Case Report

Bardet-Biedl Syndrome: A Case Report

Wahiduzzaman M[a], Rahim MA[b], Iftekhar M[c]

Abstract

Bardet-Beidl syndrome is a rare autosomal recessive disorder having heterogeneous clinical manifestations including rod-cone dystrophy, polydactyly, obesity, learning difficulties, development delay, speech deficit, diabetes mellitus, congenital heart disease etc. Most of these symptoms are not present at birth but appear and progressively worsen during the first and second decades of life. The combined occurrence of diabetes mellitus and retinitis pigmentosa is rare. Here, we present such a case.

Key words: *Bardet-Beidl syndrome, diabetes mellitus, retinitis pigmentosa.*

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Introduction

Bardet-Biedl syndrome (BBS) is a rare genetic disorder with multiple systems involvement and wide spectrum of clinical features. Principal manifestations of this disorder are rod-cone dystrophy (sometimes called atypical retinitis pigmentosa), postaxial polydactyly, central obesity, mental retardation, hypogonadism and renal dysfunction.[1] Other features include hepatic fibrosis, diabetes mellitus, speech and language deficits, behavioral traits, facial dimorphism, dental anomalies and developmental delay.[1,2] BBS is distinguished from the much rarer Laurence-Moon syndrome in which retinal pigmentary degeneration, mental retardation, and hypogonadism occur in conjunction with progressive spastic paraparesis and distal muscle weakness without polydactyly.[3] Recent research has demonstrated that some individuals with the clinical findings of Laurence-Moon syndrome have had mutations in genes linked to BBS.

Author Information

a. Dr. Miah Wahiduzzaman, Senior Medical Officer, Medicine Outpatient Department, BIRDEM General Hospital, Dhaka, Bangladesh.

b. Dr. Muhammad Abdur Rahim, Assistant Professor, Department of Nephrology, BIRDEM General Hospital, Dhaka, Bangladesh.

c. Dr. Mahboob Iftekhar, Senior Medical Officer, Endocrinology Outpatient Department, BIRDEM General Hospital, Dhaka, Bangladesh.

Address of correspondence: Dr. Miah Wahiduzzaman, Senior Medical Officer, Medicine Outpatient Department, BIRDEM General Hospital, Dhaka, Bangladesh. Email: miahwahiduzzaman @yahoo.com

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Case Report

A 25-year-old male presented with the complaints of progressive deterioration of vision since 10 years of age. His parents enrolled him in a school, but he was eventually dropped out because of visual problems. He learnt to walk at the age of two and half years. He also had complains of decreased frequency of shaving since adolescence.

He was obese with a body mass index (BMI) of 31.2 kg/m^2. Other findings were polydactyly (Figure 1), micropenis with normal volume testis. He had retinitispigmentosa on fundoscopy.

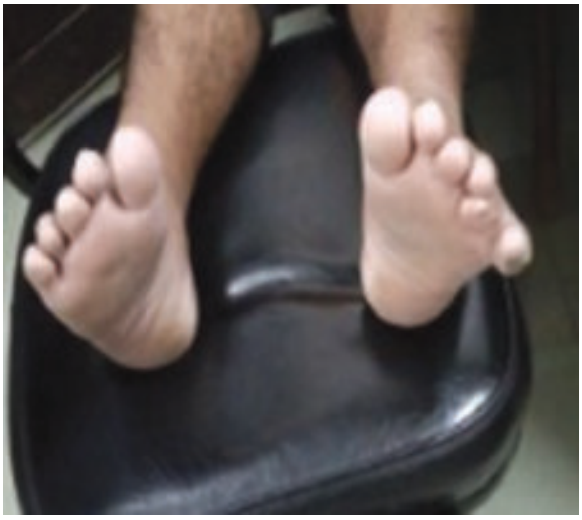

Figure 1. *Polydactyly in the left foot*

Laboratory examination included proteinuria (++), impaired renal function (serum creatinine 2.3 mg/dl), hypothyroidism (thyroid stimulating hormone 11.9 IU/ml), hypogonadism and retinitis pigmentosa (Figure 2).

