CASE REPORT OLGU SUNUMU

Wilms' Tumor in a Child with Cystic Fibrosis: A Case Report and Review of the Literature

Kistik Fibrozisli Bir Çocukta Wilms Tümörü: Olgu Sunumu ve Literatür Taraması

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Abstract

Cystic fibrosis (CF) is the most common life-limiting autosomal recessive disorder. While gastrointestinal and pulmonary complications are frequent in CF, the coexistence of malignancy is rare. We aimed to present a case of bilateral Wilms' tumor (BWT) in a child with CF and to review the relevant literature. A female child diagnosed with CF in the neonatal period was referred to our clinic at the age of 2 years, when bilateral Wilms' tumor was detected. Clinical history, laboratory findings, and imaging results were evaluated, and the diagnostic, therapeutic, and follow-up process were described in detail. In addition, a literature review was performed to identify similar cases.In addition to the typical clinical manifestations of CF, abdominal imaging revealed bilateral Wilms' tumor. The clinical course and treatment process of the patient were reported. Literature review demonstrated that the coexistence of CF and Wilms' tumor is extremely rare, with only a limited number of cases described. Although malignancy is rare in children with CF, it should not be overlooked. This case highlights the unusual coexistence of CF and Wilms' tumor and emphasizes that reporting such cases provides a valuable contribution to the literature. Clinicians should consider the possibility of malignancy during the long-term follow-up of patients with CF, particularly as survival improves.

Öz

Kistik fibroz (KF), yaşamı sınırlayan en yaygın otozomal resesif hastalıktır. KF'de gastrointestinal ve pulmoner komplikasyonlar sık görülürken, malignite birlikteliği nadirdir. KF'li bir çocukta bilateral Wilms tümörü (BWT) olgusunu sunmayı ve ilgili literatürü incelemeyi amaçladık. Yenidoğan döneminde KF tanısı almış bir kız çocuğu, 2 yaşındayken bilateral Wilms tümörü tespit edilmesi üzerine kliniğimize sevk edildi. Klinik öykü, laboratuvar bulguları ve görüntüleme sonuçları değerlendirildi ve tanı, tedavi ve takip süreci ayrıntılı olarak anlatıldı. Ayrıca benzer vakaları belirlemek için literatür taraması yapıldı. KF'nin tipik klinik bulgularına ek olarak, abdominal görüntülemede bilateral Wilms tümörü saptandı. Hastanın klinik seyri ve tedavi süreci raporlandı. Literatür taraması, KF ve Wilms tümörü birlikteliğinin son derece nadir olduğunu ve sınırlı sayıda vakanın tanımlandığını

Keywords
Wilms' tumor, child, cystic fibrosis

Anahtar kelimeler
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göstermiştir. Kistik fibrozisli çocuklarda malignite nadir görülse de göz ardı edilmemelidir. Bu olgu, Kistik fibrozis ve Wilms tümörünün alışılmadık bir şekilde bir arada bulunmasını vurgulamakta ve bu tür olguların bildirilmesinin literatüre değerli bir katkı sağladığını vurgulamaktadır. Klinisyenler, özellikle sağ kalım arttıkça, Kistik fibrozisli hastaların uzun dönem takibinde malignite olasılığını göz önünde bulundurmalıdır.

Introduction

Cystic fibrosis (CF) is the most common life-limiting autosomal recessive disease affecting the pancreas, intestine and hepatobiliary system, and respiratory tract in Caucasians. CF is caused by different mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. With the use of modulators aimed at correcting the CFTR protein, the prognosis of CF has improved markedly over the past 20 years. In this way, the number of patients reaching adulthood is increasing. Approximately 85% of patients with CF have pancreatic insufficiency at birth. Gastroesophageal reflux disease (GERD) is more common in patients with CF but is thought to adversely affect lung health. Distal small bowel obstruction syndrome (DIOS), caused by meconium-like stool plugs, occurs at any age after the neonatal period and affects 15-20% of patients with CF (1). With the increasing survival rate, many patients with CF, especially gastrointestinal system cancers, are more common in advanced age compared with the normal population. Recently, it has been reported that different types of cancer are seen during the follow-up of patients CF in the pediatric age group (2,3). In this report, by reviewing the literature, we evaluated a bilateral Wilms tumor (BWT) detected in a 2-year-old girl who was followed up for CF.

