UT Southwestern
Harold C. Simmons
Comprehensive Cancer Center

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MEN1 Mutations (Multiple Endocrine Neoplasia, Type 1)

What You Should Know About *MEN1* Mutations

Individuals with an *MEN1* mutation have a condition called Multiple Endocrine Neoplasia, Type 1 (MEN1). This condition causes a person to develop tumors in their endocrine glands, primarily involving the parathyroid, pancreas and pituitary gland, as well as other tumors. MEN1 can be clinically diagnosed when an individual has at least two of the three main MEN1 endocrine tumors (parathyroid, pituitary, and pancreatic). Individuals who do not meet this definition may also have MEN1.

The Types of Tumors Associated with *MEN1* Mutations

MEN1 is associated with the following risks for endocrine tumors:

- Parathyroid tumors or parathyroid hyperplasia (98% risk by age 50). Tumors that develop in the parathyroid glands in individuals with MEN1 are typically not cancerous, but they can produce excessive amounts of hormone. This causes calcium to be moved from the bone to the blood, which causes the bones to become weak and can cause kidney stones to develop.
- Pituitary tumor (35% lifetime risk)
- Pancreatic gastrinoma (40% lifetime risk)
- Pancreatic insulinoma (10% lifetime risk)
- Carcinoid (bronchial and thymic cancer) (10% lifetime risk)
- Adrenocortical cancer (20-40% lifetime risk)

MEN1 is also associated with non-endocrine tumors, including facial angiomas, collagenomas, and multiple lipomas.

The Risks to Family Members

Mutations in the *MEN1* gene are inherited in an autosomal dominant fashion. This means that first degree relatives (children, brothers, sisters, and parents) of individuals with a *MEN1* mutation have a 1 in 2 (50%) chance of having the mutation as well. Approximately 10% of individuals with MEN1 do not have a family history of the disease, and thus have a new mutation (de novo).

Managing the Tumor Risks

Below are guidelines based on current National Comprehensive Cancer Network (NCCN v1.2019) Clinical Practice Guidelines in Oncology for individuals with MEN1. The following recommendations can help lower the risk of developing a tumor or catch a tumor in the developing stage:

Biochemical Testing

- Annual parathyroid hormone and calcium annually
- Annual Prolactin and IGF-1 hormones every 3-5 years

Imaging

- Brain MRI with contrast, every 3-5 years
- Consider abdominal/pelvic CT or MRI, every 1-3 years
- Consider chest CT or MRI with contrast, every 1-3 years

- Consider serial endoscopic ultrasounds (EUS)

Surgery

- Sub-total or total parathyroidectomy is usually recommended

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