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 Fax: (205) 996-2929			Accession: For	MGL Laborat	tory Use only	
			Test Requ	isition Form		
- This form mu - Billing instru	ist accompany all ctions are availab	specimens rec le on page 5	eived	- All spec - Testing	cimens receive must be order	d must include two patient identifiers red by a qualified clinician
	I	Additional test	ting information is availal	ble at www.genetics.u	ab.edu/medge	enomics
	Patien	t Information	:		Orderii	ng Physician:
Date of specin	nen collection:			\Box Please check box	x if physician	should receive report directly
Patient Name:	(First) (M	(Last)		Name:		NPI:
DOB: (MM/D)	D/YY)	MRN:		Institution:		
Address:				Address:		
City:		State:	Zip:	City:	S	tate: Zip:
Gender:		SSN:		Email:		
Phone:		Email:		Phone:		Fax:
Parent or Guar	dian Name (if mi	nor):			Addition	nal Reports to
Please list othe	er information her	e:		Name		
				Address:		
				City:	Sta	tte: Zip:
				Institution:		Email:
				Phone:		Fax:
	For MG	L Lab Use on	ly:		Lab/Hospi	tal Information:
	Initials:	Date:	Comment:	□ Please check box	x if Lab/Hosp	ital should receive report directly
Received:				Name:		
Reviewed:				Address:		
Accession:				City:	Sta	tte: Zip:
Billing:				Email:		
Other:				Phone:		Fax:
			Informe	d Consent:		
Provider's sta consent handou Informed conse	tement: I acknow ut; and I have disc ent has been obtai	vledge the risks cussed the test(ined from the p	s, benefits, limitations, and (s) requested with the patien batient/guardian and the har	implications of genetic nt/guardian and I have d copy will be maintai	c testing as out answered his/h ned.	lined on the complete informed her questions regarding testing.
	P	rovider's Sign				
Patient History (Please check all that apply)						
	diseases (AIDS, H	Hepatitis, etc.)		Patient has had a	chemotherapy	in the past 6 months
Patient has	had a bone marro	ow transplant		□ Patient or family	member is pr	egnant LMP:
			Previous 7	Festing History		
		Ha	s this patient or relatives ha	d previous testing?	Yes □ No	
Name/Relationship to	patient:			Test/Mutation/Lab:		
Name/Relationship to	patient:			Test/Mutation/Lab:		

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Patient Name: (First) (MI) (Last)	DOB: (MM/DD/YY)		
Comprehensive Testing for Constitutional/Mo	osaic Mutations with Deep Coverage via NGS		
If multiple tests are requested, please specify order in which testing sh	ould 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 	<u>Key used below:</u> Next Generation Sequencing (NGS) Sanger Sequencing (Sanger) Deletion/Duplication analysis (Del/Dup)		
RUSH Analysis: Testing completed wit	hin 15 working days of receipt of sample		
(Additional \$600 RUSH fee applied; on	ly available for tests listed on page 2)		
NF1/SPRED1 and Other RASopathy Related Conditions	NF2/Schwannomatosis/Meningiomatosis		
I NF1-NG: NGS and Del/Dup: NF1 only	□ NF2-NG: NGS and Del/Dup: NF2 only		
■ NFSP-NG: NGS and Del/Dup: NF1 and SPRED1 only	SCH-NG: NGS 3 genes: <i>LZTR1, NF2,</i> and <i>SMARCB1</i> Del/Dup : <i>NF2, LZTR1,</i> and <i>SMARCB1</i>		
MAP2K1, MAP2K2, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2, and SPRED1 and Del/Dup: SPRED1	■ MEN-NG: NGS 4 genes: NF2, SMARCB1, SMARCE1, and SUFU: Del/Dup: NF2 and SMARCB1		
\square RAS-NG: NGS 17 genes: RRAF CRI HRAS KRAS MAP2KI	Peripheral Nerve Sheath Tumor Testing		
MAP2K2. NF1. NRAS. PPP1CB. PTPN11. RAF1. RASA2. RIT1.	PNT-NG: NGS 6 genes: NF1. NF2. KRAS. LZTR1. PTPN11 and		
SHOC2, SOS1, SOS2, and SPRED1 and Del/Dup: NF1 and SPRED1	SMARCB1; Del/Dup: NF1, NF2, LZTR1, and SMARCB1		
SPD1-NG: NGS and Del/Dup : <i>SPRED1</i> only	Rhabdoid Tumor Predisposition Syndrome		
CST-NG: NGS: <i>HRAS</i> only	RT-NG: NGS and Del/Dup : <i>SMARCB1</i> only		
Tuberous Sclerosis Complex	Capillary Malformation Arteriovenous Malformation Syndrome		
TSCP-NG: NGS and Del/Dup : <i>TSC1</i> and <i>TSC2</i> only	RASA-NG: NGS and Del/Dup : <i>RASA1</i> only		
Important points of cor	sideration for testing		
The MGL offers next generation sequencing testing options that provi 3% of the alleles, depending on coverage in the regions of interest.	de the ability to identify variants (indels and substitutions) as low as		
The average coverage of our panel is >1800x. Specifically for the <i>NF1</i> gene, the NGS array covers >99.8% of the <i>NF1</i> coding region at \geq 350X and 100% \geq 200X, allowing detection of very low level mosaicism, down to 3-5% MAF respectively (regions covered by \geq 350X respectively \geq 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 o Please contact the lab via phone (205) 934-5562 or via email at medgenon For additional information, please visit our website at www.genetics.uab.e	rder form. nics@uabmc.edu if you have any questions when completing this form. du/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:		



