# Multiple endocrine neoplasia type 2

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# Definition

Multiple endocrine neoplasia is a group of disorders that affect the body's network of hormone-producing glands called the endocrine system. Hormones are chemical messengers that travel through the bloodstream and regulate the function of cells and tissues throughout the body.

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MEN2 is a rare autosomal dominant familial cancer syndrome caused by a mutation in the RET gene located on the tenth chromosome.

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# MEN2 HAS 3 SUBTYPES

01
MEN2A

02
MEN2B

03
FMTC

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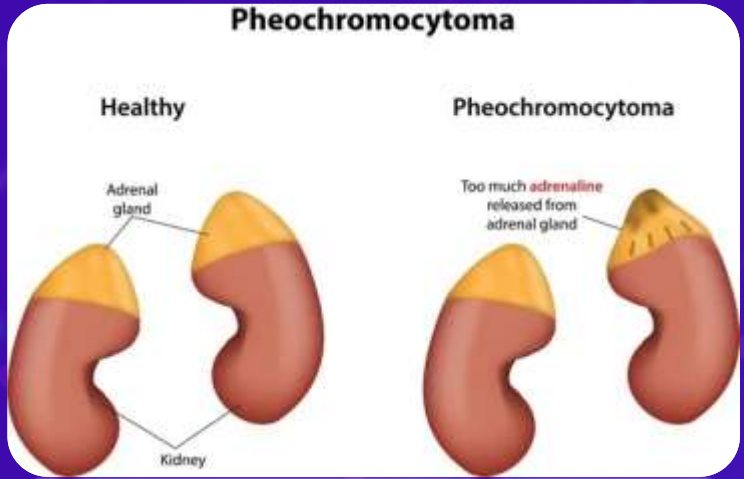
# MEN subtype 2A

Medullary thyroid cancer, Pheochromocytoma, primary hyperparathyroidism, Hirschsprung disease.

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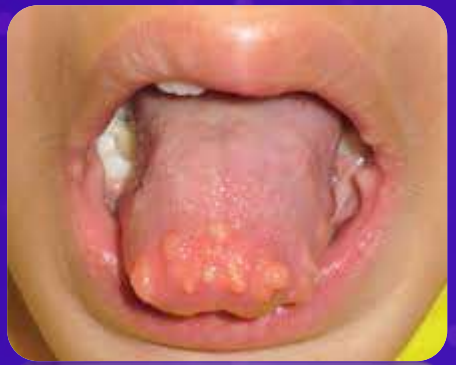
# MEN subtype 2B

Medullary thyroid cancer, pheochromocytoma, mucosal neuromas, ganglioneruomatosis of the gastrointestinal tract.



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# Familial medullary thyroid cancer

Medullary thyroid cancer in at least four family members, with documented absence of other endocrinopathies.

