

PTEN-RELATED DISORDERS

Target Mutation Analysis - **Test 2**

- updated 08-10-09

DESCRIPTION

Mendelian Inheritance in Man number: [*601728](#) (*PTEN* gene); [158350](#) (Cowden syndrome, CS); [153480](#) (Bannayan-Riley-Ruvalcaba syndrome, BRRS); [176920](#) (Proteus syndrome, PS); [605309](#) (Macrocephaly/Autism syndrome)

Click here for [Gene Reviews](#) Clinical Summary.

Please see [PTEN Test 1](#) description for detailed clinical information

INDICATIONS FOR DIRECT TESTING

- Identification of pre-symptomatic carriers of a *PTEN*-associated mutation among family members of patients.
- Individuals who want to prepare for prenatal / pre-implantation diagnosis

TESTING METHODOLOGY

We offer a **targeted detection** of a previously characterized *PTEN* mutation within the family. Depending on the mutation identified previously in the family, targeted testing can involve direct sequencing of a specific region or copy number analysis by MLPA.

Test 2 is provided **free of charge** to all relevant relatives of a proband in whom a novel **missense** alteration was found that needs further clarification to come to a final conclusion. As the final conclusion on the pathogenicity of a missense alteration relies on accurate phenotypic data, the testing in relevant relatives is provided free of charge only if a phenotypic checklist is filled out by a healthcare professional that made the clinical assessment of the relatives. The correct interpretation of the results also relies on the correct disclosure of the biological relationships.

SPECIMEN REQUIREMENTS

We require 1 milliliter of whole blood. Blood samples must be collected in EDTA (purple topped) tubes.

TRANSPORT

If specimen is from clinics within UAB or Kirklin Clinic, please call 934-5562 for pick-up. If specimens are being sent from some other location, please ship via UPS or Federal Express.

1. Be sure that the shipping air bill is marked "**Priority**", either Domestic or International.
2. Specimens must be packaged to prevent breakage and absorbent material must be included in the package to absorb liquids in the event that breakage occurs. Also, the package must be shipped in double watertight containers (e.g. a specimen pouch + the

shipping companies Diagnostic Envelope). **You can use our collection kits, which we will send to physicians directly upon request.**

TURN AROUND TIME

2-3 weeks

CPT CODES AND PRICES

Please note that prices listed correspond to institutional rates; please contact the lab for insurance rates.

\$250, -USD ([currency converter](#))
83891 (x1), 83894 (x4), 83898 (x4), 83904 (x3), 83912 (x1)

REQUIRED FORMS

[PTEN Test Requisition including the phenotypic data form](#)
[Form for customs \(International shipment\)](#)

Note: Requests for Molecular Genetic testing for *PTEN* will **not** be accepted for the following reasons:

- No label (patient's full name and date of collection) on the specimens
- No referring physician's or genetic counselor's names and addresses
- No billing information
- No informed consent
- No phenotypic checklist
- **No phenotypic checklist:** we offer **free of charge** targeted testing to all relevant relatives of a proband in whom a **novel missense variant** was identified. Testing of these relatives may allow us to make a final conclusion on the pathogenicity of the novel missense variant and allow us to provide better counseling now and in the future. Free of charge targeted testing will only be provided if the necessary **phenotypic information on the proband and relatives filled out by a healthcare professional** accompanies the samples. If no phenotypic information is provided, we will charge the institution for the test.

For more information, test requisition forms, or sample collection and mailing kits, please call: 205-934-5562.

REFERENCES

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