Image: 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 Fax: (205) 996-2929	For MGL Laborat	ory Use only
Patient Name: (First) (MI) (Last)	DOB: (MM/DD/YY)	
Tumor/Biopsy Based C	comprehensive Testing	
<u>Key used</u> Next Generation S Sanger Sequen Deletion/Duplication	<u>l below:</u> equencing (NGS) ccing (Sanger) a analysis (Del/Dup)	
NF1/SPRED1 on biopsied CALs and Neurofibromas	NF2/Schwannomatosis/Meningie (Please choose testing options ba	omatosis (sed on correct specimen)
NF14C: Sanger and Del/Dup : <i>NF1</i> and <i>SPRED1</i> on biopsied CALs	Fresh/Frozen Tumor	Tumor Block
□ NF14N: Sanger and Del/Dup: <i>NF1</i> on biopsied neurofibromas	□ NF2-NG: NGS and Del/Dup: NF2 only	■ NF24: Sanger and Del/Dup: NF2
** 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.**	□ SCH-NG: NGS 3 genes: <i>LZTR1, NF2,</i> and <i>SMARCB1</i> and Del/Dup : <i>NF2, LZTR1,</i> and <i>SMARCB1</i>	SCHP: Sanger and Del/Dup : <i>NF2, LZTR1,</i> and <i>SMARCB1</i>
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	■ MEN-NG: NGS 4 genes: NF2, SMARCB1, SMARCE1, and SUFU; Del/Dup: NF2 & SMARCB1	
Tuberous Sclerosis Complex	Rhabdoid Tumor Predisposition	Syndrome
TSCP-NG: NGS and Del/Dup : <i>TSC1</i> & <i>TSC2</i> on Fresh/Frozen Tumor	Fresh/Frozen Tumor RT-NG: NGS and Del/Dup: SMARCB1	Tumor Block SB14RT: Sanger and Del/Dup : <i>SMARCB1</i>
□ Please check here if blood is p	rovided for confirmation testing.	1
Important points of con	nsideration for testing	
When proceeding with biopsy based testing for NF1, RNA-based tissue cu laboratory before ordering this test as media must be provided in advance.	ulture analysis would be the suggeste	d starting point. Please contact the
The MGL offers next generation sequencing testing options that provi 3% of the alleles, depending on coverage in the regions of interest.	ide the ability to identify variants (indels and substitutions) as low as
When proceeding with tumor based testing for NF2, test code "SCH-N also has additional findings unique to NF2.	NG" (NF2, SMARCB1, and LZTR1) is suggested unless the patient
Please contact the lab via phone (205) 934-5562 or via email at medgenon For additional information, please visit our website at www.genetics.uab.e	nics@uabmc.edu if you have any que edu/medgenomics.	estions when completing this form.
Specimen requirements vary based on test requested; please see www	.genetics.uab.edu/medgenomics for	r more details.
Peripheral Blood (EDTA); # Tubes:	\Box Saliva (kit must be provided by	MGL)
L Extracted DNA; Source:	U Other, please describe:	
□ Biopsy-CAL-spot; # biopsies:	□ Biopsy-Neurofibroma; # biopsie	28:
□ Tumor (specify location on checklist): □ Frozen □ Fresh □ Parat	ffin Block 🗆	Paraffin Curls
Pathology:		

