
Lab Dept: **Anatomic Pathology**

Test Name: **NEUROFIBROMATOSIS TYPE 2 (NF2) KNOWN MUTATION**

General Information

Lab Order Codes: NF2K

Synonyms: NF2 known mutation; Neurofibromatosis Known Mutation

CPT Codes: 81403 – Molecular pathology procedure, Level 4 (analysis of single exon by DNA sequence analysis, analysis of >10 amplicons using multiplex PCR in 2 or more independent reactions, mutation scanning or duplication/deletion variants of 2-5 exons)

Test Includes: A targeted detection of a previously characterized NF2 mutation within the family.

Logistics

Test Indications: NF2 Targeted Mutation detection testing is for individuals at risk of inheriting an already known familial NF2 mutation.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: University of Alabama Medical Genomics (UAL Test: KT2)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 15 days

Special Instructions: A completed [requisition form](#) and informed consent with a phenotypic checklist must accompany each sample. For questions regarding the forms, please call 205-934-5562.

Samples collected on Friday before 1400 can be shipped for Saturday delivery with special arrangements. Friday after 1400, Saturday/Sunday and holiday collections, will be held in the lab and shipped on Monday, or next business day.

NOTE: Detailed and accurate completion of the requisition is necessary for reporting purposes. The Medical Genomics Laboratory issues its clinical reports based on the demographic data provided by the referring institution

on the lab requisition form. It is the responsibility of the referring institution to provide accurate information. If an amended report is necessary due to inaccurate or illegible documentation, additional reports will be drafted with charge.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	6 mL (Minimum: 3 mL) whole blood must be in EDTA (Lavender) tubes
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection
Special Processing:	<p>Lab Staff:</p> <ol style="list-style-type: none">1. Do Not centrifuge. Send whole blood at room temperature.2. DO NOT SHIP ON ICE.3. Include completed forms and requisition.4. Be sure the shipping air bill is marked "Priority", Domestic.5. 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 <p>Shipping:</p> <p>Monday- Thursday, ship specimen as priority with proper forms, at ambient temperature via overnight courier.</p> <p>Friday before 1400 specimens can be shipped at ambient temperatures for Saturday delivery. Call the University of Alabama Genomics lab (205-934-5562) for special instructions.</p> <p>Friday after 1400, Saturday or Sunday and holidays specimens should be held in the lab at ambient temperatures and shipped ambient on Monday or the next business day (Monday-Thursday).</p> <p>Note: Blood collections are stable for 1 week after collection.</p>
Patient Preparation:	None
Sample Rejection:	Requests for Molecular Genetic testing 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 Phenotypic checklist form; Mislabeled or unlabeled specimens; Incorrect specimen type; Specimen frozen; Mislabeled specimens

Interpretive

Reference Range: Interpretive report

Critical Values: N/A

Limitations: 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. Testing of these relevant relatives may allow a final conclusion on the pathogenicity of the novel missense variant and allow better counseling now and in the future.

Methodology: A targeted mutation of a previously characterized NF2 mutation within the family. DNA is extracted directly and the target region is amplified and analyzed for presence or absence of the specific mutation.

References: [University of Alabama Medical Genomics Laboratory](#) December 2023

Updates:
1/13/2013: CPT update
6/13/2018: Updated collection and shipping information
12/18/2023: Updated turnaround time.