

For UAB MGL Use Only		Received	Reviewed	Lab Accession	Billing Review	Other	Status/Notes: UAB MGL Accession:
	Initials						
	Date						

This form must accompany all samples received. Please contact the lab at (205) 934-5562 or medgenomics@uabmc.edu if you have any questions.
All samples must include two patient identifiers and a collection date, and billing information must be included.

PATIENT INFORMATION				
First Name		MI	Last Name	
Patient ID/MRN		Sex at Birth F M Other	Gender (Optional)	Date of Birth (MM/DD/YYYY)
Check all that Apply	☐ Patient or family member is pregnant EDD: _____		☐ Patient has had bone marrow transplant	
	☐ Please HOLD testing for this sample until notified		☐ Patient has had chemotherapy in the past 6 months	
	☐ Previous genetic testing (please attach a copy of report)		☐ Infectious diseases (AIDS, Hepatitis, etc)	

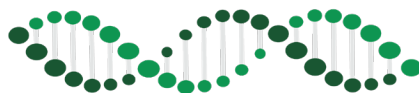
ORDERING PHYSICIAN				SENDING FACILITY (optional)			
Name		NPI		Name			
Institution				Institution			
Mailing Address				Mailing Address			
City	State	Zipcode	Country	City	State	Zipcode	Country

RESULTS DELIVERY			
Please provide contact information and select who should receive a copy of this report.			
☐ The ordering provider listed	Email ☐	Fax ☐	Phone ☐
☐ The sending facility listed	Email ☐	Fax ☐	Phone ☐
Another Party (Name) ☐	Email ☐	Fax ☐	Phone ☐

Sample Type	Requirements	Required Patient Sample Information
☐ Peripheral Blood # of tubes: _____	3-6ml EDTA *Received within 60 hours of collection for RNA-based testing	Collection Date: _____ ☐ For paired tumor/biopsy testing (confirmation testing) ☐ For Maternal Cell Contamination (MCC) studies (blood only)
☐ Saliva	Saliva kit must be Genotek OGD-575; kits provided upon request	
☐ Extracted DNA Source: _____	A minimum of 25uL at 3ug, O.D. value at 260:280nm ≥1.6 (must be extracted in a CLIA or equivalent certified lab)	Collection Date: _____ Volume & concentration if known: _____ ☐ For paired tumor/biopsy testing (confirmation testing) ☐ For Maternal Cell Contamination (MCC) studies (blood only)
☐ Fresh/Frozen tumor	Minimum 5-mm cubed, 60% pure tumor content, and marked with collection location/tumor type.*	Collection Date(s): _____ ☐ Pathology Report Included *See Tumor Specimen Submission Checklist for more information.
☐ Tumor Block ☐ Paraffin curls # of blocks: _____	Pathology report required for each sample. 60% pure tumor content, 70% nucleated cells. Tumor block preferred. Minimum 5-mm squared OR at least 3-6 loose paraffin curls (no slides) 30-50 microns thick.*	
☐ Café au lait biopsies ☐ Neurofibromas	Special media transport is required; request for kit send-out needed 1 week prior to the biopsy. 3 containers of media per kit.	Collection Date: _____
☐ Cultured Fibroblasts	Two T25 flasks (>70% confluent) required	Collection Date: _____
☐ Direct CVS (cleaned)	Minimum 10 mg cleaned villi required	Collection Date: _____
☐ Direct Amnio Fluid	Minimum 10 mL fluid	Location of back-up culture: _____
☐ Cultured CVS ☐ Cultured Amniocytes	Two T25 flasks (>70% confluent) required	
☐ Semen/sperm	Fresh, sterile semen collection using a local sperm bank/medical facility/fertility clinic/cryobank	Collection Date: _____

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

Medical Professional's Signature:	Date:
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Patient Name:	DOB:
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BILLING

☐ **Institutional Bill** ☐ Please **check** this box if the billing institution should receive a report directly

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

☐ **Self-Payment** ***Credit card information must be provided with sample*** *Please ensure all information is legible*

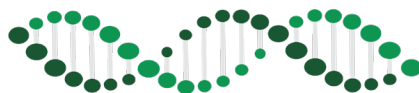
Name as it appears on card:		Cardholder's Signature:	
Type: ☐ Visa ☐ MasterCard ☐ Discover ☐ American Express			
Card Number:		Expiration: /	Security Code:
Cardholder's preferred form of contact:	☐ Email:	☐ Phone:	

☐ **Bill Third Party Insurance Company** *Please include a copy of the patient's insurance card, front and back*

Prior authorization approval number:		ICD-10 Code(s):
Patient insurance carrier:	☐ Please check if you have submitted a benefits investigation request	

Important Billing Considerations – By completing this form, you agree that you have discussed the UAB MGL's billing policies with your patient.

