CASE REPORT

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Bilateral Cystic Nephroma with *DICER1* Mutation

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ABSTRACT Cystic nephroma (CN) is a rare, asymptomatic, benign kidney tumor. Clinically and radiologically, CN is difficult to distinguish from malignant tumors such as cystic partially differentiated nephroblastoma and cystic Wilms tumor, and the definitive diagnosis is made by histopathological examination. The effective method of treatment is removal of the tumor. Depending on the location and size of the mass, total nephrectomy or nephron-sparing surgery are the treatment options. *DICER1* gene mutation is frequently seen in bilateral cases, and *DICER1* gene mutation was also positive in our fourteen-month-old male patient. Since the gene has a predisposition to malignancy. We aimed to present our approach with the literature.

Keywords: Cystic nephroma; *DICER1*; kidney tumor

A benign kidney tumor called a cystic nephroma (CN) is a rare and asymptomatic. Cysts may have septation and can be of different sizes. It is usually separated from the kidney by a fibrous capsule. Its etiology is unknown. With the widespread use of imaging methods, awareness of the tumor has increased. Cystic Wilms tumor and cystic partly differentiated nephroblastoma are two malignant tumors that are difficult to identify from CN clinically and radiologically. The definitive diagnosis is made by histopathological examination. The effective method of treatment is removal of the tumor. Depending on the location and size of the mass, total nephrectomy or nephron-sparing surgery are the treatment options.

We describe a patient with bilateral CN in this case report. The bilaterality of CN is very rare. *DICER1* gene mutation can be seen frequently in bilateral cases. This gene is a gene associated with malignant cancers. Therefore, early recognition and close follow-up are important.

CASE REPORT

Necessary permissions were obtained from the patient's family. This case report does not require institutional review. The patient provided the required consents. Fourteen-month-old male patient was admitted with significant abdominal distension and hematuria in his diaper. The abdomen was distended. A mass measuring approximately 10x10 cm in the left kidney lodge and approximately same size mass in the right kidney lodge were palpated. In the abdominal ultrasonography, cystic masses, approximately 10x6 cm in size in the upper half of the right kidney and 12x6.5 cm in the left kidney, consisting of multiple cysts separated from each other by multiple thin septa and causing significant suppression and thinning in the adjacent parenchyma were observed. Abdominal tomography revealed a well-circumscribed cystic mass of 11x9.5x8 cm in the right kidney and 12x8x9.5 cm in the left kidney, with multiple

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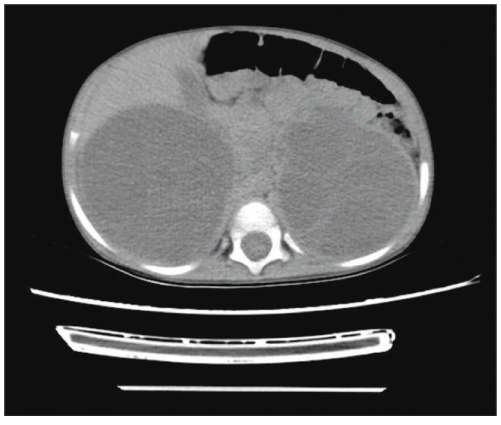

FIGURE 1: Abdominal tomography.

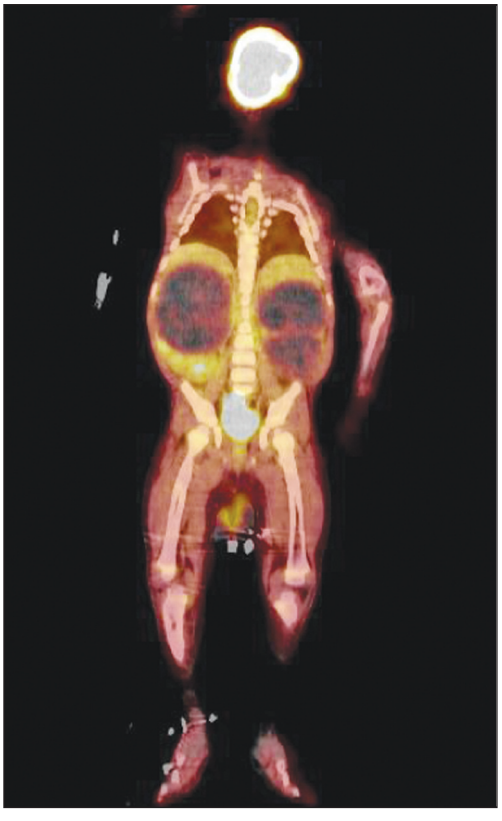

FIGURE 2: F-18 positron emission tomography.

fine septations and macrolobule contours in the left kidney (Figure 1). No solid component was found in the mass. CN and cystic nephroblastoma were considered in differential diagnosis. The F-18 positron emission tomography showed the kidney tissue in the small area in the lower pole of the right kidney, and no significant activity uptake was observed in the cystic lesion with septa. Metabolic activity was not detected in the lesions (Figure 2).

