

<b>UAB MEDICAL GENOMICS LABORATORY</b> 720 South Twentieth Street, Suite 330      Tel: (205) 934-5562 Birmingham, Alabama 35294-0005      Fax: (205) 996-2929 <a href="http://www.genetics.uab.edu/medgenomics">www.genetics.uab.edu/medgenomics</a>				Accession: For MGL Laboratory Use only	
<b>Test Requisition Form</b>					
- This form must accompany all specimens received			- All specimens received must include two patient identifiers		
- Billing instructions are available on page 5			- Testing must be ordered by a qualified clinician		
<b>Additional testing information is available at <a href="http://www.genetics.uab.edu/medgenomics">www.genetics.uab.edu/medgenomics</a></b>					
<b>Patient Information:</b>			<b>Ordering Physician:</b>		
<b>Date of specimen collection:</b>			<input type="checkbox"/> <b>Please check box if physician should receive report directly</b>		
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):			<b>Additional Reports to</b>		
Please list other information here:			Name		
			Address:		
City:		State:	Zip:		
City:		State:	Zip:		
Institution:		Email:			
Phone:		Fax:			
<b>For MGL Lab Use only:</b>			<b>Lab/Hospital Information:</b>		
			<input type="checkbox"/> <b>Please check box if Lab/Hospital should receive report directly</b>		
Received:			Name:		
Reviewed:			Address:		
Accession:			City:		State:      Zip:
Billing:			Email:		
Other:			Phone:		Fax:
<b>Informed Consent:</b>					
<b>Provider's statement:</b> 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.					
<b>Provider's Signature:</b> _____					
<b>Patient History</b> (Please check all that apply)					
<input type="checkbox"/> Infectious diseases (AIDS, Hepatitis, etc.)			<input type="checkbox"/> Patient has had chemotherapy in the past 6 months		
<input type="checkbox"/> Patient has had a bone marrow transplant			<input type="checkbox"/> Patient or family member is pregnant    LMP:		
<b>Previous Testing History</b>					
Has this patient or relatives had previous testing? <input type="checkbox"/> Yes <input type="checkbox"/> 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)
<b>Comprehensive Testing for Constitutional/Mosaic Mutations with Deep Coverage via NGS</b>	
<b>If multiple tests are requested, please specify order in which testing should be performed.</b>	
<b>Acceptable Specimen Types</b> <ul style="list-style-type: none"> <li>• <b>Blood</b>, (3-6ml EDTA; no time limitations associated with receipt)</li> <li>• <b>Saliva</b>, (OGR-575 DNA Genotek; kits are provided upon request)</li> <li>• <b>DNA</b>, (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)</li> <li>• <b>Fibroblasts</b></li> </ul>	<b>Key used below:</b> <b>Next Generation Sequencing (NGS)</b> <b>Sanger Sequencing (Sanger)</b> <b>Deletion/Duplication analysis (Del/Dup)</b>
<input type="checkbox"/> <b>RUSH Analysis: Testing completed within 15 working days of receipt of sample</b> (Additional \$600 RUSH fee applied; only available for tests listed on page 2)	
<b>NF1/SPRED1 and Other RASopathy Related Conditions</b>  <input type="checkbox"/> <b>NF1-NG: NGS and Del/Dup: <i>NF1</i> only</b>  <input type="checkbox"/> <b>NFSP-NG: NGS and Del/Dup: <i>NF1</i> and <i>SPRED1</i> only</b>  <input type="checkbox"/> <b>NNP-NG: NGS 16 genes (no <i>NF1</i>): <i>BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2,</i> and <i>SPRED1</i> and <b>Del/Dup: <i>SPRED1</i></b>   <input type="checkbox"/> <b>RAS-NG: NGS 17 genes: <i>BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2,</i> and <i>SPRED1</i> and <b>Del/Dup: <i>NF1</i> and <i>SPRED1</i></b>   <input type="checkbox"/> <b>SPD1-NG: NGS and Del/Dup: <i>SPRED1</i> only</b>   <input type="checkbox"/> <b>CST-NG: NGS: <i>HRAS</i> only</b> </b></b>	<b>NF2/Schwannomatosis/Meningiomatosis</b>  <input type="checkbox"/> <b>NF2-NG: NGS and Del/Dup: <i>NF2</i> only</b>  <input type="checkbox"/> <b>SCH-NG: NGS 3 genes: <i>LZTR1, NF2,</i> and <i>SMARCB1</i></b> <b>Del/Dup: <i>NF2, LZTR1,</i> and <i>SMARCB1</i></b>  <input type="checkbox"/> <b>MEN-NG: NGS 4 genes: <i>NF2, SMARCB1, SMARCE1,</i> and <i>SUFU</i></b> <b>Del/Dup: <i>NF2</i> and <i>SMARCB1</i></b>  <b>Peripheral Nerve Sheath Tumor Testing</b> <input type="checkbox"/> <b>PNT-NG: NGS 6 genes: <i>NF1, NF2, KRAS, LZTR1, PTPN11</i> and <i>SMARCB1</i>; <b>Del/Dup: <i>NF1, NF2, LZTR1,</i> and <i>SMARCB1</i></b>   <b>Rhabdoid Tumor Predisposition Syndrome</b>  <input type="checkbox"/> <b>RT-NG: NGS and Del/Dup: <i>SMARCB1</i> only</b>   <b>Capillary Malformation Arteriovenous Malformation Syndrome</b>  <input type="checkbox"/> <b>RASA-NG: NGS and Del/Dup: <i>RASA1</i> only</b> </b>
<b>Tuberous Sclerosis Complex</b>	
<input type="checkbox"/> <b>TSCP-NG: NGS and Del/Dup: <i>TSC1</i> and <i>TSC2</i> only</b>	
<b>Important points of consideration for testing</b>	
<p>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.</p> <p>The average coverage of our panel is &gt;1800x. Specifically for the <i>NF1</i> gene, the NGS array covers &gt;99.8% of the <i>NF1</i> 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).</p> <p>For all remaining genes on our panels, the NGS array covers &gt;99.5% of the coding region at ≥350X and 99.2% covered at ≥200X. Remaining regions are covered at ≥30X.</p> <p>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 <a href="mailto:medgenomics@uabmc.edu">medgenomics@uabmc.edu</a> if you have any questions when completing this form.          For additional information, please visit our website at <a href="http://www.genetics.uab.edu/medgenomics">www.genetics.uab.edu/medgenomics</a>.</p>	
<b>Specimen requirements vary based on test requested; please see <a href="http://www.genetics.uab.edu/medgenomics">www.genetics.uab.edu/medgenomics</a> for more details.</b>	
Date collected:	
<input type="checkbox"/> Peripheral Blood (EDTA); # Tubes:	<input type="checkbox"/> Saliva (kit must be provided by MGL)
<input type="checkbox"/> Extracted DNA; Source:	<input type="checkbox"/> Other, please describe:



