720 Twentieth Street South, Suite 330 Birmingham, Alabama 35294-0005 Phone: (205) 934-5562 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 Additional information is available online at warmers.			-All specimens received must include <u>two</u> patient identifiers and <u>collection date</u> -Testing must be ordered by a qualified clinician lab.edu/medicine/genetics/medical-genomics-laboratory						
	Additi	onal Information		isition Form		edicai-ge	enomics-iaborat	ory	
	Patier	nt Informatio	·			Ord	ering Physici	an:	
Sample Collected: (MM/DD/YY)			□ Please check box if physician should receive report directly						
		(c)		Name:				NPI:	
Legal Name: (First) (MI) (Last)			In with this co.						
				Institution:					
DOB: (MM/DD/YY)		MRN:		Address:					
Address:				City:			State:	Zip:	
City:		State:	Zip:	Country:	untry: Phone		:		
Sex at Birth:		SSN:		Please check presult deliver	Please check preferred Fax				
Parent or Guardian name	e (if minor):	:		□ Email:		•			
	Referrir	ng Lab/Hospit	tal:			Addit	ional Report	s to:	
□ Please check box if lab/	hospital sh	ould receive rep	ort directly	Name:					
Name: Institution:				Institution:					
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City:		State:	Zip:	Country:		Phone	<u> </u> :		
Country:	Country: Phone:			Please check p	oreferred	dd			
Please check preferred result delivery:				result delivery:					
☐ Email:				□ Email:					
			Previous Te	esting Histor	У				
Charle III that and	☐ Patient or family member is pregnant. LMP:				☐ Patient has had chemotherapy in the past 6 months				
Check all that apply:	☐ Patient has had a bone marrow transplant				☐ Infectious diseases (AIDS, Hepatitis, etc.)			Hepatitis, etc.)	
		На	as this patient or relatives ha	nad previous testing? ☐ Yes ☐ No					
Name/Relationship to patient:			Test/Variant/Lab:						
Name/Relationship to patient:			Test/Variant/Lab:						
			Informe	d 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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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 NF2/Schwannomatosis/Meningiomatosis ☐ 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 ☐ SCH-NG: NGS: 3 genes: LZTR1, NF2, and SMARCB1; and Del/Dup: □ NNP-NG: NGS: 17 genes (no NF1): BRAF, CBL, HRAS, KRAS, LZTR1, NF2, LZTR1, and SMARCB1 MAP2K1, MAP2K2, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2, and SPRED1; and Del/Dup: SPRED1 and LZTR1 ☐ MEN-NG: NGS: 4 genes: NF2, SMARCB1, SMARCE1, and SUFU; and Del/Dup: NF2 and SMARCB1 ☐ RAS-NG: NGS: 18 genes: BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2. NF1. NRAS. PPP1CB. PTPN11. RAF1. RASA2. RIT1. SHOC2. SOS1. Peripheral Nerve Sheath Tumor Testing SOS2, and SPRED1; and Del/Dup: NF1, SPRED1, and LZTR1 ☐ PNT-NG: NGS: 6 genes: NF1, NF2, KRAS, LZTR1, PTPN11 and ☐ CST-NG: NGS: HRAS only SMARCB1; and Del/Dup: NF1, NF2, LZTR1, and SMARCB1 McCune-Albright Syndrome **Rhabdoid Tumor Predisposition Syndrome** ☐ GNAS-NG: NGS: GNAS exons 8 and 9 only ☐ RT-NG: NGS: SMARCB1 and SMARCA4; and Del/Dup: SMARCB1 only **Tuberous Sclerosis Complex** Capillary Malformation Arteriovenous Malformation Syndrome ☐ TSCP-NG: NGS and Del/Dup: TSC1 and TSC2 ☐ 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 \geq 350X and 99% \geq 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 \geq 200X. Remaining regions are covered at \geq 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 Information:			
Specimen requirements vary based on test requested; please see our	□ Peripheral Blood (EDTA); # Tubes:			
website for more details.	□ Extracted DNA; Source:			
-Blood: 3-6ml EDTA (receipt within one week of collection)	□ Saliva (kit must be provided by MGL)			
-Saliva: OGR-575 DNA Genotek (kits are provided upon request)	□ Other, please describe:			
A: extracted from lymphocyte cells, a minimum of 25ul at 3µg, O.D. e at 260:280nm ≥1.6 (must be extracted in a CLIA or equivalent ified lab)	Please note: failure to provide a date of collection can delay release of results			
-Fibroblast cells	Sample Collected Date (required):			

