

# PTEN-RELATED DISORDERS

Prenatal Detection of Known Mutation - **Test 3**

- 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

- Prenatal predictive testing for individuals at risk of inheriting an already known *PTEN* mutation

## TESTING METHODOLOGY

We offer a **targeted detection** of a previously characterized *PTEN* mutation within the Family. For prenatal diagnosis, fresh or cultured CVS or amniocytes can be used for diagnostic purposes. DNA is extracted directly, the target region is amplified and the sequence is analyzed for the presence or absence of the specific mutation. Maternal cell contamination is analyzed by amplification of microsatellite markers in the DNA of the maternal and fetal sample. All prenatal samples are performed in duplicate and independently by two technicians.

## SPECIMEN REQUIREMENTS

- (1) minimum of 15 mg of chorionic villus specimen. Send specimen in transport media in 15-mL centrifuge tube.
- (2) 20 mL of amniotic fluid. Send specimen refrigerated, but not frozen.
- (3) 2-T25 flasks of cultured CVS (>70% confluent), sent at ambient temperature.
- (4) 2-T25 flasks of cultured amniocytes. (>70% confluent), sent at ambient temperature. Please also send 1-5 ml of blood or buccal swab sample from the mother for maternal contamination studies.

## 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.**

3. Please contact us (Email – [mgl@genetics.uab.edu](mailto:mgl@genetics.uab.edu), Phone – 205-934-5562) prior to sending a sample for prenatal testing and provide us with the date of shipment and the tracking number of the package, so that we can better ensure receipt of the samples within the 60-hour window

## TURN AROUND TIME

6 business days after sample is received

## CPT CODES AND PRICES

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

\$750, -USD ([currency converter](#))

83891 (x3), 83894 (x20), 83896 (x6), 83898 (x20), 83904 (x8), 83912 (x1)

## REQUIRED FORMS

### [PTEN Test Requisition](#)

\*Phenotypic checklist does not need to be filled out for prenatal tests.

### [Form for customs \(International shipment\)](#)

**Note:** Requests for Molecular Genetic testing for *PTEN-test 3* will **not** be accepted for the following reasons:

- No label (patients full name and date of collection) on the specimens
- No referring physician's or genetic counselor's names and addresses
- No billing information if being paid for by an institution
- No payment if being paid for by an institution
- No informed consent

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

## REFERENCES

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