Hereditary Pheocromocytoma-Paraganglioma Syndrome. A case Report.

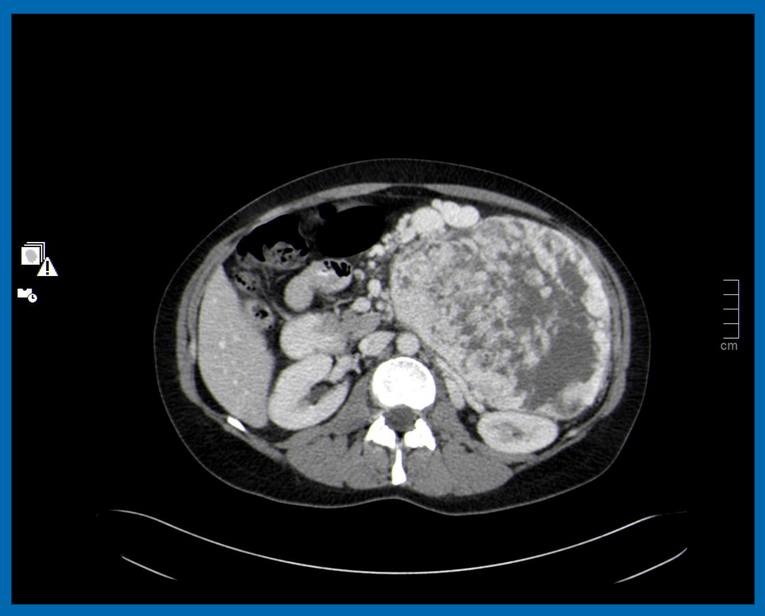
M.Teresa Gallego. Lucía Vera, Amparo Meoro, Cristina Del Peso ,R. Soriano .

Servicio de Endocrinología y Nutrición. Hospital General Universitario Reina Sofía. Murcia. Spain.

EP274

INTRODUCTION: Paraganglioma (PGL) develops from cells of the parasympathetic and sympathetic system. It usually manifests as a slow- growing and painless mass.PGL may be hereditary, benign, malignant, unilateral or bilateral tumors. In most cases PGL is located around the common carotid artery but may also be located within the middle ear or in the abdomen. Non functional retroperitoneal PGL are rare tumors, usually asymptomatic, and can attain big dimensions. Mutations in the succinate dehydrogenase gene complex have been identified as a cause of inherited Pheocromocytoma-Paraganglioma Syndrome (PGL/PCC). Malignant PGL is a uncommon presentation diagnosed by local recurrence after total resection of primary mass or findings of distant metastases.

CASE REPORT: A 47 years-old woman with a non functional retroperitoneal PGL. The patient presented as a single symptom constipation. A large pararenal tumor of 15x10cm and 1100 gr was found in imaging studies and removed completely. No distant metastases were seen. After a follow up period of 10 months the patient was in a good health, asymptomatic, but a second operation was performed because of evidence of tumor recurrence. After surgery the patient was treated with neoadjuvant radiotherapy.
Genetic analysis revealed Succinate Dehydrogenase B mutation as well as in two of her three children and four of her five brothers.



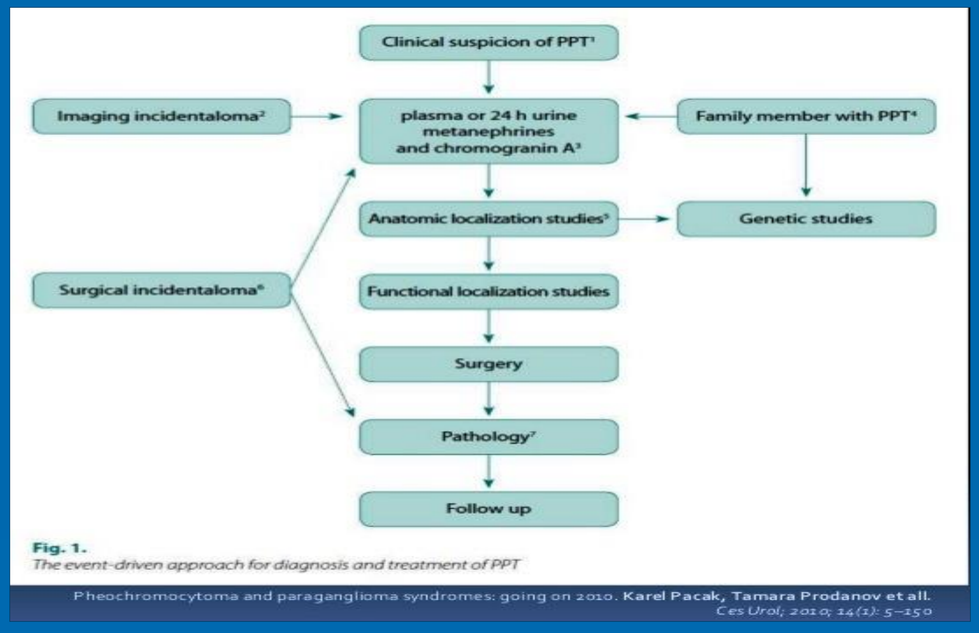


Fig. 1.
The event-driven approach for diagnosis and treatment of PPT

Pheochromocytoma and paraganglioma syndromes: going on 2010. **Karel Pacak, Tamara Prodanov et all.** *Ces Urol; 2010; 14(1): 5–150*

INDICATIONS FOR GENETIC TESTS IN PGL/PCC

*All of PGLs.
*Bilateral PCCs.
*PCC and family history of inherited *PGL/PCC.*
*PCC in young people under the age of 45 .
*Associated to PGL/PCC genetic syndromes.
*Metastatic or Dopamine PCCs.

DISCUSSION: The majority of sympathetic PGL/PCCs produces catecholamines, in advanced cases resulting in typical symptoms and signs such as palpitations, headache, diaphoresis, and hypertension. The state-of-the-art diagnosis and localization of sPGL/PCCs are based on measurement of plasma and/or 24-h urinary excretion of (fractionated) metanephrines and methoxytyramine (MT).sPGL/PCCs can subsequently be localized by anatomical(computed tomography and/or magnetic resonance imaging) and functional imaging studies(123I-metaiodobenzylguanidine-scintigraphy, 111In-pentetreotide cintigraphy, or PET with radiolabeled dopamine or dihydroxyphenylalanine.Although most PGL/PCCs are benign, factors such as genetic background ,tumor size, tumor location, and high MT levels are associated with higher rates of metastatic disease. Surgery is the only curative treatment. Treatment options for patients with metastatic disease are limited. PGL/PCCs have a strong genetic background, with at least one-third of all cases linked with germline mutations in 11 susceptibility genes. As genetic testing becomes more widely available, the diagnosis of PGL/PCCs will be made earlier due to routine screening of at-risk patients. Early detection of a familial PGL allows early detection of potentially malignant PGLs and early surgical treatment, reducing the complication rates of this operation.

CONCLUSIONS:

The malignant potential of the PGL is determined by local invasion as well as distant metastases as there are no characteristic cellular changes. In retroperitoneal PGL 50% are thought to be malignant. The SDHB mutation plays an important role in malignant PCC/PGL. The present case indicates that conducting genetic testing, including SDHB mutation analyses, is required to determine the prognosis in patients highly suspected of having malignant tumors in the context of a PGL/PCC Syndrome.