

Contribution of functional imaging in the follow-up of a patient with pituitary macroadenoma, paraganglioma, renal tumor, and SDHC gene variant.

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Introduction:

Patients carrying SDHx mutations have been described with rare pituitary adenomas and kidney cancers.

Observation:

A 54-year-old patient presented a 22 mm macroprolactinoma (prolactinemia 627ug/L). Treatment with cabergoline, 1.5 mg per week, normalized prolactin in 3 months and stabilized the tumor. Patient reported previous surgery of left jugulo-tympanic paraganglioma at the age of 32, followed by radiotherapy without any recent follow-up

NGS sequencing of paraganglioma susceptibility genes identified a heterozygous constitutional variant of undetermined significance in the SDHC gene (c.160C>A; p.(Pro54Thr)). This variant was classified as probably pathogenic, thankful by a loss of SDHB expression in paraganglioma in immunohistochemical analysis.

To test the hypothesis of causality of the SDHC mutation in the emergence of macroprolactinoma, an MRI spectroscopy was performed (figure 1), It showed an accumulation of succinate in the pituitary adenoma.

A 68Ga-DOTATOC PET scan revealed a fixation on the residual cervical lesion and an intense fixation of the left kidney corresponding, on CT, to a 4 cm lesion compatible with a clear cell renal carcinoma (RCC).

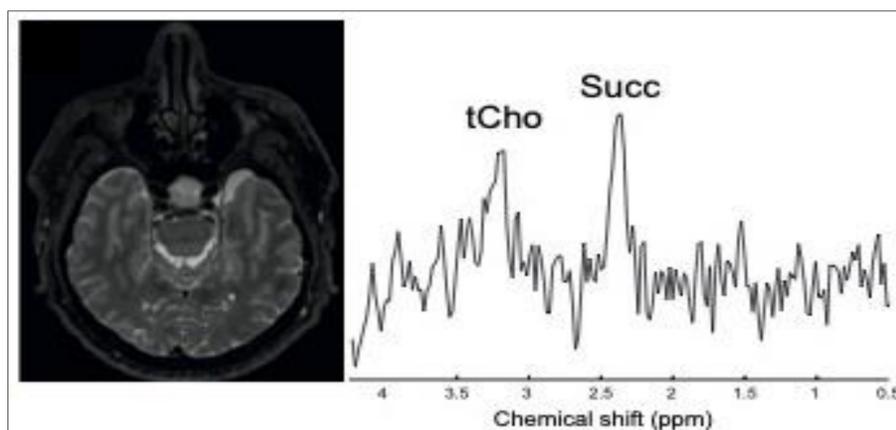


Figure 1. MR spectra acquired in the neck PGL of this patient and in the pheochromocytoma . The location of the VOIs is shown on coronal or axial T2-WI. Voxel sizes was 11 x 11 x 11 mm³ (PA) for this case.
tCho: choline containing compounds; Succ: succinate.

Discussion:

We report the case of a patient with a pathogenic variant of SDHC. This mutation was responsible for jugulo-tympanic paraganglioma, macroprolactinoma, and an RCC. New functional imaging techniques recently developed in vivo (MRI spectroscopy and 68Ga-DOTATOC PET) and in vitro (SDHB immunohistochemistry) allow optimal management of patients with SDHx mutation.