MASSACHUSETTS GENERAL HOSPITAL
CANCER CENTER

RET gene: What You Need to Know

What does it mean to test positive for a *RET* gene mutation?

Mutations in the *RET* gene cause a condition known as Multiple Endocrine Neoplasia Type 2 (MEN2) syndrome. There are three subtypes of this syndrome: MEN2A, MEN2B and familial medullary thyroid cancer (FMTC) syndrome.

Mutations in the *RET* gene can also cause a different genetic disease known as Hirschsprung disease. It is important to note that individuals with MEN2 rarely develop Hirschsprung disease.

What is my risk for cancer if I have a *RET* gene mutation?

If you have a *RET* gene mutation, you have a greater risk of developing certain types of cancers and benign tumors of the endocrine system. The endocrine system is made up of endocrine glands, which secrete hormones to control important functions in the body such as mood, growth and development. and metabolism.

Benign tumors are not cancerous and do not spread whereas malignant tumors are cancerous and can invade nearby healthy tissue and organs. Cancerous cells have the potential to spread (metastasize) to other sites of the body. Benign and malignant tumors are often treated in different ways.

Lifetime Tumor and Cancer Risks

	MEN2A	MEN2B	FMTC
Medullary thyroid cancer	95-100%	100%	100%
Pheochromocytoma (benign adrenal gland tumor)	50%	50%	Not increased
Primary hyperparathyroidism (parathyroid adenoma)	20-30%	Not increased	Not increased

Some families with *RET* gene mutations may also develop benign tumors of the gastrointestinal tract (ganglioneuromas), lips, and tongue (mucosal neuromas).

Approximately 75% of individuals with MEN2B will have a tall slim body type, hyperflexible joints and scoliosis.

What is the chance that my family members will have a *RET* mutation if I test positive?

There is a 50% chance that a person with a mutation will pass it on to each of his/her children. In most cases, brothers and sisters of a person with a mutation have a 50% chance to have the mutation. Additionally, other family members are at risk to have the mutation.