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Bardet – Biedl Syndrome: A Rare Cause of End Stage Kidney Disease - A Case Report from Central India

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ABSTRACT

Introduction

Bardet – Biedl syndrome is a rare autosomal recessive disorder, characterized by dysfunction of basal body of ciliated cells. It includes mutations in 22 genes coding for different proteins located in the basal body and primary cilia of the cell making the syndrome an archetypal ciliopathy.

It results in myriad manifestations in various organ systems. The predominant features of this syndrome are retinitis pigmentosa, polydactyly, obesity, learning disabilities, hypogonadism, and renal abnormalities. Though structural renal abnormalities are common in these patients, chronic kidney disease as a feature of this syndrome has been reported rarely

Keywords

Bardet–Biedl syndrome, manifestations, predominant, abnormalities

CASE REPORT

A 33-year-old female had presented to our hospital with complaints swelling of legs and oliguria of 1 month duration. She was born of non-consanguineous marriage. She was born at full term by normal vaginal delivery. Patient had delayed developmental (physical and mental) milestones. Patient had difficulty in night vision since 7 years of age and difficulty indistant and

colour vision since 10 years of age. She dropped out from school due to poor scholastic performance. Patient had not attained menarche. She has one elder brother with no similar manifestations. The family history was unremarkable.

On examination, her height and weight were 130 cm (<10th centile) and 70 kg respectively with a body mass index of 41.4 kg/m^2.

Detailed clinical examination revealed pallor with bilateral pitting pedal edema. Physical examination was notable for the absence of secondary sexual characters in the form of absence of axillary, pubic hair, and poor breast bud development. She hadpolydactyly in both her legs and right hand [Figures [Figures1]. Her visual acuity was decreased to counting fingers at 1 mts in both the eyes. Ophthalmic examination revealed features of retinitis pigmentosa [Figure 2 & 3]. Her psychological evaluation showed an IQ of 60.

Figure 1

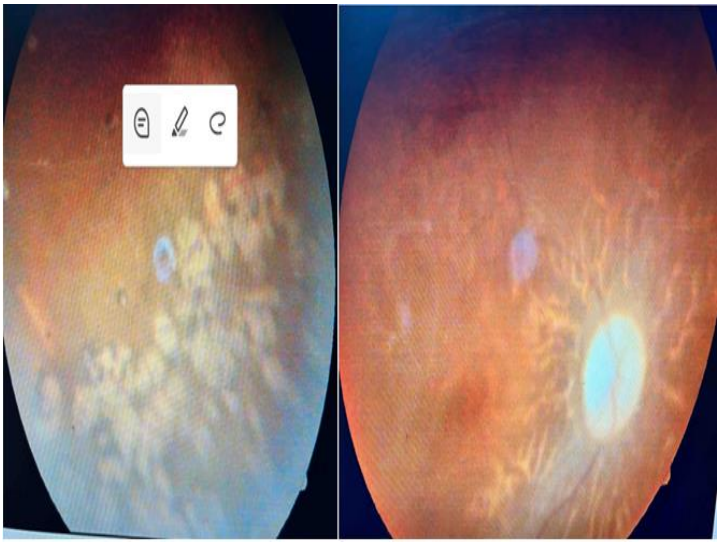

Figure 2, Figure 3

Investigations revealed hemoglobin - 7.7 gm/dl, TLC-5990 /cumm, platelet count 1.77 lakhs, urea - 287 mg/dl, creatinine –24.20 mg/dl, sodium- 140meq/L, potassium- 5.8 meq/L, calcium –5.8 mg/dl, inorganic phosphorus –4.7 mg/dl, uric acid- 16.2 mg/dl and serum albumin –3.42 gm/l. Urine analysis revealed trace proteinuria with no active sediments. Her TSH was 8.78 uIU/ml.Serum alkaline phosphatase was233 U/l and intact PTH of 418.3 pg/ml. Her fasting and post prandial blood glucose were 96 and 126 mg/dl respectively. Ultrasound examination showed reducedbilateral kidney size with raised echogenicity and poor corticomedullary differentiation in both kidneys. 2D Echocardiography showed concentric Left ventricular hypertrophy with grade I left ventricular diastolic dysfunction with RVSP 36 mmHg + RAP.

