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Occurrence of phaeochromocytoma tumours in RET mutation carriers - a single-centre study.

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Abstract

INTRODUCTION:

Multiple endocrine neoplasia type 2 (MEN 2) is an autosomal dominant genetic syndrome caused by germline mutation in RET proto-oncogene. The most common mutations are in a cysteine rich domain. Phaeochromocytoma will develop in approximately 50% of RET proto-oncogene carriers.

MATERIAL AND METHODS:

The studied population consisted of 228 RET proto-oncogene mutation carriers. Monitoring for the diagnosis of phaeochromocytoma was carried out in all patients with established genetic status. Mean time of follow up was 138 months. Surveillance consisted of periodically performed clinical evaluation, 24-hour urinary determinations of total metanephrines complementary with imaging (CT, MR, MIBG scintigraphy).

RESULTS:

Phaeochromocytoma developed in 41 patients (18% of all RET proto-oncogene mutations carriers). The mean age of diagnosis for the whole cohort was 43 years. In eight cases phaeochromocytoma was the first manifestation of the MEN 2 syndrome. Only eight (20%) patients were symptomatic at diagnosis of phaeochromocytoma. The mean size of the tumour was 4.3 cm. There was no extra-adrenal localisation. We observed one case of malignant phaeochromocytoma.

CONCLUSIONS:

In patients with MEN 2 syndrome phaeochromocytomas are usually benign adrenal tumours with high risk of bilateral development. Taking to account the latter risk and non-specific clinical manifestation of the neoplasm it is mandatory to screen all RET proto-oncogene mutations carriers for phaeochromocytoma