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
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.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	3 – 6 mL blood
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection
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:	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 March 2018 (205) 934-5562 Fax (205) 996-2929
Updates:	1/13/2013: CPT update