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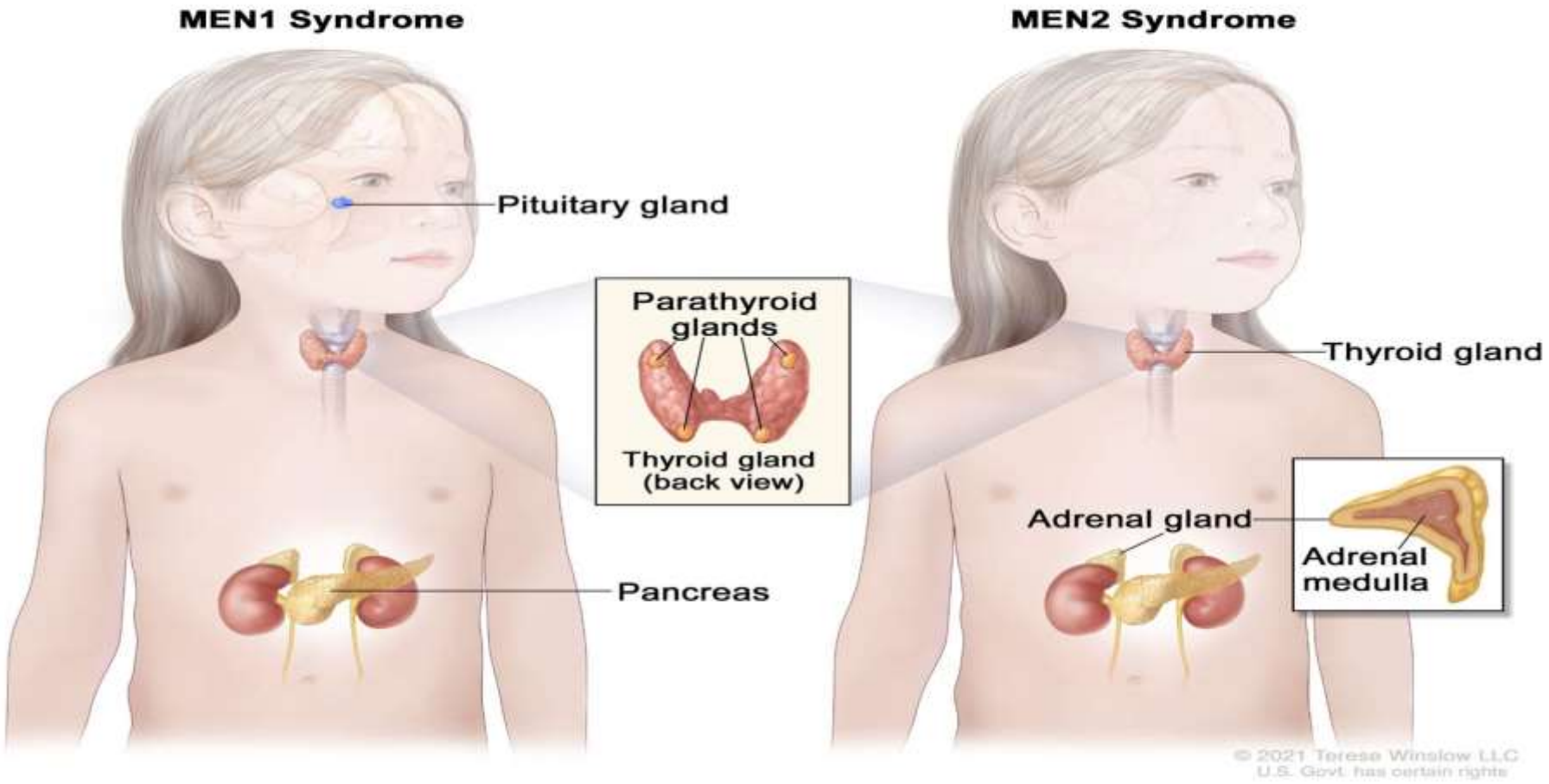
# Parts of the Body Affected by MEN Syndromes

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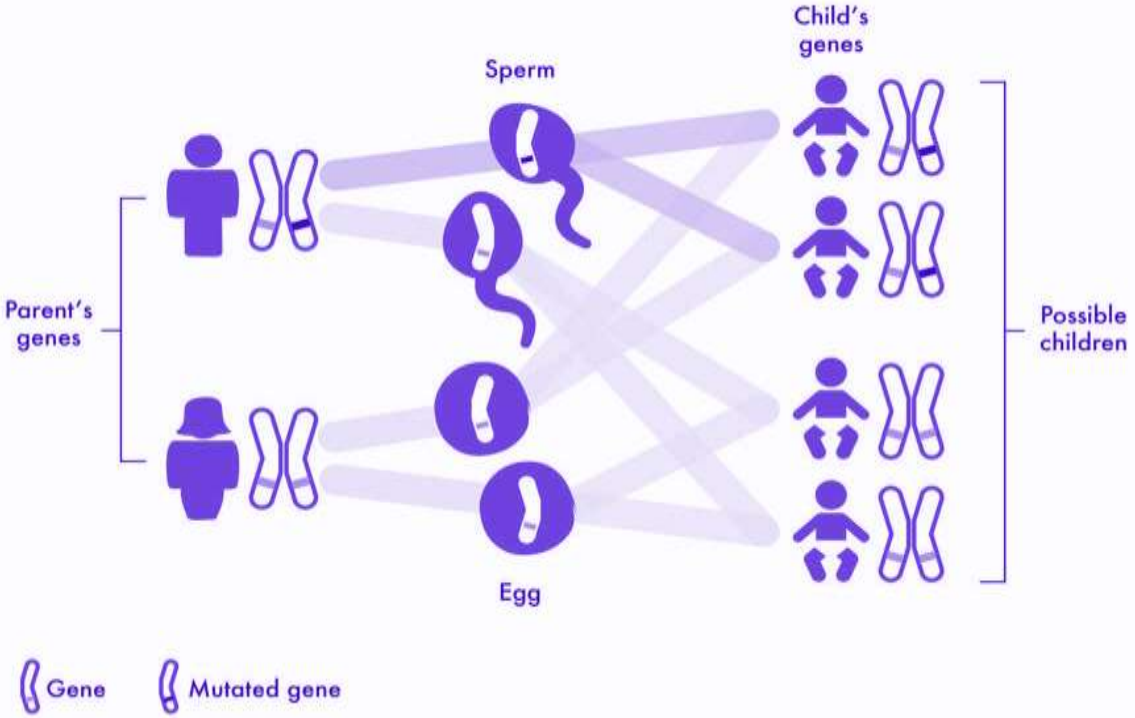
# Case study

