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LETTER TO THE EDITOR

A Rare Intersection: Wilms Tumor in a Patient with PIGO Syndrome

Şefika Akyol[1,2] | Özge Vural[3] | Gülsüm Kayhan[4] | Zafer Türkyılmaz[5] | Alper Özcan[1] | Deniz Koçak Göl[1] | Güçlü Pınarlı[3] | Ekrem Ünal[1,6]

[1]Department of Pediatric Hematology and Oncology, Erciyes University Faculty of Medicine, Kayseri, Turkey | [2]Pediatric Hematology and Oncology Clinic, Antalya Research and Training Hospital, Health Sciences University, Antalya, Turkey | [3]Department of Pediatric Hematology and Oncology, Gazi University Faculty of Medicine, Ankara, Turkey | [4]Department of Medical Genetics, Gazi University Faculty of Medicine, Ankara, Turkey | [5]Department of Pediatric Surgery, Gazi University Faculty of Medicine, Ankara, Turkey | [6]Department of Health Sciences, Hasan Kalyoncu University, Pediatric Hematology and Oncology, Gaziantep, Turkey

Correspondence: Ekrem Ünal (drekremunal@yahoo.com.tr)

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To the Editor

A 4-year-old girl was admitted with a complaint of abdominal pain and diagnosed with a mass in her left kidney with dimensions of 42 × 38 × 60 mm. Her medical history revealed that she had antenatal hydronephrosis leading to bilateral double J catheter implementation. One year before admission, the patient underwent an uroterostomy operation. No metastasis was present in her chest computed tomography (CT) imaging. She underwent surgery for a total nephrectomy of her left kidney. The pathology results revealed a stage I Wilms tumor (WT) with favorable histology. She received chemotherapy in accordance with the National Wilms Tumor Study 5 [1]. In the 7th month of follow-up, a relapse was detected in the left renal bed with the dimensions 67 × 72 mm. Surgery was implemented and total resection of the tumor was performed. Pathology results disclosed WT containing blastemal component with no anaplasia, lymph node, and vascular involvement. The second-line chemo-radiotherapy protocol was prepared according to the SIOP Umbrella Study [2]. A total of 13.5 Gy radiotherapy was applied over 9 days with a daily dose of 1.5 Gy fractions for the entire abdomen using 6 MV photon energy in a linear accelerator. She completed her second-line treatment and is still in remission.

Owing to intellectual disability, extremity anomalies, epilepsy, and hydronephrosis, whole genome sequencing was implemented. No mutation known to predispose to WT was detected. However, biallelic pathogenic variants were detected in the *PIGO* gene. The c.2708_2710delITCT (p.Phe903del) (de novo) and c.1944C>G (p.Cys648Trp) (maternal) variants detected as compound heterozygous in the patient are compatible with hyperphosphatasia with impaired intellectual development syndrome-2 (OMIM #614749).

PIGO syndrome is an autosomal recessively inherited disease caused by the mutations in the *PIGO* gene encoding the phosphatidylinositol glycan anchor biosynthesis class O protein synthesis [3]. The disease is mainly a congenital glycosylation disorder affecting the synthesis of glycosylphosphatidylinositol anchor. The syndrome is reported to be associated with dysmorphism, psychomotor disability, epilepsy, and hyperphosphatasemia. No predisposition to solid tumor development has been reported yet. Nevertheless, WT is one of the embryonal tumors of childhood and is generally associated with predisposing syndromes not only in the *WT1* gene and 11p15 locus but also in the mutations leading to other cancer-predisposing syndromes like DICER1, Li-Fraumeni, and Bloom syndrome, and Fanconi anemia [4, 5].

To the best of our knowledge, the current patient is the first patient in literature with PIGO syndrome who developed a WT and also experienced a localized relapse within the first year.

Author Contributions

All authors certify that she/he has participated sufficiently in the intellectual content and the analysis of data. All the authors took part in the follow-up and care of the patient. Each author has reviewed the final version of the manuscript and approved it for publication.

Acknowledgment

The authors have nothing to report.

Conflicts of Interest

The authors declare no conflicts of interest.

Consent

The family of the patient signed the free and informed consent form.

Data Availability Statement

The data are not publicly available due to privacy or ethical restrictions.

Şefika Akyol
Özge Vural
Gülsüm Kayhan
Zafer Türkyılmaz
Alper Özcan
Deniz Koçak Göl
Güçlü Pınarlı
Ekrem Ünal

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