
Lab Dept: **Anatomic Pathology**

Test Name: **MULTIPLE ENDOCRINE NEOPLASIA TYPE 1
(MEN1) SEQUENCING**

General Information

Lab Order Codes: MEN

Synonyms: MEN1; Multiple Endocrine adenomatosis; MEA1; Werner syndrome; Family Isolated Hyperparathyroidism

CPT Codes: 81405 – Molecular Pathology procedure, Level 6

Test Includes: Analysis is performed bi-directional sequencing of the coding regions and splice sites exons 2-10 of the MEN1 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: **Multiple endocrine neoplasia type 1 (MEN1)** is characterized by endocrine tumors, particularly in the parathyroid glands, anterior pituitary, and pancreatic islet cells. Primary tumors may be found in more than one endocrine organ and/or multiple tumors may be found in the same organ. The presence of MEN1-associated endocrine tumors causes an array of clinical and biochemical manifestations secondary to hormone hypersecretion: hyperparathyroidism [the most frequent MEN1-symptom with potential central nervous (CNS), hypercalcemia, gastrointestinal, renal cardiovascular, and skeletal involvement), hypercortisolism, gigantism and acromegaly, prolactinoma (with associated oligomenorrhea, amenorrhea, and galactorrhea in females and sexual dysfunction in males), gastrinoma, and insulinoma. Non-endocrine tumors also are common and can include facial angiofibromas and collagenomas of the skin, lipomas, meningioma and ependymoma of the CNS, and skin collagenomas, and leiomyomas. Mutations in the MEN1 gene are highly penetrant; approximately 50% of MEN1 mutation carriers are symptomatic by age 20 and 95% are symptomatic by age 40.

Familial Isolated Hyperparathyroidism (FIHP) is characterized by parathyroid adenoma/hyperplasia (and possibly carcinoma) in the absence of other associated endocrinopathies, is also associated with mutations in the MEN1 gene. However, FIHP is genetically heterozygous and can be cause by mutations in other genes, such as CASR and HRPT2.

Reasons for referral:

1. Confirmation of a clinical diagnosis
2. To differentiate MEN1-related FIHP from other causes
3. Identification of at-risk family members
4. Determination of appropriate surveillance and treatment protocols

- 5. Genetic counseling
- 6. Prenatal diagnosis

Lab Testing Sections:	Anatomic Pathology - Sendouts
Referred to:	GeneDx, Inc. (GDX#: 1761)
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:	6 - 7 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 during warm temperatures, 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
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Critical Values: N/A

Limitations: Buccal specimens are not acceptable for this testing.

Methodology: Bi-directional sequence analysis

References: [GeneDx, Inc.](#) March 2012
(301) 519-2000 Fax (301) 519-2892

Update: 2/7/2013: CPT update