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 – NF2 Known family variant, DNA sequence analysis, each variant exon (Molecular Pathology Level 4)
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 - Sendout
Referred to:	University of Alabama, Medical Genomics Laboratory, Dept of Genetics (UAL test: KT2)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours (See Special Instructions)
Turnaround Time:	10 days
Special Instructions:	Requests must include request form with referring or genetic counselor's name and address, billing information, completed informed consent and phenotypic checklist. All forms can be found at NF2 Req form.pdf. Targeted testing to all relevant relatives of a proband in whom a novel missense variant was identified is performed free of charge. 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, there will be charges to the test. Monday – Thursday collections are preferred. 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 this document 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: Lab Staff:

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

Shipping:

Monday- Thursday, ship specimen as priority with proper forms, at ambient temperature via overnight courier.

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.

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).

Note: Blood collections are stable for 1 week after collection.

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 June 2018 (205) 934-5562 Fax (205) 996-2929
Updates:	1/13/2013: CPT update 6/13/2018: Updated collection and shipping information