- Institutional hold pending insurance confirmation can be placed by selecting "Please HOLD testing for this sample until notified" on page 1.**
- Requests for cancellation, test change, or change in billing information must be submitted to the laboratory within three working days of sample arrival. Individuals or institutions submitting change requests after the three working day window may still incur charges for the cost of testing.
- 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.
- The UAB MGL will contact the insurance provider to check CPT code coverage for all samples submitted for insurance payment. The healthcare provider will be emailed the benefit findings within 48 hours after the request is received. The benefit findings will have an **estimated** patient responsibility including the expected OOP/deductible. The ordering provider/clinician's office is responsible for obtaining prior authorizations, pre-determinations and letter of medical necessity if required for coverage.
- Please note: benefits investigation is not a guarantee of payment. **Out of state Medicaid is not accepted. All RUSH fees must be paid up front.**
- Full information on the UAB MGL billing policy & list of contracted carriers can be found at <https://www.uab.edu/medicine/genetics/medical-genomics-laboratory/billing> or by calling our billing coordinator at 205-934-5523.



Patient Name:	DOB:
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NEXT GENERATION SEQUENCING (Blood/Saliva and Tumor)

- Constitutional testing can be done on unaffected tissue including **saliva, blood (EDTA), or fibroblasts**.
- Tumor testing can be done on **fresh/frozen tumors, tumor blocks, and loose tumor curls**. Slides are NOT accepted.
- Extracted DNA** from the above sources can be used for NGS constitutional/tumor tests. Please contact the lab for any other unlisted specialized tests.
- Tumor testing includes up to 2 tumors and a blood or saliva specimen for loss of heterozygosity and confirmation Sanger Sequencing to identify germline disease.
- A common variant in two anatomically distinct tumors/biopsies is required to identify mosaic conditions when no variant is present in blood.
- Paraffin-embedded tumor may not pass quality control for NGS. *NF2, LZTR1, and SMARCB1* are the only genes that can reflex to Sanger sequencing as an alternative.

Blood/ Saliva	Tumor	Test Selection	☐ Please RUSH testing (Blood/Saliva ONLY- additional \$600 fee applies)
		NF1-NG NGS and del/dup of <i>NF1</i> only <i>(Please contact the laboratory if NF1 NGS tumor testing wanted)</i>	
		NFSP-NG NGS and del/dup of <i>NF1</i> and <i>SPRED1</i>	
		RAS-NG NGS of 18 genes: <i>BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1/2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1/2, SPRED1</i> and del/dup of <i>NF1, SPRED1, LZTR1</i>	
		NNP-NG NGS of 17 genes: <i>BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1/2, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2, SPRED1</i> and del/dup of <i>SPRED1</i> and <i>LZTR1</i> (no <i>NF1</i>)	
		NF2-NG NGS and del/dup of <i>NF2</i> only with Sanger sequencing reflex of <i>NF2</i> as needed	
		SCH-NG NGS and del/dup of <i>LZTR1, NF2, SMARCB1</i> with Sanger sequencing reflex of <i>NF2, SMARCB1, and LZTR1</i> as needed	
		MEN-NG NGS of <i>NF2, SMARCB1, SMARCE1</i> and <i>SUFU</i> and del/dup of <i>NF2</i> and <i>SMARCB1</i> with Sanger sequencing reflex of <i>NF2, SMARCB1, SMARCE1, and SUFU</i> as needed	
		RT-NG (Rhabdoid Tumor Predisposition) NGS of <i>SMARCB1, SMARCA4</i> and del/dup of <i>SMARCB1</i> with Sanger sequencing reflex of <i>SMARCB1</i> and <i>SMARCA4</i> as needed	
		PNT-NG NGS of <i>NF1, NF2, KRAS, LZTR1, PTPN11, SMARCB1</i> and del/dup of <i>NF1, NF2, LZTR1, SMARCB1</i>	
		TSCP-NG (Tuberous Sclerosis) NGS and del/dup of <i>TSC1/2</i> with Sanger sequencing reflex of <i>TSC1/2</i> as needed	
		RASA-NG (CM-AVM) NGS of <i>RASA1</i> and <i>EPHB4</i>	
		GNAS-NG (McCune-Albright) NGS of <i>GNAS</i> exons 8 and 9 only	
		Single Gene NGS Please select: <i>BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1/2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1/2, SPRED1, NF2, SMARCB1, SMARCE1, SMARCA4, SUFU, RASA1, EPHB4, TSC1, TSC2</i>	