The combination of pigmentary retinopathy, polydactyly, mild mental retardation, obesity, and renal failure fits well with the diagnosis of BBS. Patient was initiated on hemodialysis via right internal jugular HD catheterization followed by A-V Fistula creation. At present, she is on maintenance hemodialysis.

DISCUSSION

Bardet–Biedl syndrome is named after Georges Bardet and Arthur Biedl. The first known case was reported by Laurence and Moon. Laurence-Moon-Biedl-Bardet syndrome (LMBBS) is no longer considered as a valid term as patients of Laurence and Moon had paraplegia but no polydactyly and obesity, which are the key elements of the BBS.

Bardet–Biedl syndrome is genetically heterogenous. Mutations in 16 genes (BBS1 to BBS12, MKS1, NPHP6/CEP290, SDCCAG8, and SEPT7) have been identified to cause the BBS phenotype.[5]BBS has been recently included in the broad category of “ciliopathies”, a class of genetic diseases that occur due to primary ciliary dysfunction. Increasing evidence suggests that primary cilia are key coordinators of signalling pathways during development and in tissue homeostasis.[6]

The primary features of BBS are early-onset retinal dystrophy, obesity, limb defects, mental retardation, hypogonadism, and renal abnormalities. It occurs

throughout the world with prevalence rates of 1:140000–1:160000.[7]

Rod-cone dystrophy is the most common (93%) feature.[8] The onset is often earlier and progression more rapid than in isolated typical retinitis pigmentosa. Almost all patients present with visual disturbance by 2nd decade. Macular involvement has been observed in this condition resulting in reduced visual acuity. Truncal obesity is reported to occur in 72% of cases.[9] It usually starts in childhood and severity increases with age.

Polydactyly (63-81%) is the most common limb defect, though brachydactyly (6-100%), partial syndactyly, fifth finger clinodactyly, and a prominent gap between the first and second toes were also noted.[10] Mental retardation occurs with varying frequency and severity. Hypogonadism is more common in males while genital abnormalities are more common in females.[11]

The renal abnormalities as a part of this syndrome have been recognized only recently. In a seminal study by Beales *et al*.,[2] 26 (46%) out of 57 patients had renal structural abnormalities which included renal parenchymal cysts, communicating calyceal cysts, calyceal clubbing and blunting, fetal lobulation and scarring, dysplastic kidneys, unilateral agenesis, vesicoureteric reflux, bladder obstruction, horseshoe kidney, and ectopic kidney. Though only 5% had chronic renal failure, renal failure is the most common cause of mortality in these patients. All the three modalities of renal replacement therapy namely hemodialysis, peritoneal dialysis, and transplantation can be optimally used in these individuals.[12]

The other secondary features are hearing loss, speech disturbances, pigmented naevi, hypertension, diabetes mellitus, congenital heart disease, cardiomyopathy, hepatic fibrosis, nephrogenic diabetes insipidus, hypothyroidism, Hirschsprung's disease and abnormal dentition.[13]

As the diagnostic criteria of BBS are mainly clinical, so patients may remain in quiescent stage for many years until it has become flourished. Diagnostic dilemma has been found in some cases.

Multidisciplinary approach is needed for management of BBS. Both proper diet and exercise programs should be organized and scheduled to address obesity. As Eye problems are of central concern so patients with BBS should be encouraged for periodic vision evaluation by ophthalmologic examinations. Regular routine follows up should be arranged. Some of the physical abnormalities can be corrected with surgeries like, polydactyly for cosmetic purposes, and some genitourinary developmental anomalies and congenital cardiac conditions. As it is a genetic condition so Genetic Counselling may play a pivotal role in the management spectrum of the condition. Both the affected individual and his/her families should be encouraged for proper counselling by health professionals.[14]

CONCLUSION

BBS is a disease of genetic complexity. To the best of our knowledge, only 13 cases of BBS have been reported from India with End stage kidney disease (ESKD) being an infrequent manifestation.BBS is a rare cause of ESKD. Elucidation of the genetics and pathogenesis of this disease may yield novel therapeutic options in the future.

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