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Case report

Bardet Biedel Syndrome: a Rare Cause of Chronic Kidney Disease

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Abstract

Bardet Biedl syndrome (BBS) is characterized by obesity, retinitis pigmentosa, hypogonadism, mental retardation and polydactyly. Additionally, renal, cardiac and neurological manifestations may be seen. We report a case of BBS with chronic kidney disease (CKD) at the age of 43.

Keywords: Bardet Biedl syndrome, chronic renal disease, retinitis pigmentosa

Introduction

Bardet Biedl syndrome (BBS) is an autosomal recessive condition characterized by obesity, retinitis pigmentosa, hypogonadism, mental retardation and polydactyly. It has prevalence of 1 in 1,40,000-1 in 1,60,000 worldwide [1]. Renal involvement in the form of various structural and functional abnormalities is common and renal insufficiency is noted in 5-25%, with progression to end-stage renal disease (ESRD) in 4-10% [2-4].
In our country a small number of cases with this syndrome has been reported. We report a case of BBS with chronic kidney disease (CKD) at the age of 43.

Case report

A 43-year-old female patient with swelling in the legs presented with nausea and a decrease in her oral intake for one-month-period. In the patient's medical history and the family tree blindness has occured at the age of 7 and she underwent a polydactyly surgery. From mother and father relatives, there is one brother with a loss of vision in the patient's family history (Figure 1).
On physical examination, she was with central obesity, her vital signs were: heart rate 80 per minute, respiratory rate 23/dk, blood pressure 150/90 mm/hg; weight 82 kg; height 148 cm; body mass index, 37.43.kg/m² and body temperature 36.5°C.
Positive findings of physical examination, bilateral +/+ pretibial edema, abdominal obesity, internal strabismus, common hyperpigmentation, left upper and lower extremity reconstruction polydactyly from the surgery with scar lesions are presented in Figure 2. The eye examination revealed severe retinitis pigmentosa.
Laboratory findings were: urea 135 mg/dL, creatinine 5.8 g/dL, Modification of Diet in Renal Disease (MDRD) Study glomerular filtration rate (GFR) value based on 4 variables (age, race, gender, plasma creatinine) was 8 ml/min/1.73 m2; calcium 8.6 mg/dL, phosphorus 5.2 mg/dL, sodium 140 mmol/L, potassium 4.7 mEq/L, chloride 115 mEq/L, ALT: 24 U/L, AST: 26 U/L, GGT 32 U/L, alkaline phosphatase 112 U/L, lactate dehydrogenase 421 U/L, total protein 7.8 g/dl, albumin 4.5 g/dl, triglycerides 226 mg/dl, total cholesterol 217mg/dL, serum iron levels of 58 mg/dL, serum iron binding capacity of 338 mg/dL, saturation index 17.2%, ferritin 47 ng/mL, vitamin B12

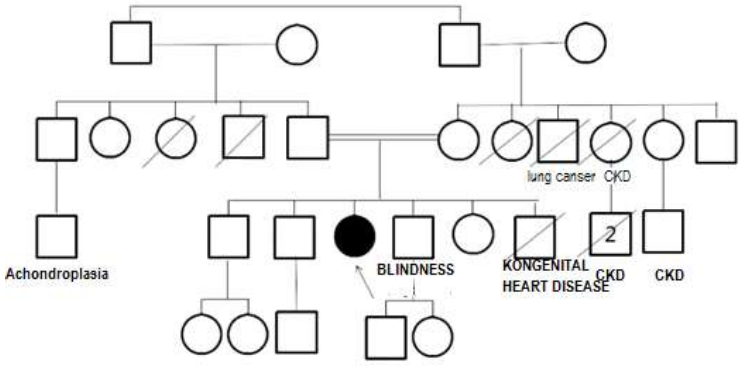

Fig. 1. The family three of the patient

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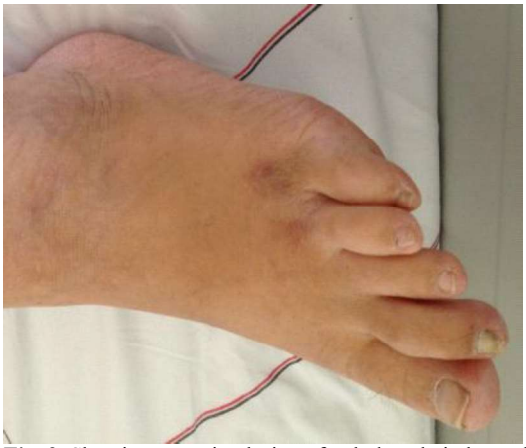

Fig. 2. Showing operation lesion of polydactyly in lower limb

was 275 pg/ml, and folate 5.9 ng/ml. Blood gas analysis were: pH 7.25, CO^2: 41 mm Hg, HCO_3 16.6 mmol/L. Evaluation of urine and the sediment: pH: 7,0 specific gravity: 1005, protein +, glucose +, and microscopic examination of the urine showed rare leukocytes.

The patient's initial diagnosis was Bardet Biedl. We sent a blood sample for the study of mutations in a genetic analysis center. Renal ultrasonography detected reduced kidney size and renal echogenicity increased bilaterally. The patient was considered as progressing towards ESRD, and renal replacement therapy was initiated.

Discussion

The diagnostic criteria for BBS as major features include retinal dystrophy (90%), post axialpolydactyly (21%), truncal obesity (72%), hypogonadism (more frequent in males), renal anomalies, hypertension (50-66%) and chronic renal failure (30-60%). Minor features include learning disabilities, speech delay, developmental delay, behavioral abnormalities, eye abnormalities, brachydactyly/syndactyly, ataxia, mild hypertonia, diabetes mellitus, orodental abnormalities, cardiovascular anomaly, and anosmia [1-4]. Four major or three major and two minor criteria are required for the diagnosis. BBS worldwide changes may be frequently found. Prevalence rates in North America and Europe is with 1:140000 - 1:160000 of live births.

ESRD in BBS patients has been reported at age range of 4-57 years [5-9]. We report a case of BBS with CKD in a 43-year-old female from Turkey. This diagnosis should be considered in patients with renal disease and the characteristic phenotype of retinitis pigmentosa, postaxial polydactyly and central obesity. Renal involvement is common and renal failure is most common cause of death in BBS. These patients should undergo regular monitoring of renal function test for an early diagnosis and treatment of CKD to prevent the progression and respective morbidity and mortality. Renal transplantation is a possible option of RRT in these patients. These findings are valuable for comparing phenotype of BBS patients with CKD from various national and international centers. Since our patient was diagnosed at a late stage once on hemodialysis, the patient and the relatives were informed about renal trasplantation, as well.

In conclusion, a patient presenting with uremia, polydactyly, obesity, mental retardation and if accompanied with retinitis pigmentosa, Bardet Biedl syndrome should be considered as most probable diagnosis.

Conflict of interest statement. None declared.

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