Radiologically and clinically, the distinction between CN and nephroblastoma could not be made. Since there was no intact parenchyma tissue in the left kidney, a left nephrectomy was performed. CN was the definite diagnosis following the histopathological examination. Parenchymal echogenicity and thickness of the right kidney were normal in the lower half. No growth was observed in the cystic mass in the right kidney in the 1st, 3rd and 6th month follow-ups. The patient had hypertensive values in the clinical follow-up. Antihypertensive was started. A slight increase in creatinine values was observed in laboratory findings.

DICER1 gene mutation is frequently seen in bilateral cases, and *DICER1* gene mutation was also positive in our case. Since the gene has a predisposition to malignancy, partial nephrectomy was planned on the right side.

DISCUSSION

In childhood CN may develop, a rare benign multicystic kidney tumor, especially in boys between the ages of three month and twenty four months. Approximately two-thirds of CN cases are children.[1] In a comprehensive WT study, the frequency of CN is reported to be less than 1%.[2]

It is important to distinguish between benign and malignant diagnoses in the differential diagnosis of cystic kidney diseases seen in early childhood. Clinically and radiologically, CN may not be differentiated from malignant tumors, and the definitive diagnosis is made only by histopathological examination.[1] Differential diagnosis is important in kidney masses, and early diagnosis is of great importance in cases that are confused with each other. The diagnosis of the mass could not be made in our patient by performing abdominal tomography, magnetic resonance imaging, F-18 positron emission tomography. The diagnosis was made histopathologically by left nephrectomy.

CN is usually a unilateral and sporadic tumor. It is usually seen as a single large (2-14 cm) mass macroscopically, bilateral involvement is rare. The exact pathogenesis is unknown. An association with

a mutation in the familial *DICER1* gene was found in bilateral cases. *DICER1* syndrome is a dominantly inherited disease. *DICER1* gene mutation was also detected in the father of our patient. Patients are at risk of developing pleuropulmonary blastoma, multinodular goiter, thyroid cancers, testicular cancers.[3] It has not been detected in our patient for the time being.

In children, the appearance is comparable to Wilms tumor. It may present with abdominal pain, abdominal mass, hematuria or hypertension.[4,5] In our case, it was noticed with an abdominal mass and hematuria.

The purpose of the operative procedure in the CN is to remove the tumor tissue while preserving the normal renal parenchyma. Partial nephrectomy or enucleation may be attempted to preserve functional renal parenchyma. However, if the CN has affected most of the kidney, total nephrectomy is indicated. It is not malignant. There is no need for additional treatment after the surgical procedure.[5-7] In their experience with nine pediatric multiloculated cystic tumors, Kurian et al. underlined the diagnostic and therapeutic dilemma associated with these tumors and suggested a similar surgical approach.[8] Van den Hoek et al. suggested that surgery should be considered first in cystic neoplasms without solitary components and enucleation should be performed without further treatment if possible.[9] They stated that partial excision can be applied in rare bilateral cases.[10]

In our case, there was a large mass involving the entire left kidney and extending to the hilum. Left nephrectomy was performed. The mass was limited to the upper pole of the right kidney in the follow-up. The studied *DICER1* gene mutation was positive. Since the gene is also associated with pleuropulmonary blastoma, thyroid and testicular cancers, partial nephrectomy was planned.

Source of Finance

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Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

Idea/Concept: *Dilşad Dereli;* ***Design:*** *Çiğdem Arslan Alıcı;* ***Control/Supervision:*** *Baran Tokar;* ***Data Collection and/or Processing:*** *Dilşad Dereli;* ***Analysis and/or Interpretation:*** *Nuran Çetin;* ***Literature Review:*** *Aslı Kavaz Tufan;* ***Writing the Article:*** *Dilşad Dereli;* ***Critical Review:*** *Nuran Çetin;* ***References and Fundings:*** *Baran Tokar;* ***Materials:*** *Dilşad Dereli.*

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