

NEUROFIBROMATOSIS Type 1

Target Mutation Analysis - **Test 2**

- updated 08-10-09 -

DESCRIPTION

Mendelian Inheritance in Man number: [162200](#)

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

Neurofibromatosis type 1 is a completely penetrant, autosomal dominant disorder with a frequency of 1/3500 births in all ethnic populations. NF1 is a progressive disorder, characterized by multiple café-au-lait spots, neurofibromas, and Lisch nodules, although additional features may develop. NF1 is notorious for its variable expression. About 50% of cases are due to new dominant mutations, where neither parent has signs of the disorder. An affected individual has a 50% risk of transmitting NF1 to each offspring, although the degree of severity can differ from person to person, even within the same family.

INDICATIONS FOR DIRECT TESTING

- Predictive testing for individuals at risk of inheriting an already known *NF1* mutation
- Individuals who want to prepare for prenatal / pre-implantation diagnosis
- Targeted testing of all relevant relatives of a proband in whom a **novel missense variant** was identified.

TESTING METHODOLOGY

We offer a **targeted detection** of a previously characterized *NF1* mutation within the family. Depending on the mutation identified previously in the family, targeted testing can involve FISH analysis, 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 934-7107 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

[NF1 Test Requisition including the phenotypic data form](#)
[Form for Customs \(International shipment\)](#)

Note: Requests for Molecular Genetic testing for *NF1* 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 if this is a fee for service test
- No informed consent
- **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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