

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

**UAB MEDICAL GENOMICS LABORATORY**  
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 Birmingham, Alabama 35294-0005 Fax: (205) 996-2929  
[www.genetics.uab.edu/medgenomics](http://www.genetics.uab.edu/medgenomics)

**PTEN TEST REQUEST FORM**

<b>THIS FORM AND PHENOTYPIC CHECKLIST MUST BE FILLED OUT COMPLETELY</b>				
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):		
<b>PTEN MUTATION ANALYSIS TESTING</b>		Physician's Name:		
<input type="checkbox"/> Sequence of the entire coding region and core promotor (Tier 1) and Copy Number Analysis (Tier 2) as needed (Test 1 comprehensive) <input type="checkbox"/> by RNA <input type="checkbox"/> by DNA <input type="checkbox"/> Sequence of the entire coding region and core promotor <i>only</i> (Test 1-Tier 1) <input type="checkbox"/> by RNA <input type="checkbox"/> by DNA <input type="checkbox"/> Copy Number Analysis by MLPA <i>only</i> (Test1-Tier2) <input type="checkbox"/> Core promoter sequence analysis <i>only</i> <input type="checkbox"/> Targeted Mutation Analysis (Test 2) Proband _____ <input type="checkbox"/> Prenatal Targeted Analysis (Test 3) Proband _____		Physician's Address: _____ _____ _____  Phone: _____ Fax: _____ NPI Number: _____ Email: _____		
Facility where specimen obtained:		Date:		
<b>REQUIRED</b> Diagnosis (ICD-9) Code (only in US):		ADDITIONAL REPORTS TO: Name: _____ Mailing Address: _____ _____ Phone No: _____ Fax No: _____		
Is Patient Pregnant? <input type="checkbox"/> No <input type="checkbox"/> Yes If yes, date of LMP:	Please check if applicable: <input type="checkbox"/> Infectious diseases (AIDS, Hepatitis, etc)			
Specimen type:		<input type="checkbox"/> Amniotic Fluid <input type="checkbox"/> Cultured Amniocytes <input type="checkbox"/> Cheek Swabs; # Swabs: <input type="checkbox"/> Direct CVS <input type="checkbox"/> Cultured CVS <input type="checkbox"/> Peripheral Blood (EDTA); # Tubes: <input type="checkbox"/> Other:		

**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](http://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 PTEN Testing**

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I, \_\_\_\_\_, hereby agree to participate in testing for *PTEN* 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. Approximately 80% of individuals who meet the diagnostic criteria for Cowden syndrome (CS), ~65% of individuals with a clinical diagnosis of Bannayan-Ruvalcaba-Riley Syndrome (BRRS), ~50% of individuals with a Proteus-like syndrome, and ~20% of individuals with Proteus syndrome have a germline mutation in the *PTEN* gene identifiable by sequencing of all coding exons. Additionally, 1-16% of individuals with Macrocephaly/Autism syndrome have a germline mutation in the *PTEN* gene identifiable by sequencing. Approximately 10% of individuals with CS who do not have an identifiable *PTEN* mutation in the coding exons, have a point mutation in the *PTEN* core promoter region. Of individuals with BRRS, ~11% have a partial or complete *PTEN* gene deletion that will be detected by MLPA.
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 *PTEN* 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. A. 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. Any results obtained could not be related to the original source, so no results would be reported.  
B. I indicate my desire to opt out of participation in anonymized research studies using my DNA/RNA sample by checking this box
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 are confidential and will not be released from the Medical Genomics Laboratory. 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: \_\_\_\_\_



Patient ID: \_\_\_\_\_ Date of Birth (MM/DD/YY) \_\_\_/\_\_\_/\_\_\_  
 Referring Physician \_\_\_\_\_ Date of Exam (MM/DD/YY) \_\_\_/\_\_\_/\_\_\_

**DEMOGRAPHIC INFORMATION**

Gender:  Male  Female  
 Ethnicity: Mother -  White  Black  Native American  Hispanic  Asian  Other: \_\_\_\_  
 Father -  White  Black  Native American  Hispanic  Asian  Other: \_\_\_\_

**CLINICAL DIAGNOSIS**

- Cowden syndrome (CS)
- Proteus syndrome (PS)
- Macrocephaly/Autism syndrome
- Bannayan-Riley-Ruvalcaba syndrome (BRRS)
- Proteus-like syndrome
- Other \_\_\_\_\_

**FAMILY HISTORY**

Sporadic  Familial  Unknown  
 Familial cases: Please provide pedigree and details on the affection status of family members on a separate page.

