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 – NF2 (neurofibromin 2 [merlin]) (eg, neurofibromatosis type 2), deletion/duplication analysis (Molecular Pathology Level 6)
81479 – NF2 microsatellite analysis (Molecular Pathology Unlisted)

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 Genomics Laboratory, Dept of Genetics (UAL) (NF2-NG)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Monday - Thursday

Turnaround Time: 4 – 5 weeks, specimens are batched and performed once or twice weekly

Special Instructions: Restricted draw times. See Test Availability. 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.

Specimen

Specimen Type: Whole blood

Container:	Lavender top (EDTA) tube
Draw Volume:	3 mL (Minimum: 2 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture
Special Processing:	Lab Staff: **Do Not** centrifuge. Specimen should remain in original collection container. Specimen should remain at room temperature and received at reference lab within 60 - 72 hours of collection. Send Monday-Thursday via UPS or Federal Express marked “priority”. 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. Please contact the reference lab prior to sample shipment and provide them with the date of shipment and the tracking number of the package, so that sample delivery can be better ensured within the 60 - 72 hour window.
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:	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 January 2017 (205) 934-5562 Fax (205) 996-2929

Updates: 1/15/2013: CPT update
1/11/17: Draw volume and CPT update.