Lab Dept: Anatomic Pathology

Test Name: NEUROFIBROMATOSIS TYPE 2 (NF2) SEQUENCING & DELETION/DUPLICATION

General Information

Lab Order Codes:	NF2U
Synonyms:	NF2 gene analysis, NF2 Sequencing and deletion/duplication
CPT Codes:	81405 -Molecular Pathology Procedure, Level 6 (analysis of 6-10 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 11-25 exons, regionally targeted cytogenomic array analysis) 81406-Molecular pathology Procedure, Level 7 (analysis if 11-25 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 26-50 exons)
Test Includes:	Direct sequencing and MLPA analysis of the NF2 gene.

Logistics

Test Indications:	Neurofibromatosis type 2 is characterized by bilateral vestibular schwannomas with associated symptoms of tinnitus, hearing loss and balance dysfunction. Other findings include meningiomas of the brain, schwannomas of other cranial nerves or of the dorsal roots of the spinal cord and juvenile posterior subcapsular cataract.
Lab Testing Sections:	Anatomic Pathology - Sendouts
Referred to:	University of Alabama Medical Genomic Laboratory (UAL Test: NF2-NG)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	30 working 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 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 EDTA (Minimum pediatric: 3 mL) whole blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture

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 overnight 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: Interpretive report

Critical Values: N/A

Limitations: Using this methodology, mutation detection rate in leukocytes is 90% in non-founder NF2 patients. Mutations detected include truncating mutations (nonsense, frameshift, splicing mutations including deep intronic splice mutations), missense mutations, multi-exon deletions or duplications and total gene deletions. In about 25-30% of founders (simplex cases, patients with unaffected parents), mutations are not detected in blood lymphocytes as a result of somatic mosaicism. Only mutations with mosaicism levels greater than 10% can be detected in lymphocyte DNA (Evans et al.2007). Identification of the majority of mosaic mutation requires testing of tumor tissue (Evans et al.2007).

Methodology:	A direct test using cDNA-based direct sequencing of the entire coding region and MLPA analysis to detect copy number changes. Copy number changes are confirmed by long range RT-PCR, quantitative PCR and/or a CGH.
References:	University of Alabama Medical Genomics Laboratory December 2023
Updates:	1/15/2013: CPT update 1/11/17: Draw volume and CPT update. 12/18/2023: Updated turnaround time, blood volumes and CPT code. Clarified test availability and special instructions by removing outdated collection restrictions.