

# Multiple Endocrine Neoplasia Type 2 (MEN2)

## WHAT IS THE THYROID GLAND?

Your thyroid gland is a butterfly-shaped gland located in the lower front of your neck. The job of your thyroid gland is to make thyroid hormones. Thyroid hormones are released into your blood and carried to every tissue in your body. Thyroid hormones help your body use energy, stay warm and keep your brain, heart, muscles, and other organs working normally.

## WHAT IS MULTIPLE ENDOCRINE NEOPLASIA TYPE 2 (MEN 2)?

MEN 2 (Multiple Endocrine Neoplasia Syndrome type 2) is a group of diseases including a type of thyroid cancer called *Medullary Thyroid Cancer (MTC)*. MEN2 is due to a change (mutation) in a gene called RET. This gene mutation can be found with a blood test for genetic testing. If you have changes in this gene, you [and your family members] are at a higher risk of getting MTC and other tumors.

There are two types of MEN2: MEN 2A and MEN 2B. It is important for your doctor to know the exact change you have in the RET gene to know your risk for getting the diseases in MEN2.

If you are diagnosed with either form of MEN2, you will need life-long monitoring by an endocrinologist. Depending on the diseases identified as part of the MEN2 syndrome, you may need a team of doctors to monitor for these disorders.

MEN 1 is a different condition. It is not associated with thyroid cancer.

## MEDULLARY THYROID CANCER (MTC)

**WHAT IS MTC?** MTC is a less common type of *thyroid cancer*. Only one to two (1-2) out of 100 thyroid cancers are MTC. MTC comes from cells in the thyroid gland called parafollicular cells or C-cells. These C- cells make proteins called “calcitonin” and “carcinoembryonic antigen (CEA).” MTC is a slow growing type of cancer.

## What are the Symptoms of MEN2?

- **IF YOU HAVE MEN 2A, YOU MAY HAVE:**
  - A form of thyroid cancer called medullary thyroid cancer (MTC)
  - Benign (not cancer) growth of the parathyroid glands (primary hyperparathyroidism)
  - Adrenal tumors called pheochromocytoma (PHEO)
  - A skin disease called cutaneous lichen amyloidosis (CLA)
  - A disease of the large intestine called Hirschsprung disease (HD)
- **IF YOU HAVE MEN 2B, YOU MAY HAVE:**
  - Medullary thyroid cancer (MTC) at an early age, even as a child
  - A 50% chance of getting pheochromocytoma (PHEO)
  - Other symptoms, including:
    - Long arms
    - High arch of your foot
    - Swelling of your nerve cells (neuromas or “ganglioneuromatosis”) in the lining of your eyes, mouth, and large intestine. This can cause:
      - Your eyelids to turn out
      - Difficulty making tears
      - Your lips appear swollen and lumpy
      - Your tongue may be thick and lumpy
      - Your large intestine may not work normally causing constipation or diarrhea



# MULTIPLE ENDOCRINE NEOPLASIA TYPE 2 (MEN2)

## WHAT ARE THE SYMPTOMS OF MTC?

Most people with MTC have no symptoms.

- You might feel a lump in your neck from the MTC tumor.
- If the MTC tumor is large, you can have symptoms from the tumor pressing on your windpipe or throat. This might include a hoarse voice, a cough, trouble swallowing or trouble breathing.
- If the MTC tumor has spread, you might have diarrhea or flushing.

## HOW IS MTC DIAGNOSED?

- MTC is diagnosed when a lump is found in your thyroid gland on exam, neck ultrasound or other type of X-ray.
- You might have high levels of calcitonin and/or CEA on a blood test.
- A biopsy of the lump in your thyroid gland can show MTC cells.

## HOW IS MEDULLARY THYROID CANCER TREATED?

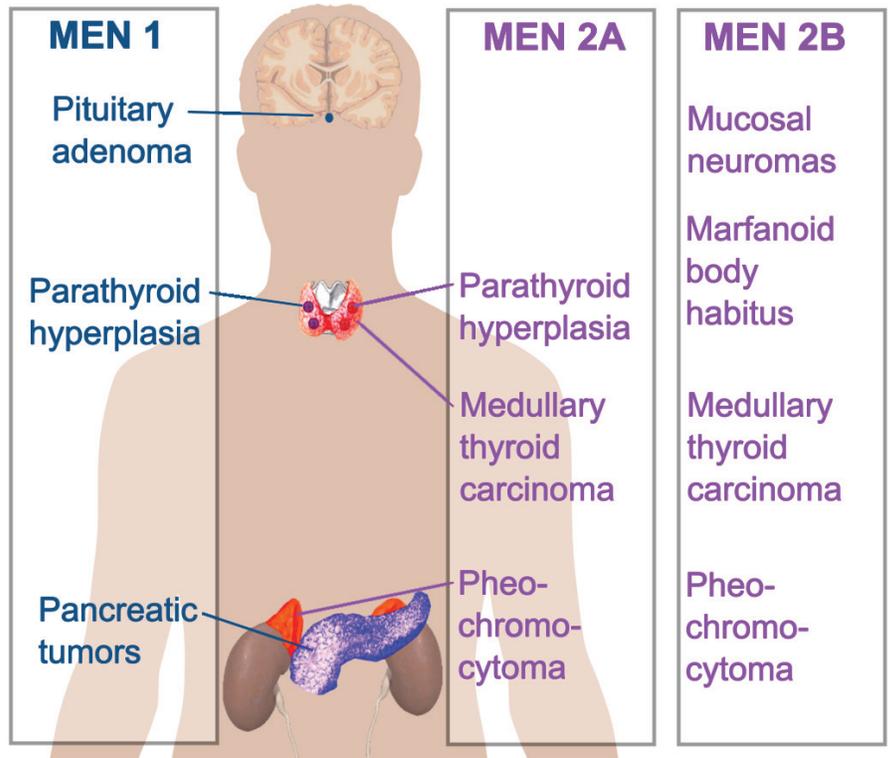
- MTC in your thyroid gland is treated with surgery.
- MTC may spread to the lymph nodes in your neck.
  - These lymph nodes can be removed with surgery
- MTC can spread to other areas of the body such as the lungs, liver, and bones.
  - This can be treated with radiation, chemotherapy pills, and other treatments

## PHEOCHROMOCYTOMA

### WHAT IS PHEOCHROMOCYTOMA?

Pheochromocytoma (PHEO) is a tumor of the adrenal glands.

