NEUROFIBROMATOSIS Type 2

Prenatal Detection of Known Mutation - **Test 3** - updated 08-10-09 -

DESCRIPTION

Mendelian Inheritance in Man number: 101000

Click here for Gene Reviews Clinical Summary.

Bilateral Acoustic Neurofibromatosis, Central Neurofibromatosis
Neurofibromatosis type 2 is characterized by bilateral vestibular schwannomas with associated symptoms of tinnitus, hearing loss and balance dysfunction. Other findings include meningiomas of the brain, schwannomas of other cranial nerves or of the dorsal roots of the spinal cord and juvenile posterior subcapsular cataract. NF2 is an autosomal dominant disorder with a frequency of 1:33-40,000 births in all populations. About 50% of patients are due to a de novo mutation, where neither parent has signs of the disorder. The offspring of an affected individual have a 50% risk of inheriting the altered *NF2* gene. For more information on the condition please refer to the review on the GeneTests website and Online Mendelian Inheritance in Man.

INDICATIONS FOR TESTING

- Prenatal analysis for pregnancies at risk of inheriting an already known *NF2* mutation

TESTING METHODOLOGY

We offer a **targeted detection** of a previously characterized *NF2* mutation within the family. For prenatal diagnosis, fresh or cultured CVS or amniocytes can be used for diagnostic purposes. DNA is extracted directly and the target region is amplified and 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 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, 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), 83909 (x16), 83894 (x4), 83896 (x6), 83898 (x20), 83904 (x8), 83912 (x1)

REQUIRED FORMS

NF2 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 *NF2*-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

Baser M, Friedman J, Aeschliman D, Joe H, Wallace A, Ramsden R, Evans DG - Predictors of the risk of mortality in neurofibromatosis 2. Am J Hum Genet 71:715. 2002 (pubmed)

Evans DG, Ramsden RT, Gokhale C, Bowers N, Huson SM, Wallace A Should NF2 mutation screening be undertaken in patients with an apparently isolated vestibular schwannoma? Clin Genet. 71 (4): 354-8, 2007 (pubmed)

Kluwe L, Nygren A, Errami A, Heinrich B, Matthies C, Tatagiba M, Mautner V Screening for large mutations of the NF2 gene. Genes Chromosomes Cancer 42:384, 2005 (pubmed)