

Genetic Testing: Multiple Endocrine Neoplasia Type 1

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Multiple Endocrine Neoplasia Type 1

Multiple endocrine neoplasia Type 1 (MEN1) is a rare disorder that affects families. It results from a *mutation* (a change) in a gene known as the *MEN1* gene. In people with MEN1, some of the endocrine glands become overactive. Endocrine glands produce substances called *hormones* that regulate various processes in the body such as growth and digestion. MEN1 affects both males and females. It also affects people of all races and occurs throughout the world.

In people with MEN1, there are tumors of one or more of three endocrine glands, and these tumors frequently produce abnormal amounts of hormones. The tumors involve the parathyroid glands, pancreas, or pituitary gland. Most individuals with only one overactive endocrine gland do not have MEN1.

The parathyroid glands, located next to the thyroid gland in the neck, are most commonly involved. Increased production of parathyroid hormone by these tumors (*hyperparathyroidism*) causes the levels of calcium in the body to become high, which can cause kidney stones to form. In patients with very high calcium levels, there may also be fatigue, confusion, pain, and changes in appetite and digestion.

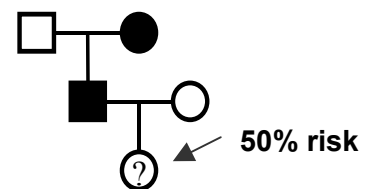
Some of the tumors that develop can be malignant, particularly those that develop in the pancreas (islet cell cancer) or chest (carcinoid tumors). These tumors can *metastasize* (spread) to other organs. Therefore, it is important for people with MEN1 to be checked often to make sure such tumors have not formed. Finding these tumors early can help lower the risk for metastasis. It is also important for individuals with this disorder to be evaluated for excessive hormone production by pituitary, pancreatic islet, and carcinoid tumors. If undetected, these hormones can cause severe diarrhea, stomach ulcers, low blood sugar, abnormalities in menstruation and fertility, low estrogen or testosterone levels, and excessive cortisol or growth hormone production.

Sometimes more than one hormone is produced, creating symptoms that are difficult to interpret. For this reason it is best to be evaluated by a physician with experience with this disorder.

Most people with MEN1 begin to show the symptoms of the disorder in their late teens or early 20s and 30s, but some do not show symptoms until as late as their 50s.

Genetic Testing for MEN1

The diagnosis of MEN1 can sometimes be made on the basis of a person's medical and family history. Other times, it is diagnosed on the basis of genetic testing. Genetic testing involves a search for a mutation in the *MEN1* gene. The test can detect a mutation in about 75% of people with MEN1. If a mutation is found, a doctor can be certain that a person has MEN1. Once a mutation is identified in a family, other close relatives may want to find out whether they also have it. For example, the children of a person with MEN1 have a 50% risk of having the disorder too.



Even though it can be beneficial, genetic testing can cause a person to have unexpected thoughts and emotions. One of the common concerns people have is that they will lose or not be able to get a job or insurance if they are found to have a mutated *MEN1* gene. A genetic counselor or trained healthcare professional can go over such concerns with you to help you and your family make a decision about having genetic testing.

MEN1 Research at M.D. Anderson Cancer Center

Genetic testing for MEN1 is now available at M.D. Anderson at no cost to patients. This is being done as part of a study on MEN1 that will help us learn more about the disorder and help other families with MEN1.

Study Description

Those who are interested in the study will first meet with a member of the study team to determine whether they are eligible for the study and are willing to participate. Then they will meet with a genetic counselor to discuss concerns and learn about MEN1 and genetic testing. Each participant will provide a small blood sample, which will be tested for mutations in the *MEN1* gene. Participants can choose whether they do or do not want to learn their test results. The results will be given in a meeting with a genetic counselor. Although it is best for the test results to be given in person, if participants cannot return to Houston, then results can be given over the telephone along with the same counseling that would be provided in person. A written report of the findings will also be sent to participants who want to learn their test results.

People will also be invited to participate in a related MEN1 questionnaire study. This questionnaire study is open to all people with MEN1 including those who do not want genetic testing.

Eligibility

Participants must be known to have MEN1 on the basis of clinical findings or be suspected of having MEN1.

Healthy participants who wish to have the test must be 1) a blood relative from a family with a known *MEN1* gene mutation and 2) at least 16 years old.

All participants, both healthy and those who are already affected with MEN1, must be registered patients at M.D. Anderson Cancer Center.

Possible Risks & Benefits

Benefits:

A positive result may confirm a suspicious diagnosis or allow for early diagnosis and therefore prompt treatment of the related disease. A negative result in a relative from a family with a known mutation in the *MEN1* gene may eliminate concern about having the disorder and excuse that relative from future medical testing related to MEN1.

Risks:

A bruise may form at the site where blood is drawn. Participants may also feel fear, anger, or depression caused by learning that they have a mutated *MEN1* gene. Because a mutation is only found in about 75% of people with MEN1, it is possible that the test will not identify an existing mutation in as many as 25% of people with MEN1.

A genetic counselor will discuss these potential risks and benefits in more detail before people decide whether to join the study.

Duration

Test results may not be available until several months later.

Those who participate but do not wish to learn their test results do not need to return for additional visits. Genetic counseling and information will be available to all participants for the entire time of the study.

Cost

There is no cost for the genetic test or for the genetic counseling in this study. This does not extend to any other unrelated tests or treatment that participants receive at M.D. Anderson Cancer Center.

Confidentiality

The results from genetic testing will be kept confidential among the researchers in the study and will not be given to a third party or appear in participants' M.D. Anderson medical records.

To Learn More

More information about this and other current research studies in endocrine disease at M.D. Anderson Cancer Center can be found online:

www.mdanderson.org/departments/endocrinesurg

www.mdanderson.org/departments/endocrinology

We also welcome your questions by phone. Please call our recruitment office in Surgical Endocrinology at (713) 745-3179.