Incidental pancreatic nodule in a patient with Tuberous Sclerosis

https://doi.org/10.56238/sevened2024.001-007

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ABSTRACT

TS is a genetic disorder that causes a cell proliferation and migration disorder related to variable clinical manifestations, ranging from benign lesions to malignant tumors. Most TS patients diagnosed with NETs have pancreatic involvement, but the presence of these tumors in other organs should be considered as part of the disease phenotype. Recent studies have shown that most tuberous sclerosis patients with pancreatic NETs have a germline pathogenic variant in the TSC2 gene. In the present report, the patient had an asymptomatic tumor in the caudal region of the pancreas. Current surgical recommendations for non-functional pancreatic NETs indicate the need for surgical removal that is larger than 20 mm in size or has very rapid growth. In the case discussed, the area of the tumor made it necessary to remove it surgically.

Keywords: Tuberous sclerose, Pancreatectomy, Neuroendocrine tumors.

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INTRODUCTION

Tuberous Sclerosis (TS) is a genetic disorder resulting from a mutation in the TSC1 or TSC2 gene, responsible for the formation of proteins acting in cell hyperplasia. Thus, their mutation leads to a cell proliferation and migration disorder related to variable clinical manifestations, ranging from benign lesions to malignant tumors. Classically, the organs affected by the disease are the brain, skin, heart, eyes, kidneys and lungs. The following report is relevant due to the approach to the case of a patient with TS and Autism Spectrum Disorder (ASD) who evolved with pancreatic lesions.

CASE PRESENTATION

A 17-year-old male patient with TS and ASD was admitted to the Oswaldo Cruz University Hospital with a history of expansive formation in the caudal region of the pancreas, measuring 2.4 cm X 2.2 cm in the major transverse axes, with heterogeneous internal areas, as evidenced by a previous MRI. The mother reported that, during this period, the patient did not present any symptoms, denied weight loss, change in eating or bowel movements, and abdominal pain. On examination: good general condition, non-verbal autistic, eupneic, normostained, hydrated, anicteric, afebrile to the touch. There were no changes in the cardiorespiratory and gastrointestinal systems. Edema-free extremities, full and symmetrical peripheral pulses. A computed tomography scan of the abdomen was previously performed, which showed a pancreas of preserved topography and contours, presenting a hypoattenuating nodular formation with calcification in the periphery located in the tail with hypoenhancement on contrast medium measuring 1.9 cm X 2.1 cm. The patient underwent splenectomy + body-caudal pancreatectomy by videolaparoscopy + excision of facial nevi, in which a solid tumor was found in the topography of the tail of the pancreas of approximately 2 cm; Absence of free fluid and peritoneal carcinomatosis; Enlarged spleen with small lesions on the anterior face. The anatomopathological examination showed a condition that favors the diagnosis of well-differentiated neuroendocrine neoplasia.

Image 1: Isolation of the body of the pancreas; site where pancreatic transection will be performed.

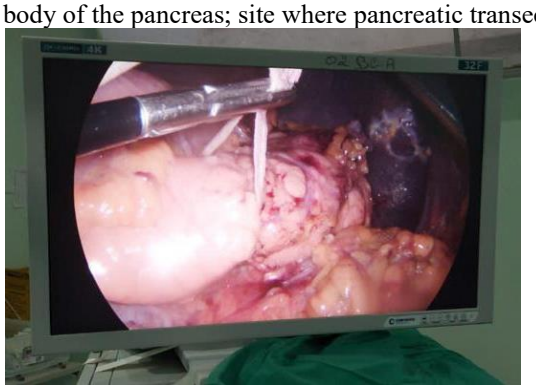

Figure of the author.

Image 2: Final appearance for removal of the spleen and pancreas.

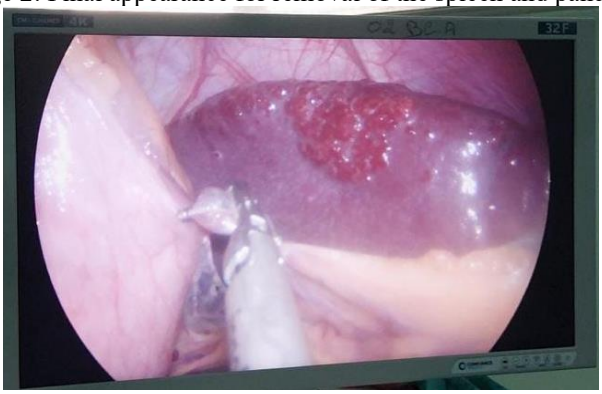

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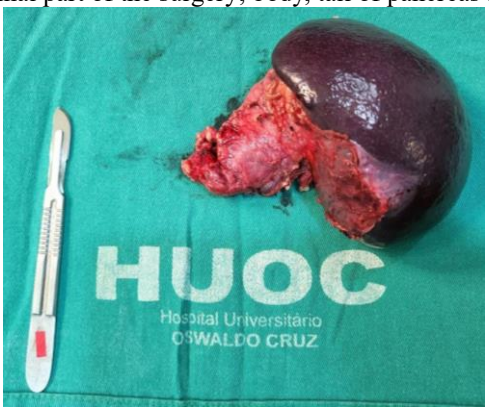
Image 3: Final part of the surgery; body, tail of pancreas and spleen.



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DISCUSSION

Pancreatic neuroendocrine tumors (NETs) are most commonly sporadic but have been previously reported in association with TS. Most TS patients diagnosed with NETs have pancreatic involvement, but the presence of these tumors in other organs should be considered as part of the disease phenotype. Recent studies have shown that most tuberous sclerosis patients with pancreatic NETs have a germline pathogenic variant in the TSC2 gene. In the present report, the patient had an asymptomatic tumor in the caudal region of the pancreas. Current surgical recommendations for non-functional pancreatic NETs indicate the need for surgical removal that is larger than 20 mm in size or has very rapid growth. In the case addressed, the area of the tumor required surgical removal, and the laparoscopic approach to the lesion was the procedure of choice.

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