 60% pure tumor content
Notes or Special Comments	

For tumor blocks only:

□ I agree to have the UAB Pathology Laboratory confirm the specimen requirements listed above for the specimen(s) submitted to the Medical Genomics Laboratory for genetic testing. I understand that this additional analysis will cost $50 per tumor specimen submitted for testing (CPT code: 88399).

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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	