RNA BLOOD AND BIOPSY CULTURE-BASED

- For biopsy-culture based testing, please **contact the laboratory** at least one week in advance of the biopsy before ordering this test as **media must be provided** and special shipping instructions apply. Please include a blood sample with biopsies for loss of heterozygosity testing and confirmation Sanger Sequencing.
- Biopsies and blood (EDTA)** collected for **RNA and culturing** must arrive at the laboratory within **60 hours** of collection. Extracted DNA **CANNOT** be used for RNA.

Test Selection	
NF14C	Sanger (RNA) and del/dup of <i>NF1</i> with automatic reflex to <i>SPRED1</i> on 2-3 café au lait macule biopsies
NF14N	Sanger (RNA) and del/dup of <i>NF1</i> on 2-3 neurofibroma biopsies
NF1-R	RNA-based Sanger and del/dup of <i>NF1</i> only on blood
NFSP-R	RNA-based Sanger and del/dup of <i>NF1</i> and DNA-based <i>SPRED1</i> on blood
NF2-R	RNA-based Sanger and del/dup of <i>NF2</i> on blood

KNOWN VARIANT TESTING

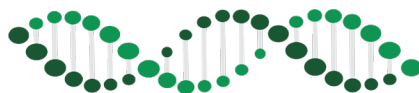
Name/Relationship to patient	Test/Variant/Lab*
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*Please attach a copy of testing done at other laboratories. Extracted DNA is accepted for KT2, KT2-NG, and PT2 tests (NOT RT2/RNA testing).

Test Selection	
KT2	Targeted testing of a known/familial variant in a gene available at our lab. Via Sanger, MLPA, and/or FISH on blood, saliva
KT2-NG	Targeted testing of a known variant via NGS to detect mosaicism as low as 3% VAF on blood, saliva, sperm
RT2	RNA-based targeted testing for VUS with a potential splicing effect on blood <60 hours old
PT2	Prenatal testing for known familial variant **MCC Required** on chorionic villi, amniocytes

OTHER

Test Selection	Test Selection
FRX PCR and, if needed, reflexive confirmatory testing by Southern blot analysis: <i>FMR1</i> on blood	MCD1 Targeted analysis of exon 11 and, if needed, reflexive full gene sequencing by Sanger: <i>ACADM</i>
VHL1 Sanger sequencing and Del/Dup of <i>VHL</i>	PTEN1 Sanger sequencing and Del/Dup of <i>PTEN</i>
Other (unlisted option; please indicate below): <i>*Please contact the lab before selecting this option</i>	



INFORMED CONSENT FOR GENETIC TESTING

I hereby consent for:

Name

DOB

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):

Test

I understand that:

- 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.
- Any samples obtained will be used attempting to determine if I and/or members of my family carry genetic changes in the disease genes ordered by my physician.
- The genetic tests performed at the MGL are highly sensitive and specific to the disease being tested. However, sensitivity and specificity are test-dependent. Additional testing details and the specific detection rates of each test can be found at <https://www.uab.edu/medicine/genetics/medical-genomics-laboratory>.
- 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.

- 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.
- 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.
- 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.
- 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.
- 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@uabmc.edu or calling the laboratory at 205-934-5562.
- 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.
- 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.
- 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

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.