Continuous Ambulatory Peritoneal Dialysis for a Patient With Bardet-Biedl Syndrome

Hossein Emad Momtaz, Ali Amanati

Division of Pediatric Nephrology, Hamedan University of Medical Sciences, Hamedan, Iran

Keywords. Bardet-Biedl syndrome, chronic kidney disease, end-stage renal disease, continuous ambulatory peritoneal dialysis

Bardet-Biedl syndrome (BBS) is a multisystem syndrome with a range of primary and secondary features. Kidney abnormalities are a major cause of morbidity and mortality in BBS and it is a significant genetic cause of chronic kidney disease in children. Conventional approach to end-stage renal disease in these patients is hemodialysis and kidney transplantation afterwards. Continuous ambulatory peritoneal dialysis, however, is not a commonly advocated modality in literature. In this report, we present a boy with BBS who underwent continuous ambulatory peritoneal dialysis, which resulted in control of kidney function impairment and better compliance for his family than hemodialysis before kidney transplantation. Of note, this is a rare case of BBS complicated with end-stage renal disease in the first decade of life.

IJKD 2008;2:237-9
www.ijkd.org

Case Report

INTRODUCTION

Bardet-Biedl Syndrome (BBS) is defined by a range of primary and secondary features. It primary features include rod-cone dystrophy, postaxial polydactyly, truncal obesity, hypogonadism, and kidney abnormalities and its secondary manifestations are speech disorder/delay, developmental delay, ataxia/imbalance, diabetes mellitus, congenital heart defects, liver disease, hearing loss, facial features, situs inversus, Hirschsprung disease, polyuria/polydipsia, dental crowding, and anosmia.[1-3] Renal involvement is observed in most affected individuals. The most common causes of kidney dysfunction are tubular impairment of the urine concentrating and acidification ability and aminoaciduria.[4] In addition, hypertension has been frequently observed.[5] Bilateral renal enlargement and increased renal parenchymal echogenicity are early findings of imaging studies, whereas after 12 months, the kidney size regresses significantly.[6]

Management of kidney problems includes control of hypertension, treatment of recurrent urinary tract infections, correction of electrolyte imbalance, and treatment of end-stage renal disease (ESRD) by early diagnosis and renal replacement therapy.[5,7] There is little information on the method of choice, especially on continuous ambulatory peritoneal dialysis (CAPD), for renal replacement therapy in these patients. Hereby, we report a boy with BBS who presented with ESRD in his 1st decade of life and underwent CAPD.

CASE REPORT

A 9-year-old boy was admitted to our hospital with loss of consciousness and elevated blood urea nitrogen (BUN) and serum creatinine levels. He had a family history of the death of 3 siblings; the first child was a girl who had died at the age of 6 month and other children were 2 boys with prominent obesity died in the neonatal period. The patient had 4 other siblings were alive with no specific problem. He had a positive history of polydypsia and frequency in his early infancy and visual problems including poor distinction of different colors and nystagmus. At the age of 4 years, he had been hospitalized because of clinically morbid obesity and polydipsia, diagnosed

as nephrogenic diabetes insipidus with the help of water deprivation test.

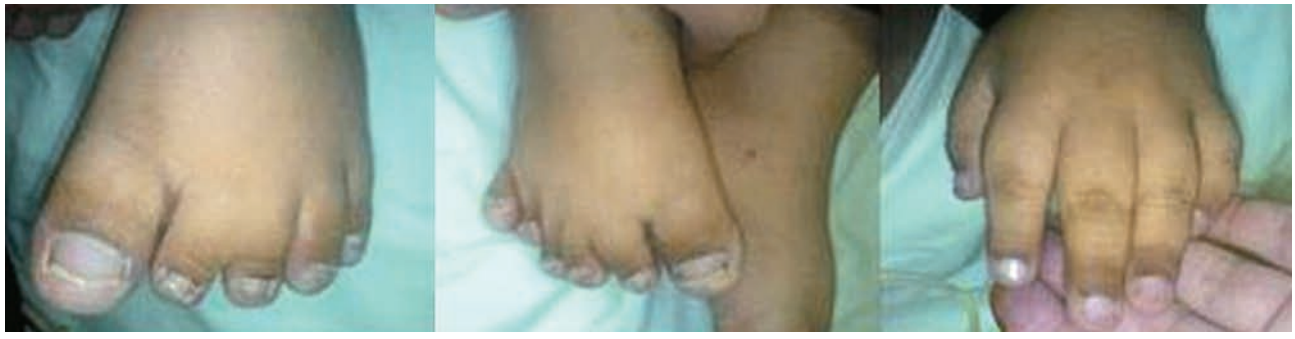
He had microtestis and microfallus on genital examination, and retinitis pigmentosa on ophthalmologic examination. Other clinical findings were polydactyly, syndactyly, and brachydactyly (Figure). The patient also suffered from behavioral problems reflecting as irritability. He had mild mental retardation and was studying in special school. According to the clinical and paraclinical evidence, he met all the 6 cardinal or primary criteria and 6 minor or secondary criteria of BBS diagnosis. In his previous admission, abdominal ultrasonography had shown bilateral small kidneys and echocardiography had revealed mild aortic and pulmonary stenoses. His karyotype was 46,XY.