Case Report

A female patient, who has been followed up with the diagnosis of CF since the neonatal period, was referred to our clinic when she was aged 2 years when bilateral hydronephrosis and a mass in both kidneys were detected in a routine abdominal ultrasonography follow-up.

When the first "immune reactive trypsinogen testing" (IRT) was found as 102 µg/L and the second IRT as 83 µg/L for the CF screening program in the neonatal period, the patient was given a sweat test. The patient was followed up when the chlorine amount was intermediate (43 eq/L) in the sweat chlorine test. In the blood tests performed during the follow-up of the patient (Na: 127 mmol/L, Cl: 96 mmol/L, Ph: 7.53, HCO3: 29 mmol/L), genetic analysis was performed and when the G85E/F1052V gene mutation was found to be a compound heterozygous mutation, she was diagnosed as having CF and treatment was started. It was confirmed through gene analysis that both parents were CF carriers.

Newborn CF screening thresholds for immunoreactive trypsinogen (IRT) vary across programs; representative fixed cutoffs are typically around 60–65 ng/mL (≈96th percentile), while some use fixed cutoffs spanning 50–100 ng/mL or floating percentiles (≈95th–99.5th). In our patient, IRT values were 102 and 83 µg/L (i.e., ng/mL), exceeding common screening thresholds.

In the physical examination of the patient, a mass was palpated in both flank regions. In abdominal ultrasonography, hydronephrosis was present in both kidneys and the larger one on the right was 4x4.5 cm. It was seen that there were multiple kidney masses on the left, the largest of which was 5x6 cm. In abdominal magnetic resonance imaging (MRI), multiple masses were detected in both kidneys (Figure 1, A-D).

She was started on chemotherapy with vincristine, actinomycin-D, and doxorubicin. Since the patient responded well, the Oncology Board decided to continue treatment beyond the usual 6–12 week window, and surgery was performed at week 25.

At that time, a left nephrectomy was performed due to multiple tumor masses, and nephron-sparing surgery was carried out on the right kidney. Pathology confirmed a regressive-type Wilms Tumor without anaplasia. Radiotherapy was applied to the left kidney lodge, while it was not administered to the right. The patient is followed up in the 3rd month after surgery, disease-free and without signs of kidney failure.

Pathology results confirmed a regressive-type Wilms Tumor without anaplasia (Figure 2). There was no tumor tissue in the surgical margins. A total of 10.8 Gy radiotherapy was applied to the left kidney lodge of the patient after the surgery. Radiotherapy was not administered to the right kidney lodge. The patient is followed up in the 3rd month after the surgery, free of disease and without signs of kidney failure.

