MEN 2B (Multiple Endocrine Neoplasia)

Multiple Endocrine Neoplasm (MEN) 2B is a group of neoplasms occurring together in characteristic organs due to mutation in the RET oncogene. The neoplasms typically originate from the medullary cells of the thyroid gland, mucosal ganglionic cells, and chromaffin cells of the adrenal gland which results in pheochromocytoma. Additionally, patients have a characteristic marfanoid appearance.

PLAY PICMONIC

Pathophysiology

RET Gene

Roulette

The RET gene, which codes for a receptor tyrosine kinase, is a proto-oncogene found on chromosome 10. It is the gain of function mutation that causes the clinical manifestations in MEN 2A and 2B.

Autosomal Dominant

Domino

The disorder has an autosomal dominant inheritance. As a result, people with this condition frequently have a strong family history.

Signs and Symptoms

Medullary Thyroid Carcinoma

Medullary Medusa and Car-gnome

Medullary thyroid carcinoma is a malignant tumor that is poorly differentiated and consists of parafollicular cells, also known as C cells. These cells secrete calcitonin, which lowers the calcium level in the blood. Cases associated with MEN 2 are more common in younger people (30s), while sporadic cases appear much later in life (60s). The tumor is also diffuse and bilateral.

Pheochromocytoma

Phiat-chrome

This is a neuroendocrine tumor of the adrenal medulla derived from chromaffin cells that secrete catecholamines. Chromaffin cells develop from neural crest cells. Palpitations, tachycardia, headaches, diaphoresis, and episodic hypertension are common clinical manifestations in both MEN 2 and sporadic forms.

Mucosal Neuromas

Mucous Nerve-mass

These are benign nerve tissue growths on the mucosal surface. MEN 2B is characterized by the presence of mucosal neuromas on the lips, tongue, and intestinal wall (intestinal ganglioneuromatosis).

Marfanoid Body Habitus

Marfan-the-martian

Patients with marfanoid body habitus have skeletal deformities, long limbs, arachnodactyly, joint hyperlaxity, and a high-arched palate. It is distinguished from Marfan syndrome by the absence of aortic or lens involvement.