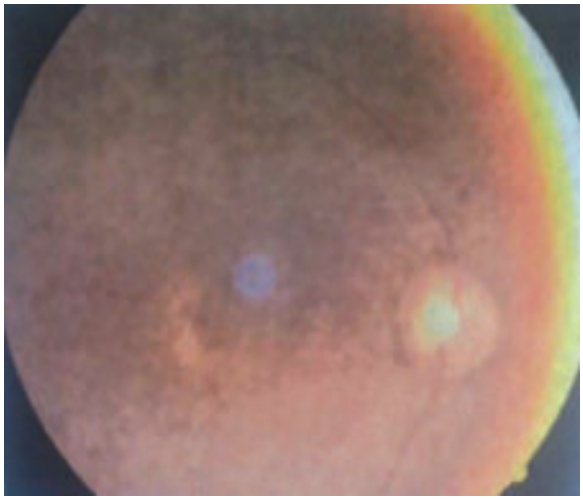

Figure 2. *Fundus photography shows features of retinitis pigmentosa*

He and his family members were counseled regarding the disease and its prognosis and treatment for hypothyroidism was initiated.

Discussion

BBS was described by Bardet and Biedl in 1920. It was later erroneously coupled with another disorder described by Laurence and Moon. BBS is distinguished from Laurence-Moon syndrome in which retinal pigmentary degeneration, mental retardation and hypogonadism occur in conjunction with progressive spastic paraparesis and distal muscle weakness without polydactyly.[4] Retinal dystrophy (100%) is the first major feature of the disorder. It is found occasionally in the first decade but present in almost all patients by the second decade.[5] Obesity is the second major feature of BBS, with a frequency of 72-96. Depending on measurement criteria. Obesity usually begins in childhood and the severity increases with age, with the majority of cases exhibiting symptoms within the first year of life.[3]

In 1999, modified diagnostic criteria were defined after a study conducted in England in 109 BBS patients.[2] Patients who had 4 primary characteristics or 3 primary and 2 secondary criteria were identified as having BBS (Table-I).

Table I. Modified diagnostic criteria[2] and clinical manifestation in present case

Primary Features	Present Case
Rod-cone dystrophy	+
Polydactyly	+
Obesity	+
Learning Disabilities	+
Hypogonadism in males	+
Renal anomalies	+
Secondary Features	
Speech disorder/delay	-
Strabismus/cataracts/astigmatism	-
Brachydactyly/ syndactyly	-
Developmental delay	-
Nephrogenic diabetes insipidus	-
Ataxia/poor coordination/imbalance	-
Mild spasticity	-
Diabetes mellitus	+
Dental crowding/hypodontia/small roots	-
Left ventricular hypertrophy/congenital heart disease	-

The treatment of BBS is directed towards the specific symptoms that are apparent in each individual. Treatment may require the coordinated efforts of a multidisciplinary team including Pediatricians, Endocrinologist, Ophthalmologists, Nephrologists, Cardiologists, Dentists, Speech Pathologists, Audiologists and other healthcare professionals. Individuals with BBS should undergo regular ophthalmologic examinations as well as periodic assessments to determine the presence of complications potentially associated with the disorder such as kidney dysfunction, diabetes mellitus, liver disfunction and high blood pressure. Early intervention is important in ensuring that children with BBS reach their highest potential. Genetic counseling may be of benefit for affected individuals and their families.

Conflict of interest: Nothing to declare.

References

1. Green JS, Parfrey PS, Harnett JD, Farid NR, Cramer BC, Johnson G, et al. The cardinal manifestations of Bardet-Biedl syndrome, a form of Laurence-Moon-Biedl syndrome. N Engl J Med 1989;321:1002-9.
2. Beales PL, Elcioglu N, Woolf AS, Parker D, Flinter FA. New criteria for improved diagnosis of Bardet-Biedl syndrome: results of a population survey. J Med Genet 1999; 36: 437-46.
3. Laurence JZ, Moon RC. Four cases of "retinitis pigmentosa" occurring in the same family, and accompanied by general imperfections of development. Ophthalmol Rev 1866;2: 32-41.
4. Schathat AP, Maumenee IH. Bardet-Biedl syndrome and related disorders. Arch Ophthal 1982;100:285-88.
5. Croft JB, Morrell D, Chase CL, Swift M. Obesity in heterozygous carriers of the gene for the Bardet Biedl syndrome. Am J Med Genet 1995;55:12-15.