IJMS
Vol 41, No 6, November 2016

Case Report

Bardet-Biedl Syndrome with End Stage Renal Disease

Rajendrakumar Parakh, MD;
Dhananjaya Matapadi Nairy, MD

Department and Institutions, SDM College of Medical Sciences and Hospital, Sattur, Dharwad-580009, State-Karnataka, India

Correspondence:
Rajendrakumar Parakh, MD;
Department of Medicine,
SDM College of Medical Sciences and Hospital, Manjushree Nagar, Sattur,
Postal Code: 15657-74419,
Dharwad-09. Karnataka (state), India.
Email: drrajendra80@gmail.com
Received: 24 December 2014
Revised: 03 May 2015
Accepted: 17 May 2015

Abstract

Bardet-Biedl syndrome (BBS) is one of the rare autosomal recessive disorders that affect multiple organs of the body. The signs and symptoms of this condition vary among affected individuals, even among members of the same family. We present a case of BBS with features of hypogonadism and features such as marked central obesity, retinitis pigmentosa, polydactyly, renal abnormalities and mental retardation, along with a brief review of the literature. The patient had end stage renal disease and managed with dialysis. This case also exemplifies the need for multidisciplinary approach in the management of such cases.

Please cite this article as: Parakh R, Nairy DM. Bardet-Biedl Syndrome with End Stage Renal Disease. Iran J Med Sci. 2016;41(6):539-542.

Keywords ● Bardet–Biedl syndrome ● Hypogonadism ● Retinitis pigmentosa ● Chronic kidney failure ● Dialysis

What's Known

- Bardet–Biedl syndrome (BBS) is a rare autosomal recessive disorder affecting multiple body organs.
- Main clinical manifestations are rod-cone dystrophy (retinitis pigmentosa), postaxial polydactyly, central obesity, mental retardation, hypogonadism, and renal dysfunction.

What's New

- Fewer than 15 cases of BBS have been reported from India.
- A few cases of BBS have been reported with end-stage renal disease.
- BBS should be suspected when the patient presents with renal failure and characteristic features of postaxial polydactyly, central obesity, and retinitis pigmentosa.

Introduction

Bardet-Biedl syndrome (BBS) is a rare autosomal recessive disorder, first described by Bardet and Biedl in the 1920.[1] It is a disorder that affects many parts of the body. This rare syndrome manifests differently among affected persons and within the members of the same family. One of the major features is vision loss. Loss of vision occurs as the retina gradually deteriorates. Initially, there is problem with night vision, which becomes obvious by middle of childhood followed later by the appearance of blind spots in peripheral vision. Later, the patient on examination presents as tubular vision. Other manifestations, which can be seen in these individuals, are blurred central vision with poor visual acuity and field of vision.

The main clinical manifestations are rod-cone dystrophy (retinitis pigmentosa), postaxial polydactyly, central obesity, mental retardation, hypogonadism, and renal dysfunction. The rare manifestations that have been described in various studies are diabetes mellitus, hepatic dysfunction and fibrosis, speech and language disturbances, behavioral abnormality, facial dysmorphism, etc.[2,3] We present here a case of BBS which is rarely seen.

The frequency of the syndrome is estimated to be 1:160,000.[4] Less than 15 cases have been reported from India.[5] The incidence varies among world populations with high incidence of 1 in 13,000 in the isolated populations of Newfoundland and Kuwait, 1 in 17,000 live births because of high level of consanguinity.[6]

Chronic renal dysfunction is a major cause of morbidity and mortality. Conventional approaches in such patients are chronic peritoneal dialysis and hemodialysis followed by kidney transplantation.

We report this case from a tertiary care hospital of North Karnataka, which is a classic case of BBS fitting into the

definite criteria to diagnose this rare syndrome presenting with end stage renal disease.

