Multiple endocrine neoplasia type 2

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Multiple endocrine neoplasia type 2 (MEN 2) is an autosomal dominant cancer syndrome with major components of medullary thyroid carcinoma (MTC), pheochromocytoma and hyperparathyroidism. The disease is caused by germline mutations of the RET proto-oncogene. Subtypes of MEN 2 include MEN 2A, MEN 2B and familial MTC (FMTC) which differ in pattern of additional lesions or - in FMTC - lack of pheochromocytoma. In 2009, after extensive review of the literature, the guidelines of the American Thyroid Association made several recommendations regarding clinical and genetic diagnostic testing and treatment options. In this article, the recently published literature is reviewed and concerns regarding future perspectives are added. In particular, a critical handling of rare DNA variants and double mutations is necessary. Up to now, mutation-specific risk profiles and mutation-associated treatment recommendations are unavailable. We emphasise the need for approved centres for treatment of patients