**General Information**

Weight \_\_\_\_ kg or \_\_\_\_ lb      Height \_\_\_\_ cm or \_\_\_\_ ft      Head circumference \_\_\_\_ cm

**CNS**

- Macrocephaly (HC > 98th percentile)
- Mental retardation/Developmental delay  
IQ \_\_\_\_\_
- Autism/ pervasive developmental disorder
- Lehermitte-Duclos disease (LDD)
- Other CNS tumors \_\_\_\_\_

**Skin/Mucosa**

- Facial trichilemmomas  
 0-2 lesions       ≥ 3 lesions
- Cutaneous facial papules
- Oral mucosal papillomatosis
- Acral keratosis  
 0-5 lesions       ≥ 6 lesions
- Lipomas
- Regional absence of fat
- Fibromas
- Pigmented macules of the glans penis
- Connective tissue nevi
- Epidermal nevus
- Vascular malformations  
 Capillary     Venous     Lymphatic
- Other skin lesions \_\_\_\_\_

**Breasts**

- Fibrocystic disease
- Breast cancer (type) \_\_\_\_\_  
BRCA testing?  No  Yes  
If yes, was mutation identified?  
 Yes \_\_\_\_\_  No

**Thyroid**

- Thyroid cancer (non-medullary)
- Benign multinodular goiter
- Adenomatous nodules and follicular adenomas
- Other thyroid lesions \_\_\_\_\_

**Gastrointestinal tract**

- Hamartomatous intestinal polyps
- GI malignancy (specify) \_\_\_\_\_

**Genitourinary**

- Benign uterine fibroids
- Endometrial cancer
- Renal cell cancer
- Bilateral ovarian cystadenomas
- Genitourinary malformation (specify) \_\_\_\_\_

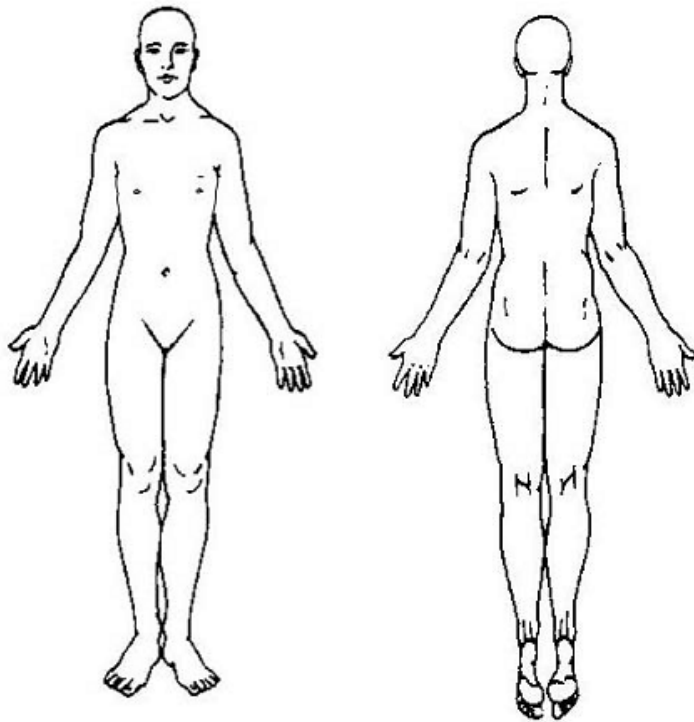
**Disproportionate overgrowth**

- Limbs (arms/legs, hands/feet/digits)
- Skull (hyperostoses)
- Vertebrae (megaspondylodysplasia)
- Viscera (spleen/thymus)

**Others**

- Parotid monomorphic adenoma
- Facial dysmorphism (specify) \_\_\_\_\_

**\*\* Please draw location/size of cutaneous lesions**



**Family pedigree – in familial cases**

