Risk of Parathyroid, Pituitary, Stomach, Small Bowel, and Other Tumors

INFORMATION ABOUT YOUR GENETIC TEST RESULT

Your Result

Positive for a known pathogenic or likely pathogenic variant in the *MEN1* gene

What This Result Means

This result means you have a change (mutation) in the *MEN1* gene. Mutations in this gene can cause multiple endocrine neoplasia, type 1 (MEN1). People with MEN1 have an increased chance of certain benign tumors and cancers. These are found mostly in the parathyroid, pituitary, pancreas, and small bowel. Some tumors, called carcinoid tumors, are located in the lungs, thymus, stomach, or pancreas. Adrenocortical tumors are found in the outer layer of the adrenal gland, which sits on top of the kidney. The risk for these tumors is shown in the table below.

This result doesn't mean you will get a tumor, and not everyone with a tumor develops cancer. But you are at higher risk than you would be if you didn't have the mutation.

Type of Tumor or Cancer	Risk of Tumor or Cancer	
	With an MEN1 Mutation	***Without an MEN1 Mutation***
Parathyroid	100%	1%
Pituitary	10 – 60%	Rare
Pancreas, small bowel	50%	Rare
Adrenocortical	20 – 40%	Rare
Carcinoid	10%	Rare
Brain and/or spinal cord	1 – 8%	Rare

Options for Managing Your Cancer Risk

Your test result shows that you have an increased risk for developing tumors and cancer. There are steps you can take to lower that risk. The options are explained briefly below. Talk with your healthcare provider to learn more and find out which is best for you.

Increased Tumor/Cancer Screening

- Parathyroid: annual calcium blood test; if high, PTH and vitamin D blood tests, ultrasound, or other imaging of the neck
- Pituitary: prolactin, IGF-1, and other hormone blood tests; MRI of the pituitary every 3 to 5 years
- Pancreas, small bowel: hormone blood tests, imaging every 1 to 3 years, possible ultrasound
- Lungs and thymus: chest imaging (CT or MRI) every 1 to 3 years

Increased screening cannot prevent you from getting a tumor or cancer. But it could detect them sooner. And, as you know, early detection improves your chances of survival.

Risk of Parathyroid, Pituitary, Stomach, Small Bowel, and Other Tumors

INFORMATION ABOUT YOUR GENETIC TEST RESULT

Quest Diagnostics™

What This Result Means for Your Family

You share genes in common with your blood relatives, so some of them may have the same mutation that you have. Your parents, brothers, sisters, and children each have a 50% chance of having it. Other blood relatives also have an increased risk.

Think about sharing your test results with your relatives. They might want to find out if they have the mutation too. If so, they should talk with their doctor and/or genetic counselor about testing. They could be tested just for the mutation that you have. Testing for a single mutation may be the best option, as it may cost less. It will still let them know if they have the same mutation you have. In some cases, testing more genes might be suggested. Either way, testing will help them know more about their risk for cancer.

Relative	Risk of Having the Same Mutation
Identical twin	100%
Fraternal twin	50%
Parent, brother, sister, child	50%
Grandparent, uncle, aunt, niece, nephew	25%
First cousin	12.5%

Your Next Steps

- ☐ Get a copy of your test results.
- ☐ Talk with your doctor or genetic counselor about things you can do to manage your risk.
- ☐ Once you and your doctor have made a plan, set up appointments to start managing your risk.
- ☐ Think about sharing copies of your test results with your family members. That way, their doctor will know which test to order if they want to be tested.
- ☐ Talk with your doctor or genetic counselor regularly. They can keep you up to date about genetic testing and risk management options.

You can help researchers learn even more about this gene and its link to cancer risk. You can do this by participating in a research registry called PROMPT. If you would like to do this, please visit **PROMPTStudy.org** to learn more.

Additional Resources

- **Association for Multiple Endocrine Neoplasia Disorders (AMEND)**
  AMEND.org.uk/
- **National Society of Genetic Counselors**
  Find a Genetic Counselor
  NSGC.org/p/cm/ld/fid=164

This information is not a substitute for medical advice, diagnosis, or treatment. The diagnosis or treatment of any disease or condition may be based on personal history, family history, symptoms, a physical examination, laboratory test results, and other information considered important by your doctor. Always talk with your doctor about the meaning of your test results and before you stop, start, or change any medication or treatment.

The classification and interpretation of the variant(s) identified reflect the current state of Quest Diagnostics' understanding at the time of the accompanying report. Variant classification and interpretation are subject to professional judgment, and may change for a variety of reasons, including but not limited to, updates in classification guidelines and availability of additional scientific and clinical information. This test result should be used in conjunction with the health care provider's clinical evaluation. Inquiry regarding potential changes to the classification of the variant is strongly recommended prior to making any clinical decision. For questions regarding variant classification updates, please call Quest Diagnostics at 866.GENE.INFO (436.3463) to speak to a genetic counselor or laboratory director.