The genetic basis of hereditary medullary thyroid cancer: clinical implications for the surgeon, with a particular emphasis on the role of prophylactic thyroidectomy.

Abstract:
Medullary thyroid cancer (MTC) may occur either sporadically or on a hereditary basis. Hereditary MTC may be observed with either multiple endocrine neoplasia syndromes (MEN 2A and MEN 2B) or as familial MTC (FMTC). Despite the rarity of these syndromes, early diagnosis is especially important, since MTC is a lethal disease if not promptly and appropriately treated. Recently, the development of genetic testing and direct DNA analysis allows the identification of asymptomatic patients. Surgical prophylaxis should be considered in these cases, ideally to prevent the development of MTC. During the recent decade, the concept of 'codon-directed' timing of prophylactic surgery emerged as a reasonable strategy in the management of these patients. Currently, genetic analysis offers the possibility to define genotype-phenotype correlations and to adjust the time of prophylactic surgery. Hereditary MTC is a model of genetically determined cancer in which both diagnostic and therapeutic strategies rely on the identification of specific mutations.