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www.genetics.uab.edu/medgenomics			
Patient Name: (First) (MI) (Last)	DOB: (MM/DD/YY)		
Sanger Testing fro	om Blood/Saliva/DNA		
If multiple tests are requested, please spec	fy order in which testing should be performed.		
Acceptable Specimen Types			
• Blood , (3-6ml EDTA; no time limitations associated with			
• Saliya. (OGR-575 DNA Genotek: kits are provided upon	Key used below:		
request)	Sanger Sequencing (Sanger)		
• DNA, , (extracted from lymphocyte cells, a minimum of 25ul at	Deletion/Duplication analysis (Del/Dup)		
$3\mu g$, O.D. value at 260:280nm ≥ 1.6 , must be extracted in a CLIA or equivalent certified lab.			
• Fibroblasts			
NF1/SPRED1 and RASopathy Related Conditions	Von Hippel Lindau		
□ NF1-R: Sanger and Del/Dup: <i>NF1(RNA)</i>	□ VHL1: Sanger & Del/Dup: VHL		
\square NFSP1-R: Sanger and Del/Dup: $NF1(RNA)$ & SPRED1 (gDNA)	PTEN Related Disorders		
	PTEN1: Sanger and Del/Dup : <i>PTEN</i>		
Autosomal Recessive Polycystic Kidney Disease	Fragile X syndrome		
PKDL: Linkage Analysis for informativity	FRX: PCR and Southern Blot analysis : <i>FMR1</i>		
	Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)		
PKDPL: Prenatal Linkage	MCD1: Targeted analysis of evon 11: ACADM		
	MCD2: Sanger: ACADM		
PARENT: Father's Name and DOB (mm/dd/yyyy):	Other (Please contact laboratory before selecting this testing option)		
PARENT: Mother's Name and DOB (mm/dd/yyyy):			
Known Mu	itation Testing		
KT2: Targeted detection of a specific, previously identified known mu	atation in any gene that is available at our lab by Sanger sequence, MLPA,		
and/or FISH analysis (Complete Previous Testing History: Page 1)			
D DT2 . Denoted testing (Complete Descions Testing History Desc 1)			
I P12 : 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 Gene	ration Sequencing (Complete Previous Testing History: Page 1)		
	nation Sequencing (complete 110/1005 1 esting 1150019/1 uge 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: Pag	e 1)		
Important points of	consideration for testing		
For additional testing options via tumor/biopsy, please see page 3 of this of Please contact the lab via phone (205) 934-5562 or via email at medgenor For additional information please visit our website at www.genetics.uab.	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.		
Specimen requirements vary based on test requested: please see www	.genetics.uab.edu/medgenomics for more details.		
Date collected:	<u>.</u>		
□ Peripheral Blood (EDTA); # Tubes:	□ Saliva (kit must be provided by MGL)		
Extracted DNA; Source:	□ Other, please describe:		
Prena	al Testing		
Amniotic Fluid	Cultured Amniocytes		
Direct CVS (cleaned)	Cultured Villus Cells		
Location of back-up culture (required):			



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Patient Name: (First) (MI) (Last)	DOB: (MM/DD/YY)		
В	illing		
□ Please hold sample until further notice from the order	ing 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 your test	investigation, there will be a 3-5 busin	ess day delay on the initiation of	
□ Institutional Bill Please check box if billing institution should receiv	e 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\aut	horization previously performed?	□ 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:			



LAB MEDICAL GENOMICS LABORATORY

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Informed Consent for Genetic Testing

This form does not need to be returned to the Medical Genomics	s Laboratory if Informed Consent F	oortion 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.



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. <u>For Prenatal Testing</u>: 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 I	Name

