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**Lab Dept:** Anatomic Pathology

**Test Name:** NEUROFIBROMATOSIS TYPE 1 (NF1) KNOWN MUTATION

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***General Information***

**Lab Order Codes:** NF1F

**Synonyms:** Von Recklinghausen disease

**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:** Targeted detection of a previously characterized NF1 mutation within the family.

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***Logistics***

**Test Indications:** NF1 is characterized by multiple café-au-lait spots, Lisch nodules and cutaneous neurofibromas. Other complications such as plexiform neurofibromas, learning disabilities, seizures and malignancies may occur. The phenotype of NF 1 is variable both within and between families. Patients with deletions of the entire gene typically have a more severe presentation including dysmorphic facial features, early onset and large number of neurofibroma and mental retardation. NF1 is an autosomal dominant disorder with a very high rate of new mutations. 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.

**Lab Testing Sections:** Anatomic Pathology - Sendouts

**Referred to:** University of Alabama Medical Genomic Laboratory (UAL test: KT2)

**Phone Numbers:** MIN Lab: 612-813-6280

STP Lab: 651-220-6550

**Test Availability:** Daily, 24 hours

**Turnaround Time:** 15 working 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 [Forms](#). 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.

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.

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### ***Specimen***

<b>Specimen Type:</b>	Whole blood
<b>Container:</b>	Lavender (EDTA) top tube
<b>Draw Volume:</b>	6 mL (Minimum: 3 mL) whole blood <b>must be</b> in EDTA (Lavender) tubes
<b>Processed Volume:</b>	Same as Draw Volume
<b>Collection:</b>	Routine venipuncture. Invert sample gently to mix. Forward immediately to the lab.
<b>Special Processing:</b>	Lab Staff: <ol style="list-style-type: none"><li>1. <b>Do Not</b> centrifuge. Send whole blood at room temperature.</li><li>2. <b>DO NOT SHIP ON ICE.</b></li><li>3. Include completed forms and requisition.</li><li>4. Be sure the shipping air bill is marked "Priority", Domestic.</li><li>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</li></ol>

### **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:** Mislabeled or unlabeled specimens; frozen specimens, contaminated specimens, absence of referring physician and address, absence of billing information, absence of informed consent, absence of phenotypic checklist

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### ***Interpretive***

**Reference Range:** No mutations detected

**Critical Values:** N/A

**Limitations:** Germline mutations in the NF1 gene have been identified in >95% of patients (with sporadic disease as well as those with a positive family history).

**Methodology:** Phytohemagglutinin-stimulated lymphocyte culture, Microsatellite analysis, Protein truncation test (PTT), cycle sequencing and long-range RT-PCR.

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

**Updates:**  
1/11/2007: CPT 2007 update  
5/6/2010: CPT updates  
1/15/2013: CPT updates  
6/13/2018: Updated collection and shipping information  
12/18/2023: Updated turnaround time.