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**Lab Dept:**                    **Anatomic Pathology**

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

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***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.

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***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.

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### ***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

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### ***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