

FOR LAB USE ONLY
ACCESSION NO:
DATE & TIME RECEIVED:
TECHNICIAN:

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

INI1/SMARCB1 TEST REQUEST FORM

THIS FORM AND PHENOTYPIC CHECKLIST MUST BE FILLED OUT COMPLETELY

PATIENT NAME:		BIRTH DATE:	DAYTIME PHONE:	SEX:	SOC. SEC. NUMBER:
PATIENT'S ADDRESS:		CITY:	STATE:	ZIP CODE:	MED REC NUMBER:
EMAIL ADDRESS:			PARENT OR GUARDIANS NAME (IF MINOR):		
INI1/SMARCB1 MUTATION ANALYSIS TESTING			Physician's Name:		
<input type="checkbox"/> Comprehensive INI1/SMARCB1 Test (Test 1): <input type="checkbox"/> by RNA <input type="checkbox"/> by DNA <input type="checkbox"/> Copy Number Analysis by MLPA <i>only</i> <input type="checkbox"/> Targeted Mutation Analysis (Test 2) Proband _____ <input type="checkbox"/> Prenatal Targeted Analysis (Test 3) Proband _____ <input type="checkbox"/> Comprehensive Test on Tumor (Test 4)			Physician's Address: _____ _____		
Facility where specimen obtained:			Phone: _____ Fax: _____		
Date:			NPI Number: _____ Email: _____		
REQUIRED Diagnosis (ICD-9) Code (only in US):			ADDITIONAL REPORTS TO:		
Is Patient Pregnant?		Please check if applicable:		Name:	
<input type="checkbox"/> No <input type="checkbox"/> Yes If yes, date of LMP: _____		<input type="checkbox"/> Infectious diseases (AIDS, Hepatitis, etc)		Mailing Address: _____ _____	
Specimen type:		Phone No: _____		Fax No: _____	
<input type="checkbox"/> Cheek Swabs; # Swabs: _____		<input type="checkbox"/> Amniotic Fluid		<input type="checkbox"/> Cultured CVS	
<input type="checkbox"/> Peripheral Blood (EDTA); # Tubes: _____		<input type="checkbox"/> Direct CVS		<input type="checkbox"/> Cultured Amniocytes	
				<input type="checkbox"/> Tumor (specify): _____	

BILLING INFORMATION

BILL INSTITUTION: (Please, provide name, address, and telephone number of entity responsible for payment)

Purchase Order Number: _____	Contact Name: _____
Billing address: _____	Phone #: _____
	Fax #: _____
	Email: _____

PAYMENT ENCLOSED:

- Cashier's Check
- VISA® MasterCard® Discover® American Express®

Card Number: _____	Expiration Date: _____
Name as it appears on card: _____	3-digit Security Code: _____
Cardholder Signature: _____	
Cardholder Email Address: _____	

BILL CONTRACTED INSURANCE COMPANY:

Please include a copy of patient's insurance card. For a list of our contracted insurance companies, please visit our website at www.genetics.uab.edu/medgenomics, under "Billing". Please include a copy of pre-approval statement if payment has been authorized. **We also need the patient's credit card information so that any balance left after insurance pays may be applied to it.**

FILE INSURANCE CLAIM WITH NON CONTRACTED COMPANY

Patient must pay full payment for test up front, (credit card or cashier's check) but UAB will file a claim for reimbursement with the patient's insurance company. Please send a copy of patient's insurance card, front and back.

Informed Consent for INI1/SMARCB1 Testing

I, _____, hereby agree to participate in testing for INI1/SMARCB1 using a RNA/DNA-based cascade of tests. I understand that biological samples (blood, cheek cells) will be removed from me using standard techniques which carry very little risk. In addition, if prenatal diagnosis is being performed, fetal cells obtained by chorionic villus sampling or amniocentesis will be used. I understand that the blood, cheek cells or fetal samples will be used for the purpose of attempting to determine if I and/or members of my family are carriers of the disease gene. In addition I hereby give permission to collect biological samples from my minor children, named below, to be used for RNA/DNA-testing for the disease listed above.