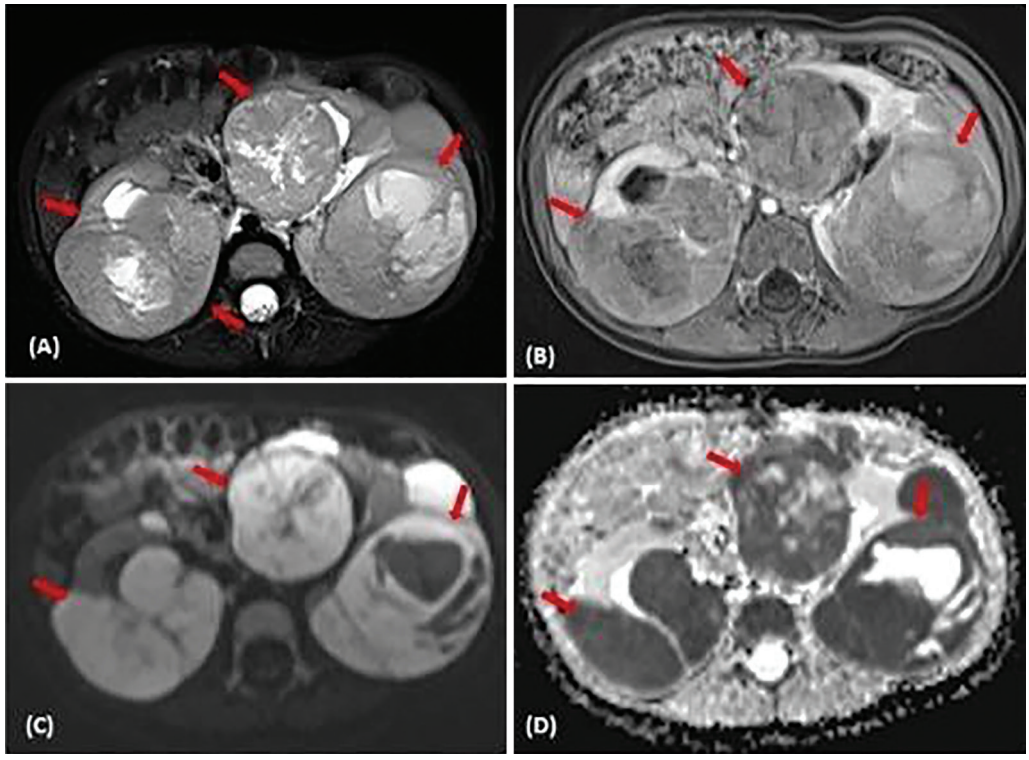

Figure 1. A-D. In both kidneys, the largest 38x45 mm on the right and 48x58 mm on the left, multiple parenchymal exophytic extensions in the collecting system with cystic and solid areas causing local dilatation and deformation, isointense with the kidney parenchyma in T2-weighted images (A), showing mild contrast enhancement on contrast-enhanced T1-weighted images (B), and marked diffusion restriction on diffusion-weighted images (C, D) solid space-occupying mass lesions are observed (red arrows)

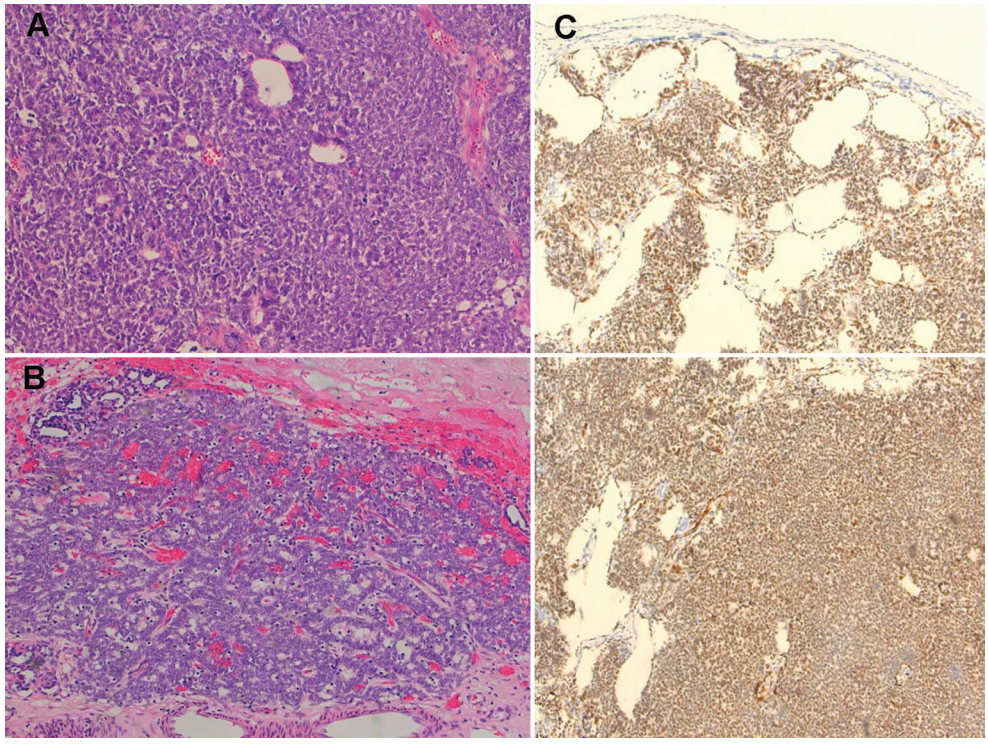

Figure 2. A: Extensive blastemal component in tumor tissue (Hematoxylin-eosin, original magnification, X40) B: Extensive epithelial component in tumor tissue (Hematoxylin-eosin, original magnification, X40)

C: Diffuse nuclear staining of tumor cells (Immunohistochemistry, WT-1 stain, original magnification, X20)

Discussion

CF is known to increase the risk of gastrointestinal cancers when compared with the general population, including specific cancers of the small intestine, colon, biliary tract, and pancreas. A screening strategy for gastrointestinal cancer needs to be developed in these patients (4).

