Lab Dept: Anatomic Pathology

Test Name: NF1 SEQUENCING & DELETION/DUPLICATION

General Information

Lab Order Codes: NF11R

Synonyms: Neurofibromatosis Type 1; NF1 Comprehensive; NF11

CPT Codes: 81408 – Molecular Pathology, Level 9
81479 – Molecular Pathology Unlisted

Test Includes: NF1 Only by NGS involves sequencing as well as deletion/duplication analysis of the entire coding NF1 region plus the alternatively splices exons 9br, 23a and 48a (60 exons total). The test uses a customized and optimized set of Agilent Haloplex captures probes, followed by sequencing of overlapping amplicons within the regions of interest using Illumina sequencing chemistry. Each coding exon plus approximately 50 bp of flanking intronic sequence are simultaneously sequenced. 5' and 3' sequences are not included.

Logistics

Test Indications: Neurofibromatosis type 1 is a completely penetrant, autosomal dominant disorder with a frequency of 1/3500 births in all ethnic populations. NF1 is a progressive disorder, characterized by multiple cafe'-au-lait spots, neurofibromas, and Lisch nodules, although additional features may develop. NF1 is notorious for its variable expression. 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.

● Patients with classic NF1 including the presence of cutaneous neurofibromatosis or Lisch nodules, as no genetic heterogeneity demonstrated so far associated with this phenotype.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: University of Alabama Medical Genomics Laboratory (UAL code: NF1-NG)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 25 working days

Special Instructions:	Include Required Forms: NF1 Test requisition. Please send completed form with the specimen or patient to the laboratory. **NOTE:** Detailed and accurate completion of this document 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:	3 mL (Minimum: 2 mL) whole blood **must be** in EDTA (Lavender) tubes.
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture
Special Processing:	Lab Staff: **IMPORTANT**! Blood samples must be received at reference lab within 60-72 hours from collection. 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 6. Contact U of Alabama medical genomics lab prior to sample shipment and provide us with the date of shipment and the tracking number of the package, so that they can better ensure the receipt of the samples within the 60-72 hour window. 7. If you wish to ship a sample on Friday for Saturday delivery, you must contact the lab by 2:00 CST on Friday to get special Saturday delivery instructions.
Patient Preparation:	None
Sample Rejection:	Requests for Molecular Genetic testing for NF1 will not be accepted for the following reasons: No label (patients full name and date of collection) on the specimens; No referring physician’s or genetic counselor’s names and addresses; No billing information; No Phenotypic checklist form; Mislabeled or unlabeled specimens; Incorrect specimen type; Specimen frozen; Mislabeled specimens

Interpretive

Reference Range:	No mutations found
Critical Values:	N/A
Limitations:	N/A
Methodology:	NGS Sequencing and Deletion/Duplication
References:	University of Alabama Medical Genomics January 2017
Updates:	1/11/17: Reflex to FISH no longer automatically done. CPT update.