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Name: 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) NF2-, LZTR1-, SMARCB1-related Schwannomatosis NF1/SPRED1 on biopsied CALs and Neurofibromas ☐ NF2-NG: Fresh/Frozen Tumor or Tumor Block for NGS with reflex *CURRENTLY UNDER MORATORIUM UNTIL 2023* to Sanger as needed and Del/Dup: NF2 only **Please contact the laboratory at least one week in advance of the ☐ SCH-NG: Fresh/Frozen Tumor or Tumor Block for NGS with reflex biopsy before ordering this test as media must be provided in advance to Sanger as needed and Del/Dup: NF2, LZTR1, and SMARCB1 and special shipping instructions apply. Biopsies must arrive within 60 hours of collection** Rhabdoid Tumor Predisposition Syndrome □ NF14C: Sanger(RNA) and Del/Dup: NF1 (with automatic reflex to ☐ RT-NG: Fresh/Frozen Tumor or Tumor Block for NGS with reflex to SPRED1) on biopsied CALs Sanger as needed SMARCB1 and SMARCA4; and Del/Dup: SMARCB1 ☐ NF14N:Sanger(RNA) and Del/Dup: NF1 on biopsied neurofibromas **RASopathy Related Conditions** Meningiomatosis ☐ MEN-NG: Fresh/Frozen Tumor or Tumor Block for NGSwith reflex to Sanger as needed: NF2, SMARCB1, SMARCE1, and SUFU; and Del/Dup: NF2 and SMARCB1 □ NNP-NG: Fresh/Frozen Tumor for NGS (no *NF1*) or Tumor Block for NGS with reflex to Sanger as needed: BRAF, CBL, HRAS, KRAS, LZTR1, Peripheral Nerve Sheath Tumor Testing MAP2K1, MAP2K2, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, ☐ PNT-NG: Fresh/Frozen Tumor for NGS with reflex to Sanger as SOS1, SOS2, and SPRED1; and Del/Dup: SPRED1 and LZTR1 needed: NF1, NF2, KRAS, LZTR1, PTPN11 and SMARCB1; and Del/Dup: NF1, NF2, LZTR1, and SMARCB1 ☐ RAS-NG: Fresh/Frozen Tumor or Tumor Block for NGS with reflex to **Tuberous Sclerosis Complex** Sanger as needed: BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2, ☐ TSC-NG: Fresh/Frozen Tumor or Tumor Block for NGS with and SPRED1; and Del/Dup: NF1, SPRED1, and LZTR1 reflex to Sanger as needed 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-related SWN, test code "SCH-NG" (NF2, SMARCB1, and LZTR1) is suggested unless the patient has findings unique to NF2.

Specimen Requirements					
Accepted Specimens	Specimen Information:				
Specimen requirements vary based on test requested; please see our website for more details.	□ Frozen □ Fresh				
	□ Paraffin Curls □ Paraffin Block				
-CALs or Neurofibromas: require special media transport (kits are provided upon request, to be arranged at least one week in advance of procedure)	□ Extracted DNA; Source:				
-Fresh/Frozen Tumors: please submit a pathology report; for additional	□ Biopsy-CAL-spot; # biopsies:				
uirements, see tumor submission checklist rmalin-Fixed Paraffin-Embedded Tumors (Tumor Block): please submit a nology report; blocks are preferred to curls, when available; for additional uirements, see tumor submission checklist	□ 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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bank/cryobank facility