Recent studies (2022–2024) have confirmed that while overall cancer risk in CF is increasing, non-GI malignancies such as Wilms tumor remain exceptionally rare. Updated reports emphasize the need for individualized management in bilateral Wilms tumor (BWT), with surgery generally recommended at week 6–12; in our case, the Tumor Board extended chemotherapy to week 25 due to ongoing favorable response. Importantly, no lung metastases were detected, and our follow-up period remains short (3 months), which is a limitation.

A study, which showed that there are various types of cancer in people with CF and its prevalence as 1.1%, stated that these types of cancer occur at a younger age than the general population. Although GI cancer is the most common, it still accounts for only 22% of total cancers (5). Although some studies did not demonstrate the relationship between existing cancer and CF (2), other authors noted that most of their patients had some degree of pancreatic insufficiency and impaired intestinal motility, and persistent pathologic changes associated with increased cell turnover. It has been shown that these changes can cause cancer and the incidence of non-gastrointestinal cancers is high (6). The coexistence of CF and Wilms tumor has been reported in only two patients in the literature, and the patients died in the early period (7). Our patient was diagnosed as having BWTs when she was aged only 2 years and she had no gastrointestinal tract symptoms.

Prospective evaluation of serum amylase and lipase measurements in all patients with newly diagnosed non-Hodgin lymphoma has been reported to be of interest to define the incidence of subclinical pancreatic disease and should bring to mind undiagnosed CF disease (8). Our patient had no signs of pancreatitis and her amylase lipase values were normal.

The coexistence of CF and acute lymphocytic leukemia (ALL) is rare and few cases have been reported in the literature (9-11). For ALL in patients with CF, the treatment of patients with severely impaired lung function should be individualized and, because teniposide clearance rates may be higher than population averages, assessment of systemic drug exposure and dose adjustment based on these findings are recommended (10). It has also been stated that some immune modulation and anti-inflammatory treatments given together with leukemia treatment are effective in lung problems caused by CF (11). Our patient had no lung problems that required treatment yet.

In another study on cancers accompanying congenital anomalies, it was stated that the possible leukemogenic effect of chloramphenicol, which is used in the treatment of CF in children with leukemia and Wilms tumor, should be considered and further studies should be conducted (7). Today, chloramphenicol is no longer used for the treatment of CF. Our patient was on medication containing only high doses of pancreatic enzymes for CF.

In the literature, cases of testicular, brain, and thyroid cancers, and neuroblastoma have also been reportedly seen with CF. The negative effects of the agents given during cancer treatment on the lung are important in patients with CF (6,12). Our patient had no lung disease due to chemotherapeutic drugs during the treatment period.

For BWTs, both the Current Children's Oncology Group (COG) and the International Society of Pediatric Oncology (SIOP) recommend preoperative chemotherapy, NSS, and subsequent treatment (13). Appropriate chemotherapy was given to our patient before the surgery, and radiotherapy was given after the surgery.

The COG and SIOP protocols mandate the removal of the entire tumor as well as lymph node sampling. Ipsilateral nephrectomy is usually performed and therefore NSS is reserved for BWT because of the size of unilateral tumors (and the fact that protocols mandate pre-chemo only of SIOP, not COG). Pre-administered chemotherapy allows the tumor to shrink significantly and makes it technically easier to separate the tumor from the surrounding normal renal parenchyma. The approach chosen should allow exposure of the retroperitoneum to facilitate tumor excision and lymph node sampling. A transperitoneal approach via a chevron incision can be used. It is necessary to protect the adrenal gland as long as it does not compromise oncologic control or other aspects of surgical quality (14). Laparoscopic partial nephrectomy for Wilms tumors has been reported, although there have been reports of large peritoneal spread by the tumor following minimally invasive NSS (15-17). Currently, the open surgical approach for NSS in patients with BWT remains the standard. Our patient first underwent a left total nephrectomy because there was more than one tumor mass in the left kidney, and NSS was not possible. Afterward, approximately 1.5 cm of tumor tissue in the mid-lower pole of the right posterior face was removed with NSS, and the double-J stent, which was placed after the opened pelvis

was repaired, was removed in the 3rd postoperative week. Both adrenal glands were not removed. There was no complication related to the surgical procedure. Renal failure did not develop.

