Abstract

Introduction: In this work we report a clinical case of PKD-TSC syndrome, with a review of current literature.

Case report: In August 2013 we admitted in our clinic a patient with a history of Tuberous Sclerosis Complex (TSC) who had a CT finding of right renal tumor in a kidney with multiple cysts. After performing a right radical nephrectomy, a genetic investigation assessed that the patient was affected by “PKD-TSC syndrome”, with a deletion on chromosome 16p 13.3.

Discussion: Genomic findings demonstrated that PKD-TSC syndrome is due to large deletions in chromosome 16 that results in deletion of part or all of both the TSC2 and the PKD1 gene, giving both aspects of Polycystic Kidney Disease and Tuberous Sclerosis Syndrome.

Conclusion: Our report is emblematic of this rare disease that has a genetic pathogenesis that needs further studies to be totally understood.

Keywords: Pkd-tsc syndrome; Renal cell carcinoma; Surgery; Genetic disorders

Introduction

Autosomal Dominant Polycystic Kidney Disease (AD-PKD) and Tuberous Sclerosis Complex (TSC) are two well described genetic disorders. However, in Literature [1] it’s documented an uncommon condition, due to a deletion on chromosome 16p13.3, which involves both PKD1 and TSC2 genes, called “PKD-TSC syndrome”. Little more than 30 cases are described in literature, with none specific relation to age [2]. We report a case of a 42-years-old man with Polycystic Kidney Disease and Tuberous Sclerosis Complex who presented with a Computed Tomography (CT) finding of a 25 mm upper pole right renal neoplasm, associated with a 20 mm angiomyolipoma, inside a cystic formation with Antero-Posterior (AP) diameter 62 mm, Latero-Lateral (LL) 55 mm and Cranio-Caudal (CC) 53 mm. The patient underwent a right radical nephrectomy, as renal function was already impaired and the patient was candidate to dialysis.

Case Report

In August 2013, a 42-years-old man came to our attention sent by his family physician, with a report of impaired renal function associated with right renal colics and pyelonephritis.

This patient was already affected by tuberous sclerosis that causes him recurrent convulsive crisis, for which he was in therapy with Carbamazepine since 2010.

Admitted in Our department, we assessed medical antibiotic therapy and we performed a Computed Tomography (CT), who showed an image of bilateral renal enlargement due to a polycystic disease (Figures 1 and 2). The patient hadn’t any previous familiar history for Polycystic Kidney Disease. At the superior third of right kidney, an exophytic cyst of 62×55×53 mm presented on its posterior-medial wall a solid 20 mm formation (Figure1). Nearby, there was a roundish 25 mm solid formation, which showed a disomogeneous density value with rapid contrast enhancement, dubious of neoplastic disease. Contemporary, a hepatic asymptomatic cystic disease was assessed. The indication was surgical and we opted for a right radical nephrectomy, as renal function was already impaired and the patient was candidate to dialysis.

Figure 1: CT appearance of upper pole right renal neoplasm into a clear polycystic disease (Varese, 2013)

The intervention was performed in October 2013, delayed as he had a convulsive episode on August, studied with a basal EEG and neurological evaluation after the regression of acute infective disease.
The day of surgery, the patient had a near-syncope with bradycardia, so the intervention was delayed of two days. Surgical access was a mono-lateral right subcostal incision and we performed a radical nephrectomy of a 15x8x7.5 cm kidney; total operating time was 2.20 hours. The first post-operative days were characterized by a temporary amylases increase, stabilized after few days. Ultrasound and radiological verifications didn’t noticed any early surgical complication.

The Pathologist report was of a 22 mm nodule of clear cell renal cell carcinoma pT1a, pNx, G2, associated with a 20 mm angiomylipoma in an adult renal multicystic disease. The patient was discharged in good general conditions and also postoperative follow up at three and six months didn’t show any early neoplastic recurrence. As Tuberous Sclerosis Complex was coexisting with a condition of multicystic renal disease, our pathology division carried out a genomic evaluation of the same chromosome 16p13.3, pathognomonic of “PKD-TSC syndrome”. The typical finding that underlines PKD-TSC syndrome is an angiomylipoma in pathologic specimen of patients with history of Polycystic Kidney Disease. Even if the most common finding in this disease is multiple hamartomas in different organs like brain, skin, kidney, liver, lung and heart, it’s also possible to develop malignant tumors, mainly renal or brain [8]. In PKD-TSC syndrome Renal Cell Carcinoma (RCC) develops within dysplastic epithelial cysts, and mean age of diagnosis for people with PKD-TSC-associated RCC is 25 years lower than common population.

Conclusion

Our clinical case is emblematic of PKD-TSC syndrome, with a phenotype clearly due to this disease. Surgical resection of the tumor is clearly the best option to adopt in this case, aiming to oncological radicality. The patient didn’t present any early recidivism during his follow-up examinations at three and six months. PKD-TSC syndrome is a genetic disease that carries both manifestations of AD-PKD and TSC, due to large deletions in chromosome 16 that results in deletion of part or all of both the TSC2 and the PKD1 gene.

References