

NEUROFIBROMATOSIS TYPE 2

Target Mutation Analysis - **Test 2**

- 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.

INDICATIONS FOR TESTING

- Predictive testing for individuals at risk of inheriting an already known familial *NF2* mutation

TESTING METHODOLOGY

We offer a **targeted detection** of a previously characterized *NF2* mutation within the family. From a fresh EDTA blood sample, DNA is extracted directly and the target region is amplified and analyzed for presence or absence of the specific mutation.

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 5 milliliters of whole blood. Blood samples must be collected in EDTA (purple topped) tubes. For pediatric patients or those for whom venipuncture is very difficult, please send a minimum of 3 ml in EDTA.

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

[NF2 Test Requisition including the phenotypic data form](#)
[Form for customs \(International shipment\)](#)

Note: Requests for Molecular Genetic testing for NF2 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
- 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

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](#))