Case Report

A 22-year male was admitted with complaints of loss of appetite, vomiting, lower abdominal pain, burning micturition and fever of 15 days duration. The patient had learning difficulty since birth, born of consanguineous marriage, a family history of obesity, learning difficulties, six digits, and visual impairment in his two younger brothers. He had six digits on all three limbs, diagnosed at birth (figure 1). His height was 126 cm, weight 52 kg with BMI of 32.9 kg/m^2. His IQ was 48 for his chronological age. He had central obesity, mild mental retardation, digital abnormalities included postaxial polydactyly which was complete in the left foot, right foot and left hand (figures 1 and 2), brachydactyly of both hands and feet. He had sparsely distributed facial and body hair and underdeveloped external genitalia. Examinations of the other systems were unremarkable.

All blood investigations were normal except for anemia (hemoglobin 4.5 g/dl), total counts 32,140 cells/cumm, and urine routine showed plenty of pus cells and deranged renal parameters with reduced daily urine output (creatinine 15.5 mg/dl). Ultrasonogram showed a corticomedullary cyst in the left kidney with grade 2-3 renal parenchymal changes. His right kidney size was 65×45 mm, left measured 62×41 mm. Fundus examination showed pallor of the optic disc and retinal pigmentary changes of retinitis pigmentosa (figure 3). Electrocardiogram and echocardiogram were normal. CT abdomen showed both Kidneys were small in size and heterogeneous enhancement in venous phase. Multiple small calcifications and small non-enhancing hypodense areas were seen in the cortex of both kidneys. On delayed images, there was no excretion of contrast into pelvicalyceal system and ureters on both sides. Renal arteries were diffusely narrowed on both sides (figure 4).

We could not perform genetic analysis due to insufficient setup in this part of the country. Moreover, the patient could not afford the cost. This is one of the limitations of the study.

The patient was treated for his urinary tract infection and hemodialysis was done for his renal failure. The patient was discharged in stable condition with the advice of regular hemodialysis. Written informed consent was obtained from the patient for the publication of this case report and accompanying images.

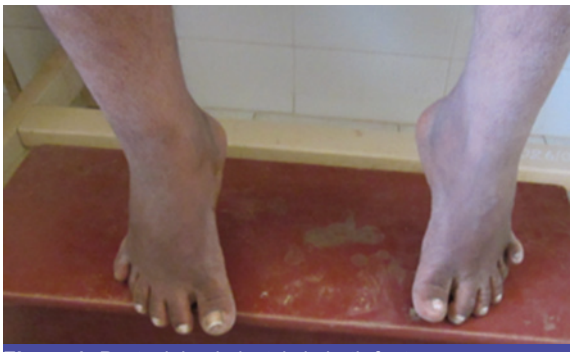

Figure 1: Postaxial polydactyly in both feet.

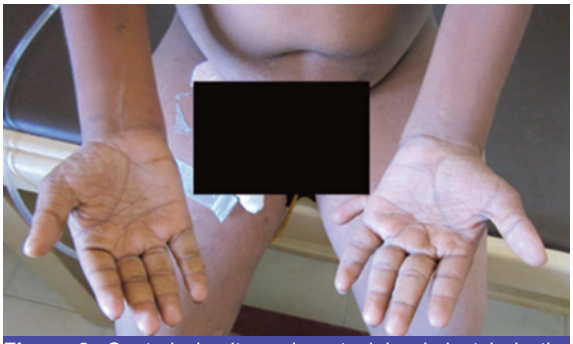

Figure 2: Central obesity and postaxial polydactyly in the left hand.

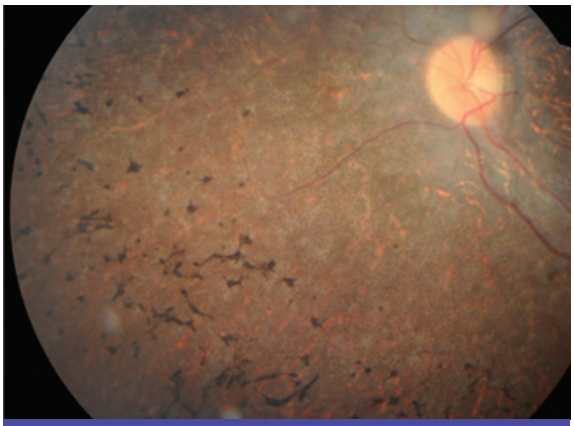

Figure 3: Retinitis pigmentosa.