Gastrointestinal system cancers can be seen in advanced age with CF. The association of non-GI system cancers with CF has been reported very rarely. Moreover, the appearance of these cancers at an early age indicates that patients with CF should be followed closely and with a multidisciplinary team. The association of CF Wilms' tumor has been reported in only a few cases in the literature. We state that since our patient had BWT, the diagnosis and treatment process were successfully managed and it is pleasing that she continued her life without developing kidney failure with NSS.

Conclusion

To date, only two cases of Wilms tumor associated with CF have been explicitly reported in the literature. Miller (1969) described two children with CF who developed Wilms tumor, both of whom died in the early period. Subsequent epidemiological reports discussed cancer risk in CF but did not provide further well-documented WT cases. Our patient therefore represents one of the very few surviving cases with detailed diagnostic and therapeutic follow-up, highlighting the exceptional rarity of this comorbidity.

Footnotes

Conflict of Interest: The authors reported no potential conflict of interest.

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References

1. Ley D, Turck D. Digestive outcomes in Cystic fibrosis. Best Pract Res Clin Gastroenterol. 2022;56-57:101788.
2. Robertson CM, Hawkins MM. Childhood cancer and cystic fibrosis. J Natl Cancer Inst. 1995;87:1486-7.
3. Neglia JP, Wielinski CL, Warwick WJ. Cancer risk among patients with cystic fibrosis. J Pediatr. 1991;119:764-6.
4. Yamada A, Komaki Y, Komaki F, Micic D, Zullow S, Sakuraba A. Risk of gastrointestinal cancers in patients with cystic fibrosis: a systematic review and meta-analysis. Lancet Oncol. 2018;19:758-67.
5. Archangelidi O, Cullinan P, Simmonds NJ, Mentzakis E, Peckham D, Bilton D, et al. Incidence and risk factors of cancer in individuals with cystic fibrosis in the UK; a case-control study. J Cyst Fibros. 2022;21:302-8.
6. Schöni MH, Maisonneuve P, Schöni-Affolter F, Lowenfels AB. Cancer risk in patients with cystic fibrosis: the European data. CF/CSG Group. J R Soc Med. 1996;89 Suppl 27:38-43.
7. Miller RW. Childhood cancer and congenital defects. A study of U.S. death certificates during the period 1960-1966. Pediatr Res. 1969;3:389-97.
8. Lucero MA, Murphy T, Faro A, Kocoshis SA, Blatt J. Pancreatitis and an abnormal sweat test at presentation in a child with non-Hodgkin's lymphoma. Pancreas. 1996;12:205-6.
9. Rizzari C, Conter V. "Childhood ALL and cystic fibrosis—treatment and outcome". Medical and Pediatric Oncology. 1995;25:223.
10. Gururangan S, Horner M, Rodman JH, Marina NM. Successful treatment of acute lymphoblastic leukemia in a child with cystic fibrosis. Med Pediatr Oncol. 1994;22:414-6.
11. Dopfer C, Kentouche K, Gruhn B, Dopfer R, Becker S, Mentzel HJ, Beck JF, Mainz JG. Acute lymphoblastic leukemia in a 10-year-old boy with cystic fibrosis - improvement of pulmonary function during chemotherapy in spite of intermittent Pseudomonas aeruginosa colonisation. Klin Padiatr. 2011;223:368-9.
12. Naselli A, Cresta F, Favilli F, Casciaro R. A long-term follow-up of residual mass neuroblastoma in a patient with cystic fibrosis. BMJ Case Rep. 2015;2015:bcr2014205284.
13. Meng D, Chang X, Ren Q, Xu J, Wang H. Congenital bilateral wilms tumor: a case report. Urology. 2021;157:242-5.
14. Kieran K, Davidoff AM. Nephron-sparing surgery for bilateral Wilms tumor. Pediatr Surg Int. 2015;31:229-36.
15. Piché N, Barrieras D. Minimally invasive nephron-sparing surgery for unilateral Wilms tumor. J Pediatr Surg. 2012;47:E1-4.
16. Rauth TP, Slone J, Crane G, Correa H, Friedman DL, Lovvorn HN 3rd. Laparoscopic nephron-sparing resection of synchronous Wilms tumors in a case of hyperplastic perilobar nephroblastomatosis. J Pediatr Surg. 2011;46:983-8.
17. Chui CH, Lee AC. Peritoneal metastases after laparoscopic nephron-sparing surgery for localized Wilms tumor. J Pediatr Surg. 2011;46:e19-21.