20.68 Multiple endocrine neoplasia (MEN) syndromes

RARE AUTOSOMAL DOMINANT SYNDROMES

MEN 1 (Wermer's syndrome)

INACTIVATING MUTATION IN MENIN (TUMOUR SUPPRESSOR GENE) ON CHROMOSOME 11

- Primary hyperparathyroidism SERUM Ca++
- Pituitary tumours SERUM PROLACTIN / MRI
- Pancreatic neuro-endocrine tumours (insulinoma, gastrinoma) SERUM OF GIT HORMONES / MRI OF PANCREAS

MEN 2 (Sipple's syndrome)

GAIN OF FUNCTION IN RET (PROTO-ONCOGEN) ON CHROMOSOME 10

- Primary hyperparathyroidism SERUM Ca++
- Medullary carcinoma of thyroid 100% WILL OCCUR, SO PROPHYLACTIC THROIDECTOMY
- Phaeochromocytoma URINARY CATECHOLAMINE METABOLITES

In addition, in MEN 2b syndrome, there are phenotypic changes (including marfanoid habitus, skeletal abnormalities, abnormal dental enamel, multiple mucosal neuromas)

Reference: Davidson's Principles and Practice of Medicine

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