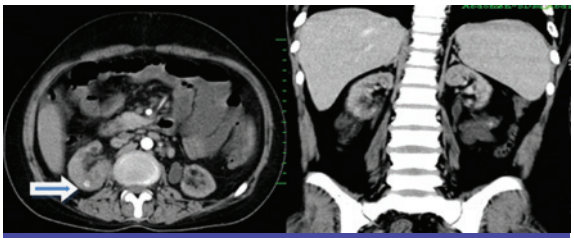

Figure 4: Small size of the kidney with multiple small calcifications.

Discussion

The syndrome was first described by Bardet-Biedl in the year 1920 and later by mistake compared to another disorder described by Laurence and Moon. It was consequently

referred to as Laurence-Moon-Biedl syndrome. Laurence-Moon-Biedl syndrome has some similar features of BBS like obesity, retinal pigmentary degeneration, mental retardation and hypogonadism occurs in association with progressive spastic paraparesis and distal muscle weakness except for polydactyly.[7]

Obesity is one of the common features found in these patients and they are more prone to hypertension and hypercholesterolemia. Ocular manifestations are the most common manifestation and become apparent between the ages of 4 and 10 years. In males, hypogonadism is commonly seen and may have primary testicular failure. The earliest and most common presenting feature is postaxial polydactyly. Renal failure is the major cause of morbidity and early mortality in BBS. The commonly seen renal anomalies are chronic renal failure, renal parenchymal cysts, clubbing of calyceal system, scarring, agenesis usually unilateral, renal calculi, and vesicoureterix reflux.

Apart from these chief presentations, a patient may also have intellectual impairment, congenital heart block, brachycephaly, sensorineural deafness, and dental abnormalities. The full spectrum of clinical features is found in only 40–45% of cases.[8]

The genetics is still unclear. Twelve genes (BBS1 to BBS12) that are responsible for the disease have been cloned. BBS is a disease under the spectrum of 'ciliopathies' because molecular basis for this disease is that it affects the centrosome and affect the ciliary transport.[5]

One of the stressful periods among these patients is during puberty and hence it is beneficial to these patients to receive guidance from an experienced counselor. Testosterone supplements may be prescribed to male patients, specifically in cases having lowered level of this hormone. Until the present, there is no official effective treatment to halt or prevent the deteriorating vision.

We stressed upon regular ophthalmological follow-up. Accessory digits are often nonfunctional and may be excised. Obesity is a primary concern, which is a part of metabolic syndrome and if not addressed it can lead to major health issues and increased health care cost to the patient. Hence, a dietary advice of low carbohydrate, low fat, and regular exercise may slow the progression of disease.[7]

Chronic kidney disease is treated with dialysis and the option of renal transplantation to be given. Early and regular screening for hypertension, diabetes, and renal involvement is required. The management of renal failure does not differ from that due to any other cause. All three modalities of renal replacement therapy, i.e. hemodialysis, chronic peritoneal dialysis, and renal transplantation can be offered to these patients.[3] In our patient, small shrunken kidney was found with altered renal parameters and hemodialysis was advised twice a week. Chronic kidney disease is an important cause of morbidity and mortality.

To conclude, so far, less than 15 cases have been reported from India, but only a few have reported BBS with end stage renal disease. This is a rare case and hence reported to create awareness among the treating clinician to consider this diagnosis when a patient presents with renal failure and characteristic features of postaxial polydactyly, central obesity and retinitis pigmentosa. The involvement of the kidney is common and renal failure is the most common cause of morbidity and mortality in BBS.

Conflict of Interest: None declared.

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