Lab Dept: Anatomic Pathology

Test Name: MULTIPLE ENDOCRINE NEOPLASIA TYPE 2B (RET) SEQUENCING

General Information

Lab Order Codes: RET2B

Synonyms: Neuromata, mucosal, with endocrine tumors; Mucosal neuroma syndrome; Wagenmann-Froboese syndrome; Ganglioneuromatosis for the alimentary tract; MEN2B

CPT Codes: 81404 – Molecular Pathology procedure, Level 5

Test Includes: Analysis is performed bi-directional sequencing of the coding exons 15 and 16 of the RET gene. Mutations found in the first of a family to be tested are confirmed by repeat analysis using sequencing, restriction fragment analysis, or another appropriate method.

Logistics

Test Indications: This disorder is characterized by multiple mucosal neuromas of the lips, tongue, and conjunctiva. Patients often have a tall, thin, Marfanoid body habitus. Muscle wasting and weakness are a complication. Gastrointestinal symptoms secondary to ganglioneuromas and colonic diverticula are observed. MEN2B is associated with an increased risk of malignancy, including pheochromocytoma (50% of patients) and medullary thyroid carcinoma (100%).

Reasons for referral:
1. Confirmation of a clinical diagnosis
2. Genetic counseling
3. Identification of at-risk family members
4. Development of a screening program for the associated malignancies

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx, Inc. (GDX#: 178)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours. Specimens collected Saturday or Sunday will be held for shipment on Monday.

Turnaround Time: 3 weeks for new patients
Special Instructions: A GeneDx signed request form must be sent with any patient or specimen to the laboratory.

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume: 2 - 5 mL (Minimum: 1 mL) blood

Processed Volume: Same as Draw Volume

Collection: Routine venipuncture for blood specimens, invert gently to mix

Special Processing: Lab Staff: Send whole blood in original collection container labeled with patient name, date of birth and medical record number, including signed consent form and requisition, with a cool pack in warm weather, via overnight or second-day courier so that the sample will arrive at GeneDx Monday through Saturday. Samples drawn on Saturday or Sunday should be held at refrigerated temperatures for shipment on Sunday. Do not freeze.

Note: Specimens may be stored at refrigerated temperatures for up to 7 days prior to shipping.

Patient Preparation: None

Sample Rejection: Unrefrigerated specimens older than 48 hours; clotted or hemolyzed for blood; frozen specimens; mislabeled or unlabeled specimens

Interpretive

Reference Range: No mutations detected

Critical Values: N/A

Limitations: N/A

Methodology: Capillary sequencing

References: GeneDx, Inc. March 2018
(309) 519-2100 Fax (319) 519-2892

Update: 2/7/2013: CPT update