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Patient Name: (First)      (MI)      (Last)		DOB: (MM/DD/YY)	
<b>Tumor/Biopsy Based Comprehensive Testing</b>			
<b>Key used below:</b> Next Generation Sequencing (NGS) Sanger Sequencing (Sanger) Deletion/Duplication analysis (Del/Dup)			
<b>NF1/SPRED1 on biopsied CALs and Neurofibromas</b>		<b>NF2/Schwannomatosis/Meningiomatosis (Please choose testing options based on correct specimen)</b>	
<input type="checkbox"/> <b>NF14C: Sanger and Del/Dup: NF1 and SPRED1 on biopsied CALs</b>  <input type="checkbox"/> <b>NF14N: Sanger and Del/Dup: NF1 on biopsied neurofibromas</b>  <b>**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.**</b>		<b>Fresh/Frozen Tumor</b>  <input type="checkbox"/> <b>NF2-NG: NGS and Del/Dup: NF2 only</b>  <input type="checkbox"/> <b>SCH-NG: NGS 3 genes: LZTR1, NF2, and SMARCB1 and Del/Dup: NF2, LZTR1, and SMARCB1</b>  <input type="checkbox"/> <b>MEN-NG: NGS 4 genes: NF2, SMARCB1, SMARCE1, and SUFU; Del/Dup: NF2 &amp; SMARCB1</b>	
<b>Peripheral Nerve Sheath Tumor Testing</b>		<b>Tumor Block</b>	
<input type="checkbox"/> <b>PNT-NG: NGS 6 genes: NF1, NF2, KRAS, LZTR1, PTPN11, and SMARCB1; Del/Dup: NF1, NF2, LZTR1, and SMARCB1 on Fresh/Frozen Tumor</b>		<input type="checkbox"/> <b>NF24: Sanger and Del/Dup: NF2</b>  <input type="checkbox"/> <b>SCHP: Sanger and Del/Dup: NF2, LZTR1, and SMARCB1</b>	
<b>Tuberous Sclerosis Complex</b>		<b>Rhabdoid Tumor Predisposition Syndrome</b>	
<input type="checkbox"/> <b>TSCP-NG: NGS and Del/Dup: TSC1 &amp; TSC2 on Fresh/Frozen Tumor</b>		<b>Fresh/Frozen Tumor</b> <input type="checkbox"/> <b>RT-NG: NGS and Del/Dup: SMARCB1</b>	
		<b>Tumor Block</b> <input type="checkbox"/> <b>SB14RT: Sanger and Del/Dup: SMARCB1</b>	
<input type="checkbox"/> <b>Please check here if blood is provided for confirmation testing.</b>			
<b>Important points of consideration for testing</b>			
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.			
<b>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.</b>			
<b>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.</b>			
Please contact the lab via phone (205) 934-5562 or via email at <a href="mailto:medgenomics@uabmc.edu">medgenomics@uabmc.edu</a> if you have any questions when completing this form. For additional information, please visit our website at <a href="http://www.genetics.uab.edu/medgenomics">www.genetics.uab.edu/medgenomics</a> .			
<b>Specimen requirements vary based on test requested; please see <a href="http://www.genetics.uab.edu/medgenomics">www.genetics.uab.edu/medgenomics</a> for more details.</b>			
Date collected:			
<input type="checkbox"/> Peripheral Blood (EDTA); # Tubes:		<input type="checkbox"/> Saliva (kit must be provided by MGL)	
<input type="checkbox"/> Extracted DNA; Source:		<input type="checkbox"/> Other, please describe:	
<input type="checkbox"/> Biopsy-CAL-spot; # biopsies:		<input type="checkbox"/> Biopsy-Neurofibroma; # biopsies:	
<input type="checkbox"/> Tumor (specify location on checklist): <input type="checkbox"/> Frozen <input type="checkbox"/> Fresh <input type="checkbox"/> Paraffin Block <input type="checkbox"/> Paraffin Curls			
Pathology:			



