HUNTSMAN CANCER INSTITUTE UNIVERSITY OF UTAH

About *RET* Gene Mutations

HEALTH UNIVERSITY OF UTAH

Genes are in every cell in our bodies. Genes are made of DNA, which gives instructions to cells about how to grow and work together. We have two copies of each gene in each cell—one from our mother and one from our father. When genes work right, they help stop cancer cells from developing.

About Cancer Syndromes

Sometimes changes to genes can happen. These changes are called mutations. Mutations can make the genes stop working and raise the risk for certain types of cancer. Some mutations can be passed through families. When that happens, family members who have the mutation have a *hereditary cancer syndrome.*

In each cell, there are genes that control how often the cell copies itself. One of these genes is *RET*. When *RET* has a mutation, the cell can copy out of control and cancer can develop. When someone has a *RET* mutation, they get a hereditary cancer syndrome called Multiple Endocrine Neoplasia Type 2 (MEN2) which has two different types: MEN2A and MEN2B. They may also get familial medullary thyroid carcinoma (FMTC).

RET Syndromes and Cancer Risk

MEN2A: About 9 out of 10 people with MEN2A will develop medullary thyroid cancer, usually in early adulthood. They also have a 5 in 10 chance to develop a tumor on the adrenal gland (a gland in the abdomen). Adrenal tumors can lead to high blood pressure.

People with MEN2A can also have high levels of parathyroid hormone or tumors in the neck area (parathyroid) called adenomas. Too much of parathyroid hormone can cause tiredness, weakness, muscle or bone pain, kidney stones, and thin bones.

MEN2B: People with MEN2B have a high chance of getting medullary thyroid cancer. This can happen when they are babies or children. They also have a 1 in 2 chance to develop tumors on the adrenal glands and in the abdomen.

People with MEN2B may be tall and thin and may develop non-cancerous (benign) growths on the tongue, lips, and eyelids before age one or two called mucosal neuromas. Some people also have benign growths in their large intestine (*ganglioneuromatosis)*, which can cause constipation or stomach pain.

FMTC: People with FMTC have a high risk of getting medullary thyroid cancer. The cancer is typically found when they are middle-aged.

Cancer/Tumor Type	Risk with MEN2A
Medullary thyroid carcinoma	9 in 10
Pheochromocytoma	5 in 10
High parathyroid hormone or adenomas	3 in 10
Cancer/Tumor Type	**Risk with MEN2B**
Medullary thyroid carcinoma	10 in 10
Pheochromocytoma	5 in 10
Cancer/Tumor Type	**Risk with FMTC**
Medullary thyroid carcinoma	8 in 10

Recommendations

For people diagnosed with a type of MEN2, the thyroid should be removed before it develops cancer. The recommended age of removal depends on which type of MEN2 the person has:

- MEN2A and FMTC: Before age 5
- MEN2B: Before 6 months of age

Other lifelong tumor screening is also recommended. These blood tests and imaging tests can catch cancer early, when it is easier to treat. These tests will vary by the type of *RET* mutation in your family.

Type of Test	How Often	Starting Age
Bloodwork	Once a year	Age of diagnosis
Blood pressure measurements	Once a year	Age 5
Abdominal MRI or CT Scan	Every 3 years	Age 15

KIDS AND SIBLINGS

Siblings and children of people with a *RET* mutation have a 1 in 2 chance of also having it. Genetic testing is recommended at birth for those with a family history of MEN2B. Children in families with MEN2A and FMTC should get tested at age 3 or 4.

It is important to determine which side of the family carries the *RET* mutation to know which family members are at risk for tumors. A genetic counselor can help you know who in your family should be tested.

Do you have questions about your risk for cancer? Our doctors and genetic counselors can help find the cancer screening plan you need.

Call Huntsman Cancer Institute's Family Cancer Assessment Clinic to learn more: 801-587-9555.

For more patient education information: Call 1-888-424-2100 toll free • Email cancerinfo@hci.utah.edu • Visit www.huntsmancancer.org
Produced by HCI © 2018 • Approved by a team of medical, health, and communications specialists • April 2018 • Review Date April 2021