Bardet Biedl syndrome - a rare case report from North Karnataka

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Abstract

Bardet Biedl syndrome (BBS) is a rare autosomal recessive condition characterised by rod-cone dystrophy, postaxial polydactyly, central obesity, mental retardation, learning difficulties, hypogonadism in males and renal involvement. Renal failure is the most common cause for morbidity and mortality in such individuals. We report the case of BBS with chronic renal failure from north Karnataka.

Key words: Bardet Biedl syndrome, autosomal recessive, renal failure

Introduction

Bardet Biedl syndrome (BBS) is an autosomal recessive condition characterised by rod-cone dystrophy, postaxial polydactyly, central obesity, mental retardation, hypogonadism, and renal dysfunction. BBS expression varies both within and between families and diagnosis is often difficult [1]. The frequency of the syndrome is estimated to be 1 in 1,60,000 [2]. Only few cases of BBS have been reported from India. We present a classical case of BBS with chronic renal failure presenting to medicine department.

Case report

A 19 year male school dropout was admitted for lower abdominal pain, burning micturition and fever of 10 days duration. An initial evaluation revealed urinary tract infection with renal failure. Our patient is the first offspring of consanguineous parents. His family history was notable for obesity, learning difficulties, six digits on two limbs and visual impairment in his 16 year old brother. He had also six digits on all four limbs, diagnosed at birth (Figure 1). Initial motor and mental development milestones were abnormal, a delay noted by the family doctor, but the diagnosis was not established in the context of family history and consanguinity of the parents at birth.

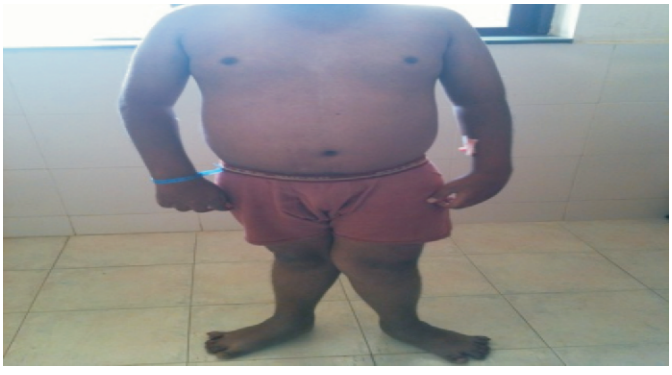

Figure 1. Central obesity

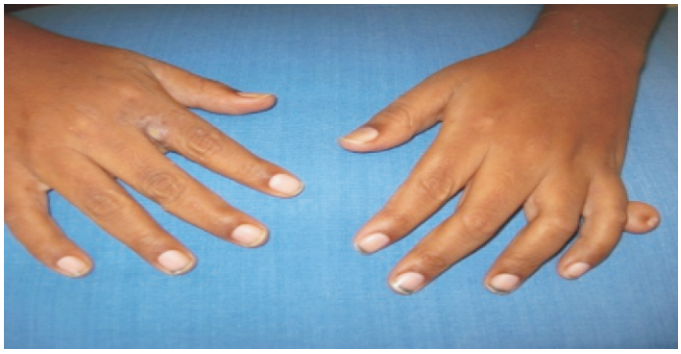

Figure2. Postaxial polydactyly in the left hand

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He had mild central obesity (Figure 1), learning difficulties, mild mental retardation and abnormal dentition were observed. Digital abnormalities included postaxial polydactyly which was complete in the left foot and hand and right foot and incomplete in the right hand (Figure 2 and 3), partial syndactyly (between the forth and the fifth toes- Figure 3), fifth finger clinodactyly of both hands (figure 2), brachydactyly of both hands and feet (Figure 2 and 3). He had sparsely distributed facial and body hair. He had underdeveloped external genitalia – a smaller than normal testes and a microphallus with hypospadias. Examinations of the cardiovascular, respiratory system and nervous system were unremarkable. All blood investigations were normal except for anemia (hemoglobin-8.8g/dl) and deranged renal parameters with adequate daily urine output (creatinine-3.9mg/dl). Ultrasonogram showed a corticomedullary cyst in the left kidney with grade 1 renal parenchymal changes. Fundus examination showed waxy pallor of the optic disc, bilaterally attenuated vessels and retinal pigmentary changes of retinitis pigmentosa (Figure 4). Electrocardiogram and echocardiogram were done to rule underlying cardiac abnormalities and were normal.