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Patient Name: (First) (MI) (Last)	DOB: (MM/DD/YY)
<b>Sanger Testing from Blood/Saliva/DNA</b>	
If multiple tests are requested, please specify order in which testing should be performed.	
<b>Acceptable Specimen Types</b> <ul style="list-style-type: none"> <li>• <b>Blood</b>, (3-6ml EDTA; no time limitations associated with receipt)</li> <li>• <b>Saliva</b>, (OGR-575 DNA Genotek; kits are provided upon request)</li> <li>• <b>DNA</b>, , (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)</li> <li>• <b>Fibroblasts</b></li> </ul>	<b>Key used below:</b> <b>Sanger Sequencing (Sanger)</b> <b>Deletion/Duplication analysis (Del/Dup)</b>
<b>NFI/SPRED1 and RASopathy Related Conditions</b>	<b>Von Hippel Lindau</b>
<input type="checkbox"/> <b>NF1-R: Sanger and Del/Dup: <i>NF1(RNA)</i></b>	<input type="checkbox"/> <b>VHL1: Sanger &amp; Del/Dup: <i>VHL</i></b>
<input type="checkbox"/> <b>NFSP1-R: Sanger and Del/Dup: <i>NF1(RNA)</i> &amp; <i>SPRED1 (gDNA)</i></b>	<b>PTEN Related Disorders</b>
	<input type="checkbox"/> <b>PTEN1: Sanger and Del/Dup: <i>PTEN</i></b>
<b>Autosomal Recessive Polycystic Kidney Disease</b>	<b>Fragile X syndrome</b>
<input type="checkbox"/> <b>PKDL: Linkage Analysis for informativity</b>	<input type="checkbox"/> <b>FRX: PCR and Southern Blot analysis: <i>FMRI</i></b>
<input type="checkbox"/> <b>PKDPL: Prenatal Linkage</b>	<b>Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)</b>
<b>PARENT: Father's Name and DOB (mm/dd/yyyy):</b> _____	<input type="checkbox"/> <b>MCD1: Targeted analysis of exon 11: <i>ACADM</i></b>
<b>PARENT: Mother's Name and DOB (mm/dd/yyyy):</b> _____	<input type="checkbox"/> <b>MCD2: Sanger: <i>ACADM</i></b>
	<b>Other (Please contact laboratory before selecting this testing option)</b>
<b>Known Mutation Testing</b>	
<input type="checkbox"/> <b>KT2:</b> 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 ( <b>Complete Previous Testing History: Page 1</b> )	
<input type="checkbox"/> <b>PT2:</b> Prenatal testing ( <b>Complete Previous Testing History: Page 1</b> )	
<input type="checkbox"/> <b>MCC:</b> Blood specimen for mother provided for maternal cell contamination studies (required)	
<input type="checkbox"/> <b>RT2:</b> Targeted RNA based testing for VOUS found during Next Generation Sequencing ( <b>Complete Previous Testing History: Page 1</b> )	
<input type="checkbox"/> <b>KT2-NGS:</b> 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) ( <b>Complete Previous Testing History: Page 1</b> )	
<b>Important points of consideration for testing</b>	
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 <a href="http://www.genetics.uab.edu/medgenomics">www.genetics.uab.edu/medgenomics</a> .	
<b>Specimen requirements vary based on test requested; please see <a href="http://www.genetics.uab.edu/medgenomics">www.genetics.uab.edu/medgenomics</a> for more details.</b>	
Date collected:	
<input type="checkbox"/> Peripheral Blood (EDTA); # Tubes:	<input type="checkbox"/> Saliva (kit must be provided by MGL)
<input type="checkbox"/> Extracted DNA; Source:	<input type="checkbox"/> Other, please describe:
<b>Prenatal Testing</b>	
<input type="checkbox"/> Amniotic Fluid	<input type="checkbox"/> Cultured Amniocytes
<input type="checkbox"/> Direct CVS (cleaned)	<input type="checkbox"/> Cultured Villus Cells
Location of back-up culture (required):	



