Renal Failure due to Bardet-Biedl Syndrome
A Case Report

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Abstract

Objective: To describe a case of Bardet-Biedl syndrome involving renal failure and retinal dystrophy. Case Presentation and Intervention: A 50-year-old female patient presented to the emergency service with uremic symptoms and metabolic acidosis. Polydactyly, retinitis pigmentosa, obesity, strabismus, nistagmus and renal failure were found. Because she had end-stage renal failure, hemodialysis therapy was started. She has been well for 18 months, without any complication on hemodialysis. Conclusion: Bardet-Biedl syndrome should be considered in patients with polydactyly, retinitis pigmentosa and renal failure.

Introduction

Bardet-Biedl syndrome, a form of Laurence-Moon-Biedl syndrome, has five recognized features: retinal dystrophy, obesity, dysmorphic extremities, renal abnormalities and hypogonadism (in male patients). Polydactyly, syndactyly or brachydactyly may be seen. The degree of polydactyly, a prominent feature of this syndrome, varies from patient to patient, ranging from a wide fifth metatarsal or metacarpal to a complete sixth digit [1]. Scores on tests of intelligence are usually low. Mental retardation, polydactyly and hypogonadism are not necessarily present in female patients [2]. The autosomal recessive disorder Bardet-Biedl syndrome is heterogeneous with at least four gene loci (BBS1–4): 11q13 (BBS1), 16q21 (BBS2), 3p12 (BBS3) and 15q22 (BBS4) [3]. A fifth locus [4] and a sixth locus [5] map to chromosome 2q31 and chromosome 20, respectively. These alleles may act in conjunction with mutations at other BBS loci to cause or modify the BBS phenotype. The prevalence is 1:160,000. It usually presents between 10 and 20 years of age. The ocular defect in Laurence-Moon-Biedl syndrome involves the photoreceptor cells [6]. In addition to retinitis pigmentosa, strabismus may be observed [7]. Early otolaryngologic, audiologic, speech pathology, and dental evaluation of these individuals is recommended [8]. Bifid epiglottis [8] and disturbance of tooth formation have been reported. Hypertrophy of the interventricular septum and dilated cardiomyopathy, insulin-resistant diabetes mellitus, empty sella, clinodactyly, and congenital hepatic fibrosis can also be found. Although hypogonadism is rare in female patients, vaginal atresia, hypoplasia of uterus

Key Words
Bardet-Biedl syndrome - Renal failure - Hemodialysis

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and ovaries, and ectopic urethra should be sought. Uremia is an important cause of morbidity and early mortality in these patients. Structural or functional abnormalities of the kidneys can be observed. The spectrum of renal involvement can range from calyceal clubbing or cysts to diffuse renal cortical loss [9]. Light microscopy may reveal varying degrees of increase in mesangialcellularity and matrix or sclerosis of the glomerular tuft [10]. Renal disease occurs in 70% of the patients [10].

We describe a patient with renal failure, who had signs and symptoms of Bardet-Biedl syndrome.

Case Report

A 50-year-old single, nulliparous woman with normal secondary sex characteristics presented to our emergency room in September 2000 with a history of nausea, vomiting and dyspnea. She had a history of frequent urinary system infections. On examination, she was pyrexic at 38.4°C, and had an erythematous lesion with yellowish discharge located near the vulva. Kussmaul’s respiration was observed. She had been blind since aged 40. Ophthalmoscopic examination revealed bilateral retinal pigmentation and strabismus. There were bilateral scars due to amputation of a sixth toe on both feet. The patient had been postmenopausal for 2 years. Her thyroid and sex hormone dysregulation revealed bilateral retinal pigmentation and strabismus. There were bilateral scars due to amputation of a sixth toe on both feet. The patient had been postmenopausal for 2 years. Her thyroid and sex hormone dysregulation. Hematology revealed Hb 9.9 g/dl, WBC 15 x 10^9/l, platelets 124 x 10^9/l, blood smear showed polymorphonuclear leukocytosis. Serum creatinine was 10.4 mg/dl, blood urea nitrogen was 111 mg/dl, calcium was 5.8 mg/dl. Blood gases revealed metabolic acidosis. Methicillin-resistant Staphylococcus aureus was isolated from the culture of the lesion. Vancomycin therapy was initiated. Renal ultrasound revealed a left kidney 77 mm in length and 8 mm in parenchymal width, with increased echogenicity of parenchyma; the right kidney was 85 mm in length and 10 mm in parenchymal width. Her body mass index was 36 kg/m², and she was 150 cm in height. She was normotensive and had left ventricular concentric hypertrophy. There was no history of blindness, consanguinity and renal failure in her family. Her mental status assessed quantitatively was normal. She was no history of blindness, consanguinity and renal failure in her family. Her mental status assessed quantitatively was normal. 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