Renal cancer in patient with polycystic kidney disease without end-stage renal disease

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Background: Polycystic kidney disease is the most commonly inherited cystic renal disease and is one of the leading causes of end-stage renal disease (ESRD). Major types of renal cancer include renal cell carcinoma (RCC) and Transitional cell carcinoma (TCC), and in either case, it is difficult to establish a treatment strategy after it has already become an ESRD. Herein, we present a case in which renal cancer was identified without end-stage renal disease in autosomal dominant polycystic kidney disease (ADPKD), and the disease was diagnosed and treated by molecular genomic analysis.

Case presentation: In November 2021, A 84 year-old man was referred to our hospital with Left kidney mass and sustaining malignant fever. The patient had a history of polycystic kidney disease, Right vertebral artery dissection and hypertension with medication. Findings on abdomen and chest CT showed multiple enlarged left kidney mass in renal parenchyma, left upper pole pelvis, metastatic para-aortic LAP, several small hepatic metastases in both hepatic lobes and multiple lung metastasis. Clinically, it suggested Lt RCC in upper pole (T3aN1M1) or Infiltrative urothelial carcinoma. An excisional biopsy of the left level IV neck node was performed. Gross pathology was suspected non-clear cell RCC such as papillary-RCC. But,the munohistochemical staining results were positive for CK7, CK20, and GATA3. This was consistent with TCC and for more accurate diagnosis, circulating tumor DNA NGS was performed through liquid biopsy. The patient was subsequently treated with platin doublet chemotherapy.

Discussion: Thanks to advances in molecular biology, genomic data of circulating tumor DNA in blood can fill in insufficient information about tissue biopsy in patients who are difficult to perform therapeutic and diagnostic nephrectomy. It was a case of how genomics research could be helpful in ADPKD patient with clinically, limited use of drugs with nephrotoxicity, limitations in radiologic work-up, limitations in invasive procedures due to comorbidity, and different results from known pidemiologic data.