

A late diagnosis of MEN 1 Syndrome in a young patient initially presenting with nephrolithiasis

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Background:

Multiple Endocrine Neoplasms Type 1 (MEN 1), originally called Wermer Syndrome, is a rare hereditary condition caused by mutations in the MEN1 tumor suppressor gene. It is characterized by tumors of the parathyroid glands, the anterior pituitary gland and pancreatic islet cells. Hyperparathyroidism is the most common manifestation of this syndrome. MEN1 can also be associated with other endocrine and non – endocrine tumors.

Case presentation:

A male patient presented with nephrolithiasis at the age of 31 and was regularly followed up by a nephrologist. At the age of 35, he was diagnosed with hyperparathyroidism and underwent partial parathyroidectomy. After almost 8 years the patient reported severe heartburn, diarrhea and significant weight loss. A CT scan showed multiple liver lesions, spinal osteoblastic lesions and pancreatic head and tail lesions. Liver biopsy revealed neuroendocrine tumor (NET), Ki-67 8%. Subsequently, genetic testing was performed and MEN1 syndrome was confirmed. The patient was initially treated with octreotide. Nevertheless, the disease had progressed. Peptide receptor radionuclide therapy (PRTT) was not an option as the majority of metastases were negative for somatostatin receptors. Consequently, several treatment options were attempted such as transarterial chemoembolization, everolimus and combined chemotherapy which included capecitabine and temozolomide. However, due to acutization of renal failure, the treatment was stopped and the patient had to undergo hemodialysis. Unfortunately, four months later, the patient developed hemodynamic shock and died despite treatment.

Conclusion:

Despite the rare occurrence of MEN1, early diagnosis is crucial for a favorable outcome of these patients. Therefore, in every young patient presenting with hyperparathyroidism one should always suspect MEN1 and follow-up because of the risk for developing neuroendocrine tumors.