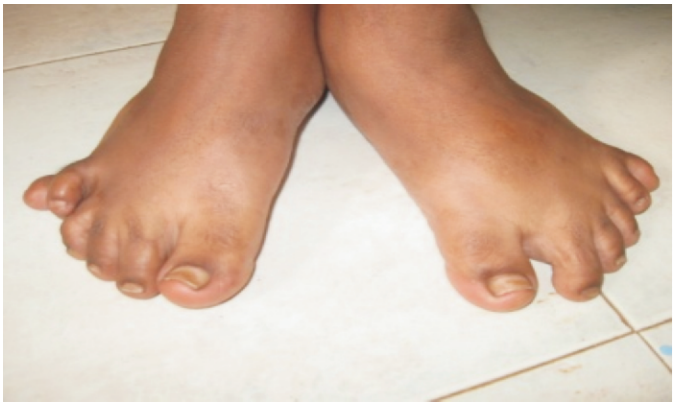

Figure3. Postaxial polydactyly in the both foot

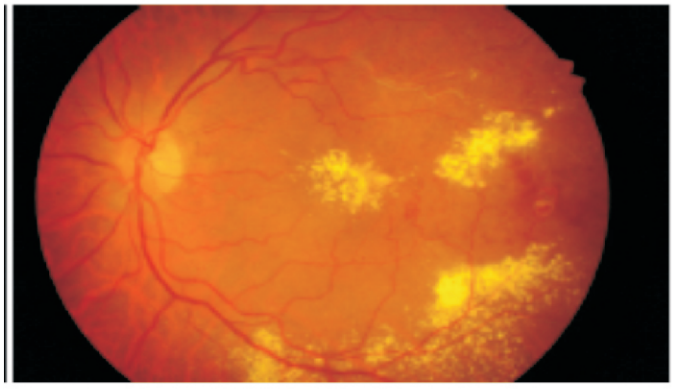

Figure4. Retinitis pigmentosa

Discussion

Bardet-Biedl syndrome (BBS) is an autosomal recessive condition with a wide spectrum of clinical features [1]. BBS is named after Georges Bardet and Arthur Biedl. The first known case was reported by Laurence and Moon in 1866. Laurence-Moon-Bardet-Biedl syndrome (LMBBS) is no longer considered as a valid term as a patient of Laurence-Moon syndrome had paraplegia but no polydactyly and obesity, which are the key elements of the BBS. Hence, Laurence-Moon syndrome is considered as a separate entity [2]. The BBS phenotype is seen in individuals with mutations in 11 different genes [3]. The exact pathogenesis of BBS is unknown. It has been recently recognised that proteins coded for by the BBS4, BBS6, BBS8, and BBS10 genes are expressed in the basal body of cilia, and BBS is now regarded as one of the 'ciliopathies'. The gene products are probably involved in the signalling pathway in the cilia; abnormalities interfere with the normal development, resulting in the diverse pathological effects of the syndrome [4].

The principal manifestations of BBS are rod cone dystrophy, postaxial polydactyly, central obesity, mental retardation, hypogonadism, and renal dysfunction [5]. Other features include hepatic fibrosis, diabetes mellitus, reproductive abnormalities, endocrinological disturbances, short stature, developmental delay, and speech deficits. Beales et al [1] suggested the diagnostic criteria for BBS which is summarised in table 1.

Table 1 Diagnostic criteria in Bardet-Biedl syndrome (BBS) (Features in our patient are shown in bold letters)

Primary features of BBS	Secondary features of BBS
Retinal dystrophy	**Developmental delay**
Post-axial polydactyly	Behavioral problems
Obesity	Neurological problems
Hypogenitalism	Speech disorder
Renal abnormalities	**Brachydactyly, syndactyly, or Clinodactyly Dental anomalies**
Learning disabilities	Nephrogenic diabetes insipidus
	Diabetes mellitus
	Hypertension
	Anosmia

Management of BBS is supportive. It includes training and rehabilitation of blind and mental retarded patients, hearing aids for deafness. There are no proven effective treatments to either prevent or alleviate the deterioration in vision. Accessory digits are non-functional and may be excised. Testosterone supplements may be advised to male patients especially in cases with low levels of this hormone. Diet control and exercises are advised for managing obesity. A low calorie and low protein diet help in obesity control and may slow the progression of renal failure in patients with BBS [6]. 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, haemodialysis, chronic peritoneal dialysis and renal transplantation can be offered to these patients [7]. As structural renal changes and derangement in renal parameters were identified in our patient, he was advised half yearly urinalysis, blood pressure measurement and urea and creatinine levels as chronic kidney disease is an important cause of morbidity in such patients.

To conclude, so far less than 15 cases have been reported from India [8], but only few have reported BBS with chronic kidney disease. Our patient is first to be reported from this part of north Karnataka. This case is reported for its rarity. This diagnosis to be considered in patients with renal failure and characteristic features of postaxial polydacytyl, central obesity and retinitis pigmentosa. The involvement of kidney is common and renal failure is the most common cause for morbidity and mortality in BBS.

References

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