Lab Dept:	**Anatomic Pathology**
Test Name:	**NEUROFIBROMATOSIS TYPE 1 (NF1) KNOWN MUTATION**

General Information

Lab Order Codes:	NF1F
Synonyms:	Von Recklinghausen disease
CPT Codes:	81403 – NF1 Known family variant, DNA sequence analysis, each variant exon (Molecular Pathology Level 4)
Test Includes:	Targeted detection of a previously characterized NF1 mutation within the family.

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 (NF1, Test 2)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	2-3 weeks
Special Instructions:	A completed requisition form with informed consent and phenotypic checklist form must accompany each sample or patient to the laboratory.

Specimen

Specimen Type:	Whole blood
Container:	Lavender (EDTA) top tube
Draw Volume:	1 mL whole blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture. Invert sample gently to mix. Forward immediately to the lab.
Special Processing:	Lab Staff: **Do Not** centrifuge. Specimen must remain in original collection container. Ship specimen ASAP, with proper forms, at ambient temperature via overnight courier Monday – Thursday. Specimens collected Friday-Sunday can be held refrigerated until shipped. Call Medical Genomics at 1-800-499-4363 and inform them a specimen is coming.
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

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 January 2013 (205) 934-5562 Fax (205) 996-2929
Updates:	1/11/2007: CPT 2007 update 5/6/2010: CPT updates 1/15/2013: CPT updates