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# More about the RET gene.

RET Is a tyrosine kinase receptor that transmits growth and differentiation signals to a variety of growing tissues.

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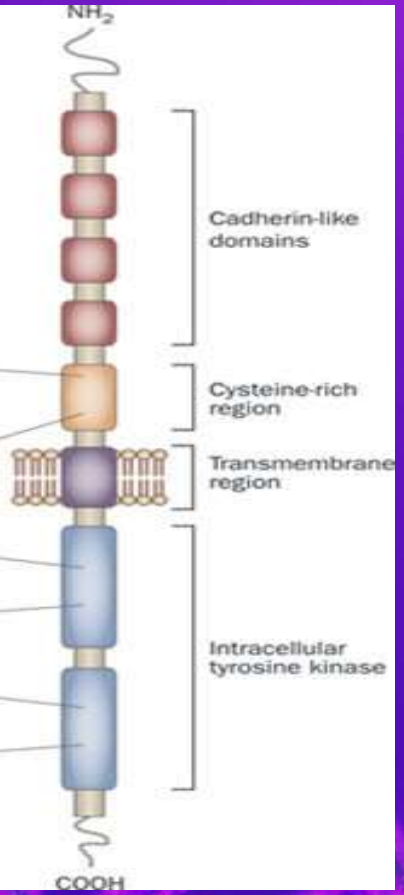
| Exon, codon and mutation | MTC | +PHEO | +PHPT | +CLA | +HSCR | +MEN2B phenotype | ATA risk | Age at testing* |
|---|---|---|---|---|---|---|---|---|
| Exon 10 | | | | | | | | |
| C609R/G/F/S/Y | ○ | ○ | ○ | | ○ | | B | 5 years |
| C611R/G/F/S/W/Y | ○ | ○ | ○ | | ○ | | B | 5 years |
| C618R/G/F/S/Y | ○ | ○ | ○ | | ○ | | B | 5 years |
| C620R/G/F/S/W/Y | ○ | ○ | | | ○ | | B | 5 years |
| Exon 11 | | | | | | | | |
| C630R/F/S/Y | ○ | | ○ | | | | B | 5 years |
| C634R/G/F/S/W/Y | ○ | ○ | ○ | ○ | | | C | 3–5 years |
| Exon 13 | | | | | | | | |
| E768D | ○ | ○ | | | | | A | 5–10 years |
| L790F | ○ | ○ | | | | | A | 5–10 years |
| Exon 14 | | | | | | | | |
| V804L/M | ○ | ○ | ○ | ○ | | | A | 5–10 years‡ |
| Exon 15 | | | | | | | | |
| A883F | ○ | ○ | | | | ○ | D | <6 months§ |
| S891A | ○ | ○ | ○ | | | | A | 5–10 years |
| Exon 16 | | | | | | | | |
| M918T | ○ | ○ | | | | ○ | D | <6 months |