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Name: DOB: (MM/DD/YY) Sanger Testing from Blood/Saliva/DNA NF1/Legius syndrome and Other RASopathy Related Conditions Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD) ☐ NF1-R: Sanger and Del/Dup: NF1 (RNA) ☐ MCD1: Targeted analysis of exon 11 and, if needed, reflexive full gene sequencing by Sanger: ACADM □ NFSP-R: Sanger and Del/Dup: NF1 (RNA) and SPRED1 (qDNA) Von Hippel-Lindau Autosomal Recessive Polycystic Kidney Disease ☐ VHL1: Sanger and Del/Dup: VHL ☐ PKDL: Linkage Analysis for informativity PTEN-Related Disorders ☐ PKDPL: Prenatal Linkage (see Prenatal Specimen Requirements) ☐ PTEN1: Sanger and Del/Dup: PTEN FATHER: Name and DOB (mm/dd/yyyy) Fragile X syndrome ☐ FRX: PCR and, if needed, reflexive confirmatory testing by MOTHER: Name and DOB (mm/dd/yyyy) Southern blot analysis: FMR1 **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: Please contact the lab via phone (205) 934-5562 or via email at medgenomics@uabmc.edu if you have any Next Generation Sequencing (NG) questions when completing this form. Sanger Sequencing (Sanger) For additional information, visit our website at www.uab.edu/medicine/genetics/medical-genomics-laboratory Deletion/Duplication analysis (Del/Dup) Specimen Requirements **Accepted Prenatal Specimens** Prenatal Specimen Information: Specimen requirements vary based on test requested; please see our website □ Amniotic fluid □ Direct CVS (cleaned) for more details. □ Cultured villus cells □ Cultured amniocytes -Direct CVS: minimum 10 mg cleaned villi -Direct amniotic fluid: minimum 10 ml fluid Location of back-up culture (required): -Cultured CVS: Two T25 flasks (>70% confluent) Sample Collected Date (required): -Cultured amniocytes: Two T25 flasks (>70% confluent) **Accepted Specimens** Specimen Information: Specimen requirements vary based on test requested; please see our website ☐ Peripheral Blood (EDTA); # Tubes: for more details. □ Extracted DNA; Source: -Blood: 3-6ml EDTA (must arrive within 60-72 hours of collection for ☐ Other, please describe: RNA-based tests) -DNA: extracted from lymphocyte cells, a minimum of 25ul at 3μg, O.D. value at Please note: failure to provide a date of collection can delay release of results 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

Sample Collected Date (required):

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mingham, Alabama 35294-0005 Fax: (205) 996-2929 www.uab.edu/medicine/genetics/medical-genomics-laboratory

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Name:	(First)	(MI)	(Last)		DOB: (MM)	DOB: (MM/DD/YY)	
Billing							
		☐ Please h	nold sample until furthe	er notice from the	ordering facil	ity.	
			Important	Information			
			agree that you have d				
						: If you are paying via self-payment or Requests for cancellation, test change, or	
	thod change of ongo	oing testing must	be submitted to the la	aboratory within t	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						
☐ Institution		Title billing polici	es is available at <u>www</u>	.aab.caa/mcaicii	ic/genetics/ii	redicting insortatory	
☐ Please check box if billing institution should receive report directly							
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Address:				T			
City:				State:		Zip:	
Contact (Name and Title):						Preferred method of contact: ☐ Email ☐ Phone	
Email: Phone:						Fax:	
☐ Self-Payment Enclosed *PLEASE ENSURE ALL INFORMATION IS LEGIBLE*							
□ Visa □ MasterCard □ Discover □ Ame				☐ Americ	an Express		
Name as it appears on card:							
Card Number: Expiration: (MM/YY)					I/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 <u>not</u> 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.							
Important Considerations for Insurance Billing							
		Impo	rtant Consideration	ons for Insura	nce 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. The ordering provider/clinician's office is responsible for obtaining prior authorization, if it is required. 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 <u>any</u> 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.