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Hormonal Mechanisms

6.3 Pheochromocytoma: Clinical, Diagnostic and Genetic Aspects: Experience in a Single Centre

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Introduction. Pheochromocytomas and paragangliomas are rare tumours with common embriogenetic origin, pheochromocytoma can be sporadic or syndromic. There are four different syndromes genetically determined (MEN 2, paraganglioma/pheochromocytoma syndrome, Von Hippel Lindau syndrome and neurofibromatosis type 1).

Methods. We have evaluated in 93 patients with pheochromocytoma, the prevalence of syndromic forms and the prevalence of malignancy. Patients and methods: 93 patients with pheochromocytoma seen in the Day Hospital of Department of Clinical Sciences of "La Sapienza" University in Rome. A clinical story and a physical examination was made in all patients, a collection of 24 urine samples was obtained to determine metanephrine and acid vanylil mandelic (VMA). A CT scan of abdomen and/or a MR abdomen was performed to detect the presence of a mass and selected cases underwent a total body scintigraphy with I-131 MIBG. After an adequate medical preparation with alpha and beta-blockers patients underwent a surgical treatment with laparoscopic approach to remove the pheochromocytoma. A genetic analysis with study of susceptibility gene for pheochromocytoma (RET, VHL, NF1, SHDB/D) was performed in most of the patients.

Results. Pheochromocyoma was sporadic in 77 cases (82%) (average age 45 years) and syndromic in 16 cases (18 %) (average age 38 years). MEN 2A was present in 5 cases (32% syndromic forms), VHL in 4 cases (25%), la NF1 in 4 cases (25%) and PGL 4 syndrome (SDHD) in 3 cases (18%). Pheocromocytoma was benign in 93% of cases, in 7% pheochromocytoma has a malignant nature.

Conclusions. Our data are similar to those reported in literature and confirm the typical course of this disease.