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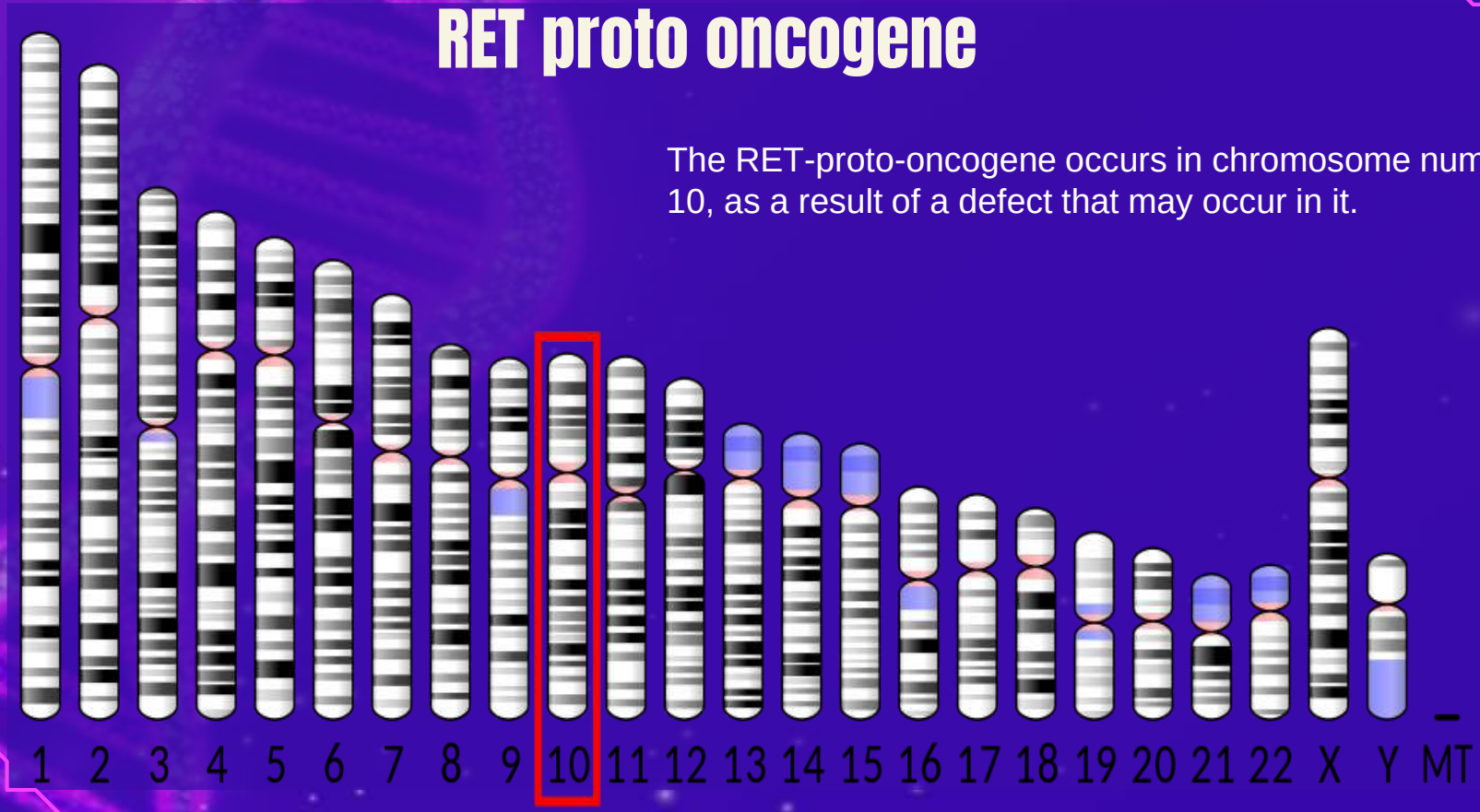
# RET proto oncogene

The RET-proto-oncogene occurs in chromosome number 10, as a result of a defect that may occur in it.