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Patient Name: (First) (MI) (Last)			DOB: (MM/DD/YY)		
<b>Billing</b>					
<input type="checkbox"/> <b>Please hold sample until further notice from the ordering facility.</b>					
<p>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 <a href="http://www.genetics.uab.edu/medgenomics">www.genetics.uab.edu/medgenomics</a>.</p>					
<b>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</b>					
<input type="checkbox"/> <b>Institutional Bill</b> Please check box if billing institution should receive report directly: <input type="checkbox"/>					
Institution:			PO#		
Address:					
City:		State:		Zip:	
Contact:			Contact Title:		
Email:		Phone:		Fax:	
<input type="checkbox"/> <b>Payment Enclosed</b> <input type="checkbox"/> Visa <input type="checkbox"/> MasterCard <input type="checkbox"/> Discover <input type="checkbox"/> American Express					
Name as it appears on card:					
Card Number:					
Expiration Date:			3-digit Security code:		
Cardholder Signature:					
Cardholder Email Address:					
<input type="checkbox"/> <b>Bill Third Party Insurance Company</b> Insurance pre-verification/authorization previously performed? <input type="checkbox"/> Yes <input type="checkbox"/> No					
<b><u>Please Note: Out of State Medicaid is not accepted under any circumstances</u></b>					
<b>ICD-10 Diagnosis Codes (required):</b>					
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 <a href="http://www.genetics.uab.edu/medgenomics">www.genetics.uab.edu/medgenomics</a> 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. <b>This service is not completed on prenatal samples. Please note: An insurance verification is not a guarantee of payment.</b>					
<input type="checkbox"/> <b>Please check box if you would not like this service to be performed by the MGL.</b>					
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:					



**MEDICAL GENOMICS LABORATORY: TSC1/TSC2 PHENOTYPIC CHECKLIST FORM**



Patient Name: \_\_\_\_\_  Male  Female Date of Birth \_\_\_/\_\_\_/\_\_\_  
Referring Physician: \_\_\_\_\_ Date of Exam \_\_\_/\_\_\_/\_\_\_

Thank you for completing this form; phenotypic information may improve our ability to interpret your results.

**DEMOGRAPHIC INFORMATION**

Ethnicity:  White  Black  Native American  Hispanic  Asian  Other:

**DIAGNOSIS**

2012 International TSC Consensus Conference Clinical Criteria: (Northrup et al. Pediatric Neuro. 2013 Oct;49(4):243-54.)  
**(Criteria are listed below with major criteria marked by # and minor criteria marked by \*.)**

- Definite TSC (2 major or 1 major plus 2 minor features)
- Possible TSC (1 major or 2 or more minor features)
- Does not meet TSC Criteria

Clinical Concern for Mosaicism:  No  Yes  
Family history:  Sporadic  Familial  Unknown

Known Familial Mutation:  No  Yes (provide information: \_\_\_\_\_)

**Familial cases:** Please provide pedigree and details on the affection status of family members on a separate page. Attach prior mutational analysis if available.

**SIGNS AND SYMPTOMS (Major criteria marked by # and minor criteria marked by \*.)**

**Dermatological**

- 1) Hypomelanotic Macules:  0  1-2  ≥3#  Unknown
- 2) "Confetti" skin lesions:  None  Present\*  Unknown
- 3) Facial Angiofibromas:  None  1-3  >3#  Unknown
- 4) Shagreen Patch:  None  Present#  Unknown
- 5) Cephalic Fibrotic Plaque:  None  Present#  Unknown
- 6) Ungal/Periungal fibromas:  None  1-2  >2#  Unknown
- 7) Hyperpigmented Macules:  0  1-2  ≥3  Unknown

Please provide detail on size and location of the dermatological findings on page 3.  
A digital picture of the skin findings would be very helpful.

