
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 (Preferred draws are Sunday - Thursday as specimens can only be received at the reference lab Monday - Friday. Specimens collected Friday or Saturday will be held for shipment on Monday.)

Turnaround Time: 4 - 5 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: 1 - 5 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, Inc. on a weekday (Monday through Friday). Samples drawn on Friday or Saturday 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: Buccal specimens are not acceptable for this testing.

Methodology: Bi-directional sequence analysis

References: [GeneDx, Inc.](#) July 2012
(309) 519-2100 Fax (319) 519-2892

Update: 2/7/2013: CPT update