On the 1st day of admission to our center, he was lethargic and febrile with a history of jerking movements of hands for 1 week. On physical examination, he had a blood pressure of 90/40 mm Hg, respiratory rate of 53/min, and axillary temperature of 37.8°C. He was ill and dyspnoeic. On chest examination, he had a III/VI systolic murmur and bilaterally diffused fine crackles. Pretibial pitting edema was notable. Due to elevated BUN and serum creatinine accompanied by uremic symptoms (loss of consciousness and clonus) urgent hemodialysis was started but because of persistent fever, positive blood cultures for microorganisms (7 days after hemodialysis), and lack of appropriate vascular access, CAPD was started on with a Tenckhoff catheter. He was followed up for 9 months without any complication with his catheter, and his glomerular filtration rate was stabilized (Table).

DISCUSSION

Most cases of BBS are diagnosed after the first decade of life and diagnosis in early infancy is very rare.[4] An interesting point in our patient was presentation with ESRD in earlier ages than most of reported cases,[5,7,8] and also the death of 2 siblings with BBS phenotype (morbid obesity) in their neonatal period, which is very rare. Nephrogenic diabetes insipidus is often the first clinical sign of BBS, as in our patient.[5,9] Renal involvement is common in BBS, but only 5% to 25% of the patients develop kidney dysfunction and 4% to 10% progress to ESRD after the second decade of life, which is the most common cause of death in these patients.[7]

Between July 1985 and January 2005, 5 patients with BBS were transplanted preemptively at Shaheed Labbafinejad Medical Center in Tehran.[8] Using CAPD is limited to situations in which hemodialysis has failed.[7] We suggest CAPD as an alternative modality in these patients because of morbid obesity and technical problems in the first step. Finally, close follow-up for the renal involvement from the early ages is highly recommended,[7] to prevent ESRD and immediately start renal replacement therapy.



Syndactyly, brachydactyly, and postaxial polydactyly in a 9-year-old boy with Bardet-Biedl syndrome.

Kidney Function Indicators During Peritoneal Dialysis in A Boy With Bardet-Biedl Syndrome

Test	Baseline	Follow-up		
		1 Week	1 Month	9 Months
Blood urea nitrogen, mg/dL	61	43	14	20
Serum Creatinine, mg/dL	7.9	5.6	2.5	2
Glomerular Filtration Rate, mL/min/1.73 m^2	11	16	36	45

CONFLICT OF INTEREST

None declared.

REFERENCES

1. Tobin JL, Beales PL. Bardet-Biedl syndrome: beyond the cilium. Pediatr Nephrol. 2007;22:926-36.
2. Chan WKY, Ho S, But B, Tse WWY. Renal disease in Bardet-Biedl Syndrome. HK J Paediatr (New Series). 2000;5:34-9.
3. Green JS, Parfrey PS, Harnett JD, et al. The cardinal manifestations of Bardet-Biedl syndrome, a form of Laurence-Moon-Biedl syndrome. N Engl J Med. 1989;321:1002-9.
4. Beales PL, Elcioglu N, Woolf AS, Parker D, Flinter FA. New criteria for improved diagnosis of Bardet-Biedl syndrome: results of a population survey. J Med Genet. 1999;36:437-46.
5. O'Dea D, Parfrey PS, Harnett JD, Hefferton D, Cramer BC, Green J. The importance of renal impairment in the natural history of Bardet-Biedl syndrome. Am J Kidney Dis. 1996;27:776-83.
6. Dippell J, Varlam DE. Early sonographic aspects of kidney morphology in Bardet-Biedl syndrome. Pediatr Nephrol. 1998;12:559-63.
7. Rathi M, Ganguli A, Singh SK, et al. Bardet-biedl syndrome with end-stage kidney disease: a case report and review of literature. Indian J Nephrol. 2007;17:10-3.
8. Sharifian M, Dadkhah-Chimeh M, Einollahi B, et al. Renal transplantation in patients with Bardet-Biedl syndrome. Arch Iran Med. 2007;10:339-42.
9. Devarajan P. Obesity and genitourinary anomalies in Bardet-Biedl syndrome after renal transplantation. Pediatr Nephrol. 1995;9:397-8.

Correspondence to:
Ali Amanati, MD
Besat Hospital, Hamedan, Iran
Tel: +98 912 444 5685
E-mail: ali_amanati_1356@yahoo.com

Received April 2008
Revised July 2008
Accepted July 2008