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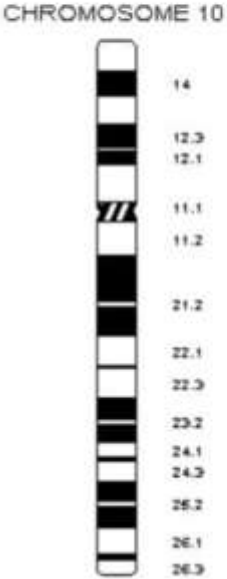
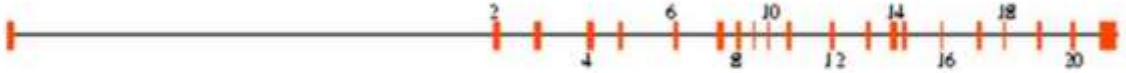
# *RET* proto-oncogene




- 10q11.2
- 21 exons over 53kb of genomic DNA

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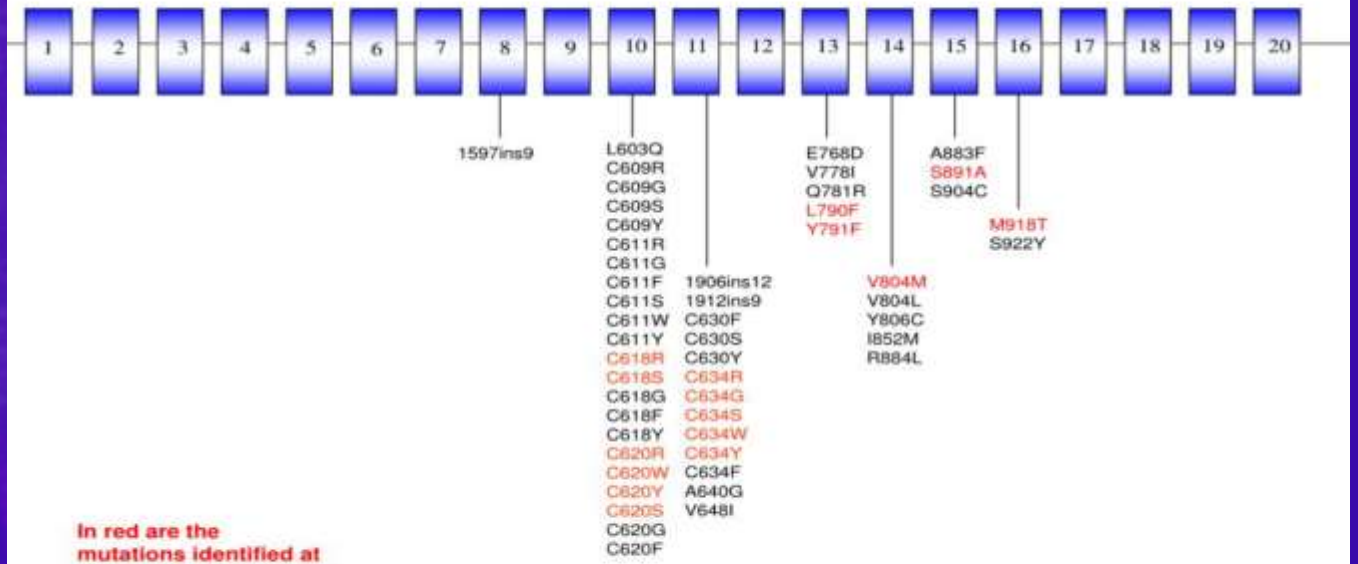
# *RET* mutations in MEN2




**In red are the mutations identified at the Exeter molecular genetics laboratory**

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# Population with MEN2

It is estimated that about 1 in 30,000 people has MEN2.
Most people with MEN2B do not have any family history of the condition. They have a de novo (new) mutation in the RET gene.
Fewer than 5% of people with MEN2A are thought to have a de novo mutation in the RET gene.

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# WHO SHOULD undergo genetic screening?

1). Clinically proven MEN2 syndrome:
bilateral, multicentric MTC and pheochromocytoma

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2). MTC or pheochromocytoma and a family member with MTC or pheochromocytoma

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3). Sporadic MEN2 related tumors:

a. Age younger than 35 (thirty five) years

b. Multicentric tumors in one organ

c. Two different organs affected

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# Treatment

Early detection of MEN2 by screening "at-risk" family members is advantageous.
And then followed by biochemical screening for the other endocrine tumors.
So Therefore, after surgery, thyroid hormones replacement therapy is required .

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