Child's name	Date of Birth	Gender (F/M)
_____	_____	_____

I understand that:

1. the techniques used for the mutation analysis of the INI1/SMARCB1 gene allow to identify nonsense, frameshift, missense, splice mutations and copy number changes (deletions or duplications of part or the entire gene) . The test is >99% accurate to detect these mutations when present in non-mosaic state. Rare variations in the DNA of individuals can sometimes be found and can cause uncertainty in predicting the carrier status.
2. 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 our current lack of knowledge of the complete gene structure or an inability of the current technology to identify certain types of mutations in the gene. I have been informed of the likelihood of finding a mutation in the gene for which I am being tested. ____
_____(Initials)
3. I understand that the RNA/DNA INI1/SMARCB1 analysis performed by the Medical Genomics Laboratory is specific for this disease and in no way guarantees my health or the health of my living or unborn children. The Medical Genomics Laboratory cannot be responsible for erroneous clinical diagnosis made elsewhere.
4. In order to perform accurate prenatal diagnosis, biological samples are required from the fetus as well as from the affected individual in the family and from the biological mother.
5. This test is relatively new and is being expanded and improved continuously. The test is not considered research, but is considered the best and newest laboratory service that can be offered. This testing is complex and utilizes specialized materials so that 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. My signature below acknowledges my voluntary participation in this test, but in no way releases the laboratory and staff from the Medical Genomics Laboratory from their professional and ethical responsibility to me.
6. I understand that my sample is not being banked. The laboratory does not return DNA samples to individuals or physicians. However, 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.
7. Once my test result is completed, an aliquot of my DNA/RNA may be made anonymous (name and all other identifiers removed) and used for research purposes. I understand declining to participate in this research will in no way negatively impact my clinical testing or treatment.

PLEASE CHECK ONE OF THE FOLLOWING

- By checking this box, I indicate my desire to opt out of participation in anonymized research studies using my DNA/RNA sample.
8. Because of the complexity of RNA/DNA based testing and the important implications of the test results, results will only be reported to me through a physician, genetic counselor or certified genetics professional. The result reports are confidential and will only be released to other medical professionals or other parties with my express written consent. All laboratory data and personal information are strictly confidential and will not be released from the UAB Medical Genomics Laboratory in full compliance with HIPPA. Participation in RNA/DNA testing is completely voluntary.
 9. I will receive a copy of this consent form.

Signature: _____
Witnessed by: _____
Date: _____

Physician's/Counselor's statement: I have explained RNA/DNA testing to this individual. I have addressed the limitations outlined above and have answered person's questions.

Signature: _____

INI1/SMARB1 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

SCHWANNOMATOSIS-related phenotypic checklist: to be completed if schwannomatosis is suspected

CNS

- Vestibular nerve (8th nerve) dysfunction
 - no evidence of vestibular nerve tumor by MRI (Age: yrs)
 - lack of symptoms of vestibular nerve tumor, but no MRI done (Age: yrs)
 - unknown

- Meningiomas Age of Symptoms: _____
If yes, please provide detail on location:

- Spinal schwannomas: Absent by MRI Present, asymptomatic Present, symptomatic Unknown
Provide location of spinal tumors: C__ to C__, T__ to T__, L__ to L__
Histopathologically confirmed? yes or no

- Cranial nerve involvement Age of Symptoms: _____
If yes, please provide further details:

SKIN

- CAL spots Age of Symptoms: _____
 - 1-5 ≥6-99 >100

- Schwannomas Age of Symptoms: _____
Please provide detail on location of schwannomas

Schwannomas present only in an anatomically limited distribution (single limb or segment of the spine): yes or no
Family history: Sporadic Familial Unknown

INI1/SMARB1 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):

Location of the tumor: brain
 kidney
 spine
 cerebral spinal fluid
 other: specify:

Result INI1-staining on the tumor specimen: abnormal (no INI1-staining)
 normal (INI1 staining)
 unknowns

Family history: Sporadic Familial Unknown
If familial: specify location and type of tumors