

# NEUROFIBROMATOSIS Type 1

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

- 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

- Prenatal predictive testing for individuals at risk of inheriting an already known *NF1* mutation

## TESTING METHODOLOGY

We offer a **targeted detection** of a previously characterized *NF1* 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 the mother for maternal contamination studies.

## 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.**
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.

## 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

### [NF1 Test Requisition](#)

\*Please note that phenotypic checklist is not required for prenatal testing.

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

**Note:** Requests for Molecular Genetic testing for *NF1*-test3 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

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

## **REFERENCES**

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