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Multiple Endocrine Neoplasia Type 2A: Case Report

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Abstract

Multiple endocrine neoplasia type 2A (MEN 2A) is a complex autosomal dominant inherited syndrome characterized by medullary thyroid carcinoma (MTC), pheochromocytoma and primary parathyroid hyperplasia. In patients with only one or two clinical features, identification of a germline RET (REarranged in Transfection) mutation or the identification of the clinical features of MEN 2A in other first degree relatives is required to make the diagnosis. We present the case of a family with MEN 2A syndrome confirmed by genetic analysis which identified RET gene mutation in 634 codon in father - DV - aged 48 years and also in daughter DM - aged 20 years. The specific feature in this case is that the index case was the daughter (diagnosed and operated for pheo-chromocytoma at the age of 19 years), the father being diagnosed later with medullary thyroid carcinoma by mutational screening in all family members. This family supports the phenomenon of anticipation, in which severity increases and the age of onset decreases in successive generations, the syndrome being discovered earlier and with a worse prognostic in the daughter.

Key words: MEN 2A, RET mutations, genetic screening

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