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EDİTÖRE MEKTUP / LETTER TO THE EDITOR

Wilms' tumor and nephrotic syndrome

Wilms tümörü ve nefrotik sendrom

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Dear Editor,

Wilms' tumor (WT) is the most frequent malignant renal tumor in pediatric age group. Approximately 10% of cases have congenital anomalies, and small proportion of them is a part of a congenital malformation syndrome[1]. Denys-Drash syndrome (DDS), Fraiser syndrome (FS), WAGR syndrome (WT, aniridia, genitourinary malformations and mental retardation) and Beckwith-Wiedemann syndrome (BWS) are among these syndromes. Nephrotic syndrome (NS) characterized by proteinuria, hypoalbuminemia, hyperlipidemia, and edema and is the most common childhood glomerular disease. NS is divided into steroid-sensitive nephrotic syndrome and steroid-resistant nephrotic syndrome (SRNS) based on the patient's response to steroid treatment[2].

In this report, an unusual case of WT associated with NS developed 30 months after the diagnosis of WT is described. A 4-year-old boy was admitted with a complaint of hematuria. Physical examination was normal and computed tomography showed a solid mass originating from the right kidney. The patient have not had aniridia, hemihypertrophy, ambiguus genitalia or any physical stigmata of DDS, FS or BWS. Patient and family history were unremarkable. His thoracal computed tomography revealed no pulmonary metastasis. After resection of the renal tumor, pathologic examination revealed Wilms' tumor with negative surgical margins and without anaplasia. The patient was treated with chemotherapy consisting of vincristine and actinomycin-D for 6 months. Two years after cessation of treatment, control laboratory tests indicated proteinuria in nephrotic range. The patient did not respond to either steroids or immunosuppressive agents and progressed to end-stage renal disease within 10 years after disease onset. A renal biopsy and WT1 mutation analysis was refused by his parents. Regular haemodialysis was started and control abdominal images showed no recurrence of WT.

The occurrence of nephrotic syndrome and Wilms' tumour in a 6-year-old child is suggestive for evaluation for syndromic associations. Evolution towards end-stage renal disease occurred in 16 years of age in our patient. In literature most of the children with WT and nephropathy were associated with DDS or FS and most of them have been reported to carry WT1 mutations[3]. DDS is characterized by ambiguus genitalia, diffuse mesangial sclerosis and/or WT. FS on the other hand includes focal and segmental glomerulosclerosis, streak gonads, ambiguus genitalia and gonadoblastoma[4]. Our patient did not have the stigmata consistent with the syndromes described above. Unfortunately we have not had tissue diagnosis to better understand the pathologic mechanism existed, because of the parents concern about the risks in kidney biopsy in a unilateral nephrectomized patient.

Most children with sporadic NS respond to steroids and have a favorable long-term prognosis; however, 10-20% of patients do not respond to steroids and

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may progress to end-stage renal disease. Mutations in genes encoding podocyte proteins such as NPHS1, NPHS2, CD2AP and WT1 are responsible for SRNS in children[5]. There is currently no guideline regarding whether all patients with SRNS should be routinely subjected to WT1 mutation analysis. Clinicians should be aware of clinical symptoms and laboratory findings of NS in patients with WT in outpatient follow-up. Kidney biopsy and WT1 mutation tests are warranted to exclude congenital malformation syndromes.

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