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LETTER TO THE EDITOR

Peritoneal dialysis as an effective kidney replacement therapy in Joubert syndrome

Joubert sendromunda etkili bir böbrek replasman tedavisi olarak periton diyalizi

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To the Editor,

Joubert syndrome is a rare autosomal recessive ciliopathy characterized by cerebellar vermis hypoplasia, developmental delay, and multisystem involvement. Kidney disease, most commonly nephronophthisis-like tubulointerstitial nephropathy, occurs in a substantial proportion of patients and represents a major determinant of prognosis. Kidney involvement may initially be subtle but often progresses to chronic kidney disease (CKD) and eventually end-stage kidney disease during childhood or adolescence[1,2]. We report a pediatric patient with genetically confirmed Joubert syndrome who developed progressive kidney disease and was subsequently managed with peritoneal dialysis (PD).

A 10-year-old boy with a clinical diagnosis of Joubert syndrome was referred to our pediatric nephrology outpatient clinic for continuation of care due to progressive CKD. He was born at term after an uneventful pregnancy and delivery. At one month of age, renal ultrasonography reportedly demonstrated cystic kidney changes. During subsequent evaluations, the presence of syndromic facial features prompted further investigation for an underlying neurogenetic disorder. Brain magnetic resonance imaging revealed cerebellar vermis hypoplasia, a characteristic "molar tooth" sign at the mesencephalon, and supracerebellar central-type atrophy, findings considered compatible with Joubert syndrome (Figure 1). Genetic testing was performed and *CEP290* gene c.5668G>T (p.G1890*) homozygosis mutation was revealed.

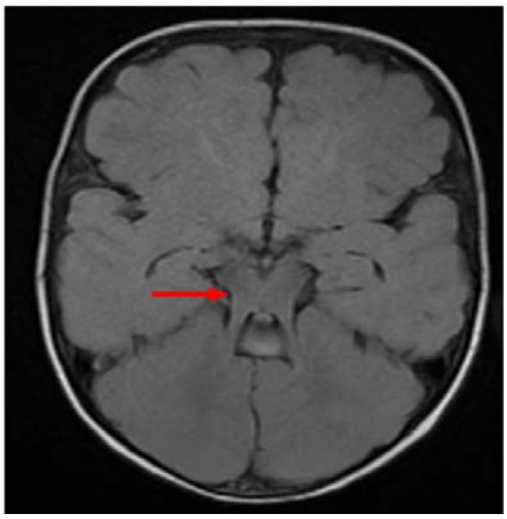


Figure 1. Brain MRI showing cerebellar vermis hypoplasia and the molar tooth sign (red arrow), consistent with Joubert syndrome.

Following the early diagnosis, the patient remained largely asymptomatic and did not require regular nephrology care until approximately one year prior to presentation, when routine laboratory tests first revealed elevated serum creatinine levels. At presentation to our clinic, he had no active complaints and preserved urine output, while physical examination revealed a syndromic

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appearance with neurodevelopmental involvement and obesity.

Initial laboratory evaluation revealed blood urea nitrogen of 39.4 mg/dL, serum creatinine of 2.31 mg/dL, and an estimated glomerular filtration rate of approximately 23 mL/min/1.73 m², findings consistent with CKD stage 4. The patient also demonstrated biochemical evidence of CKD–bone mineral disorder (BMD), with markedly elevated parathyroid hormone levels (PTH: 418 pg/mL), suggesting long-standing kidney involvement.

An abdominal ultrasonography documented that the sizes of both kidneys were consistent with the patient's age, although the renal parenchyma showed markedly increased echogenicity, without hydronephrosis or visible solid/cystic mass lesions.

The patient was started on appropriate CKD supportive therapy, including agents targeting bone mineral metabolism, and was placed under close follow-up. During follow-up, progressive deterioration in kidney function was observed. In October 2025, when the estimated glomerular filtration rate declined to approximately 8 mL/min/1.73 m² (serum creatinine 6.7 mg/dL), kidney replacement therapy was initiated. In the absence of a suitable living kidney donor for preemptive transplantation, PD was initiated as the preferred modality of kidney replacement therapy. The patient was enrolled in a PD program, and a catheter was inserted laparoscopically by the pediatric surgery team. Structured training was provided to the family both before and after the procedure, resulting in good adherence and rapid adaptation to the dialysis regimen. The patient was subsequently initiated on automated peritoneal dialysis (APD) using a standard 1.36% glucose-based dialysis solution, and residual urine output was preserved at the initiation of dialysis (approximately 1000 mL/day). The initial prescription consisted of six nightly exchanges with a dwell volume of 750 mL/m² and a dwell time of approximately 100 minutes, resulting in a total treatment duration of about 10 hours. During the initial peritoneal equilibration test (PET), the Kt/V value was 1.7, creatinine clearance 30.7 L/1.73m²/week, and the peritoneal membrane was classified as low average transport type. Daily ultrafiltration (UF) volumes ranged between 250 and 300 mL.

From the time of catheter placement to the present, the patient demonstrated excellent adherence to therapy. The family successfully completed dialysis training, and PD has been continued without interruption. No PD-related complications have been observed, and the patient's daily comfort and functional status have remained stable, with preservation of quality of life. Dialysis adequacy assessments confirmed that the PD prescription was sufficient.

In conclusion, this case highlights that PD can be a safe and effective kidney replacement therapy in children with Joubert syndrome and end-stage kidney disease. Consistent with our experience, previous reports have also demonstrated the feasibility and safety of PD in children with Joubert syndrome who develop chronic kidney disease particularly in those with accompanying neurocognitive impairment. [3-5]. Joubert syndrome is frequently associated with hypotonia and abnormal respiratory patterns, particularly episodic tachypnea and apnea[2]. These neurological features may theoretically complicate peritoneal dialysis by increasing the risk of abdominal wall weakness or hernia formation and by influencing respiratory mechanics due to increased intra-abdominal pressure. However, in our patient, PD was well tolerated without respiratory compromise or mechanical complications. Previous studies have shown that PD provides greater hemodynamic stability, preserves residual renal function, enables home-based treatment, and has been shown to be more cost-effective than hemodialysis in several settings, thereby supporting quality of life, growth, and neurodevelopment[6,7]. Our experience further suggests that, with appropriate family education and close follow-up, PD can be successfully implemented even in children with complex neurodevelopmental disorders.

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