**Neurological (Imaging)**

- 8) Cortical Dysplasia:
  - Cortical Tubers:  None  Present#  Unknown
  - Cerebral White Matter Radial Migration Lines:  None  Present#  Unknown
- 9) Subependymal nodule (SEN):  None  Present#  Unknown
- 10) Subependymal Giant Cell Astrocytoma (SEGA):  None  Present#  Unknown  
Histopathologically Confirmed?  No  Yes

**Neurological/Psychiatric (Clinical)**

- 11) Seizures:  None  Present (Describe type, if known: \_\_\_\_\_)  Unknown
- 12) Developmental/Intellectual Disabilities:  None  Present  Unknown
- 13) Behavioral/Psychiatric Diagnoses:  None  ADHD  Isolated hyperactivity  Aggression  
 Autism  unknown  Other (\_\_\_\_\_)

**Renal**

- 14) Angiomyolipomas:  None  1-2  >2#  Unknown  
Histopathologically Confirmed?  No  Yes  
Is a malignant angiomyolipoma present?  No  Yes
- 15) Renal Cell Carcinoma:  None  Present  Unknown
- 16) Renal Epithelial Cysts:  None  1-2  >2\*  Unknown
- 17) Polycystic Kidney Disease Features:  None  Present  Unknown

Note: If present, there may be increased concern for TSC-PKD contiguous deletion.

**MEDICAL GENOMICS LABORATORY: TSC1/TSC2 PHENOTYPIC CHECKLIST FORM**



Patient Name: \_\_\_\_\_ Date of Birth \_\_\_\_/\_\_\_\_/\_\_\_\_  
**(Criteria are listed below with major criteria marked by # and minor criteria marked by \*.)**

**Pulmonary**

18) Lymphangioleiomyomatosis (LAM):  None  Present#  Unknown  
 Histopathologically Confirmed?  No  Yes

**Cardiac**

19) Rhabdomyomas:  None  Present#  Formerly/Prenatally present, but regressed  Unknown

**Dental**

20) Dental Enamel Pits:  None  1-3  >3\*  Unknown  
 21) Intraoral Fibromas:  None  1-2  >2\* (Location: \_\_\_\_\_)  Unknown

**Ophthalmological**

22) Retinal Hamartomas:  None  Single  Multiple#  Unknown  
 23) Retinal Achromic Patch:  None  Present\*  Unknown  
 24) Retinal Astrocytic Hamartomas:  None  Present  Unknown

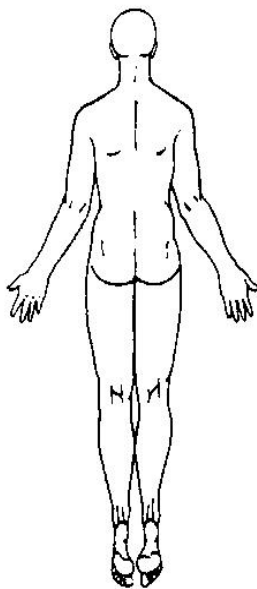
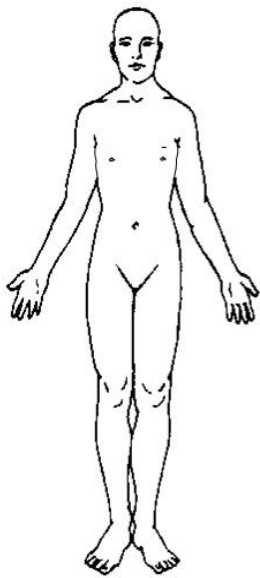
**Neuroendocrine**

25) Neuroendocrine Tumors:  None  Unknown  Yes (Specify type: \_\_\_\_\_)

**Other**

26) Nonrenal Hamartomas:  None  Present\*  Unknown  
 27) Additional Phenotypic Information:

**Indicate location/size of hypomelanotic macules or other dermatological lesions ↓**



- Hypopigmented lesions
- Hyperpigmented lesions
- Shagreen patch
- "Confetti" lesions



## 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:
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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:
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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](http://www.genetics.uab.edu/medgenomics).
  
4. The following are possible outcomes for the specific tests listed above:

<b>Positive</b>	<b>Unknown Significance</b>	<b>Negative</b>
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.