- Your adrenal glands are small glands that sit above your kidneys, one on each side.
  - Each one of your adrenal glands has an outer and inner part.
  - The outer part (called the “adrenal cortex”) makes “steroid hormones”. The adrenal steroid hormones include cortisol and aldosterone.
  - The inner part (called the “adrenal medulla”) makes adrenaline-type hormones. A PHEO is a tumor of this part of your adrenal gland.



## WHAT ARE THE SYMPTOMS OF PHEOCHROMOCYTOMA?

- Symptoms of a PHEO are due to extra amounts of adrenaline-type hormones, made by the PHEO tumor cells, and can include:
  - Racing or skipping of your heart (“palpitations”)
  - High blood pressure
  - Severe headaches
  - Increased sweating
  - Your skin appears paler than normal

## HOW IS PHEOCHROMOCYTOMA DIAGNOSED?

- Testing for PHEO can include:
  - A urine sample, usually collected for a 24-hour period
  - A blood sample to check for “metanephrines” and “normetanephrines” (adrenaline hormones)
  - A CT scan or MRI of your adrenal glands
- In MEN 2, PHEO is usually a benign tumor and not cancer

# MULTIPLE ENDOCRINE NEOPLASIA TYPE 2 (MEN2)

## HOW IS PHEOCHROMOCYTOMA TREATED?

- Treatment is required because too much of adrenaline-type hormones is harmful to the body
  - You will likely need surgery to remove the tumor
  - Your healthcare team will prepare you for surgery with medications to control your blood pressure.

## PRIMARY HYPERPARATHYROIDISM (PHPT)

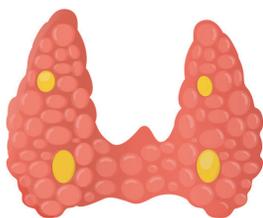
### WHAT IS PRIMARY HYPERPARATHYROIDISM (PHPT)?

PHPT is caused by a benign (non-cancer) tumor (adenoma) or overgrowth (hyperplasia) of one or more of your parathyroid glands. This causes you to produce too much parathyroid hormone (PTH).

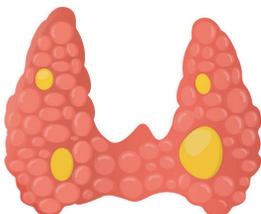
- Most people have four parathyroid glands. They are usually small and sit behind your thyroid gland, two on each side.
- Your parathyroid glands normally make parathyroid hormone (PTH) to prevent your blood calcium level from being too low.

If you make too much PTH from tumors in the parathyroid glands, you can have high calcium levels in your blood.

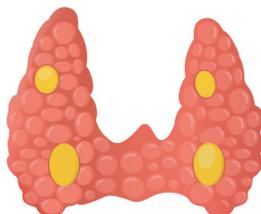
**WHAT ARE THE SYMPTOMS OF PHPT?** Symptoms are caused by the high calcium levels in the blood. These may include irritability, kidney stones, constipation, increased urination, and osteoporosis (low bone strength) leading to broken bones.



Normal parathyroid glands



Parathyroid adenoma:  
a benign tumor of the  
parathyroid gland



Parathyroid hyperplasia  
all glands are enlarged  
and a little overactive

**HOW IS PHPT TREATED?** The condition is treated with surgery.

The prognosis is usually good, but you may have low blood calcium levels after surgery and need calcium medicine to keep calcium levels normal.

## CUTANEOUS LICHEN AMYLOIDOSIS (CLA)

### WHAT IS CUTANEOUS LICHEN AMYLOIDOSIS (CLA)?

This is a rare disease in which a protein called amyloid builds up in your skin.

### WHAT ARE THE SYMPTOMS OF CLA?

- Symptoms of CLA are dark and itchy areas of your skin, usually on or between your shoulder blades.
  - The itchiness may get worse with stress and improve with sun exposure
  - The darkening can get worse with scratching

The skin changes may start before a diagnosis of MTC.

**HOW IS CLA DIAGNOSED?** The diagnosis is made by a skin specialist (Dermatologist).

**HOW IS CLA TREATED?** There are different options that are best discussed by a skin specialist.

## HIRSCHPRUNG DISEASE (HD)

**WHAT IS HIRSCHPRUNG DISEASE (HD)?** HD is caused by not enough nerve cells in your large intestine (the "colon"). The nerve cells are needed to help the muscles work to empty your large intestine.

### WHAT ARE THE SYMPTOMS OF HD?

- HD is present at birth. Symptoms usually show up in infants or young children and can include:
  - Constipation
  - Abdominal pain
  - Vomiting
  - Failure to grow
  - Diarrhea

### HOW IS HD DIAGNOSED?

- Testing for HD can include:
  - X-ray of the abdomen
  - Biopsy (sample of tissue) from your large intestine

### HOW IS HD TREATED?

- Treatment is surgery.

