

# A Case Report on the Bardet Biedl Syndrome with Hypokalaemic Paralysis

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## ABSTRACT

The Bardet-Biedl syndrome (BBS), a rare autosomal recessive disorder, was first described by Bardet and Biedl in 1920. Here, we are reporting a case of the Bardet-Biedl syndrome with hypokalaemic paralysis.

A 22 years old male patient presented with an acute onset, rapidly progressive, flaccid weakness in all four limbs. An examination revealed a moon shaped face, acanthosis nigricans, lower limb

polydactyly, central obesity, small testicular size, absence of the axillary and pubic hairs, severely impaired social adaptive functioning and retinitis pigmentosa. The central nervous system examination showed hypotonia, a grade zero power and absent reflexes. The laboratory reports showed that the patient had hypokalaemia and diabetes mellitus.

The literature showed hypokalaemic paralysis as a rare complication of the Bardet-Biedl syndrome.

**Key Words:** Bardet-Biedl syndrome, Hypokalaemic paralysis, Obesity, Retinitis pigmentosa, Ciliopathies, Autosomal recessive disorder

## CASE REPORT

A 22 years male presented with a sudden onset of weakness in all four limbs, both proximal and distal, since one day. Initially, he noticed a weakness in getting up from the squatting posture and over the next few hours, he was bedridden and could not move neither his lower limbs nor his upper limbs. However, there was no breathing difficulty. There was no history of fever, trauma, an altered sensorium, sensory disturbances, bowel bladder incontinence or cranial nerve dysfunction. His mother said that he had delayed developmental milestones and was not good at studies since childhood. Hence, he had dropped out from school. He had developed a difficulty in seeing in the dark since the past three years, which was worsening.

On physical examination, the patient was found to have a normal temperature and his vitals were stable. He had central obesity, a moon shaped face, acanthosis nigricans, lower limb polydactyly, a small testicular size and absence of the pubic and the axillary hair. His fundus examination showed the features of retinitis pigmentosa. His central nervous system examination showed hypotonia, a grade 0 power, weak neck flexors and the reflexes were absent. His sensory system was normal.

His laboratory reports showed normal blood counts, blood urea and serum creatinine. His serum potassium was 1.7 mEq/L, his random blood sugar was 286mg/dl and his fasting blood glucose was 196 mg / dl. His urine examination showed trace proteinuria. The thyroid function test, the follicle stimulating hormone, prolactin and the luteinizing hormone were within normal limits. Ultrasound of the abdomen showed calculi in the lower pole of the right kidney. ECG showed a prolonged PR interval. IQ testing showed a severely impaired social adaptive functioning.

A diagnosis of the Bardet-Biedl syndrome with hypokalaemic paralysis was made. Intravenous correction was given for the hypokalaemia and the patient improved. Potassium sparing diuretics were given on discharge. The patient was started on Insulin for his

Diabetes, which was changed to oral hypoglycaemic agents on discharge.

## DISCUSSION

The Bardet-Biedl syndrome is an autosomal recessive disorder with a wide range of clinical features. The primary clinical features include rod-cone dystrophy, postaxial polydactyly, central obesity, cognitive impairment, male hypogonadism, complex female genitourinary malformations and renal dysfunction [1,2]. The secondary features include speech disorders or delays, eye abnormalities like strabismus, cataract and astigmatism, brachydactyly or syndactyly, developmental delays, ataxia, Diabetes Mellitus, craniofacial dysmorphism, nephrogenic diabetes insipidus, hepatic fibrosis and congenital heart disease [3]. Beales et al., have suggested that the presence of four primary or three primary plus two secondary features is diagnostic [3]. The rare associations include hypothyroidism, Hirschsprung's disease, epilepsy, genital anomalies, anal stenosis and an abnormal dentition.

Our patient had polydactyly, obesity, retinitis pigmentosa, hypogonadism, mental retardation and type 2 Diabetes Mellitus i.e five primary and one secondary clinical features. Hypokalaemic paralysis was the presenting clinical feature, which made this case very rare.

So far, only 2 cases of the Bardet-Biedel syndrome have been reported from outside India with hypokalaemic paralysis [4]. Hypokalemia in BBS might be a complication of the renal dysfunction or Nephrogenic diabetes insipidus.

The Bardet-Biedl syndrome (BBS) phenotype is seen in individuals with mutations in 14 different genes [5]. BBS1 accounts for ~25-30% cases. The exact pathogenesis of BBS is unknown. It has recently been recognized that the proteins which are coded for by the BBS4, BBS6, BBS8, and the BBS10 genes are expressed in the basal body of the cilia and that BBS is now regarded as one of the 'ciliopathies' [6,7,8]. The gene products are probably involved

in the signaling pathway in the cilia; and the abnormalities interfere with a normal development, resulting in the diverse pathological effects of the syndrome.

The management of BBS is supportive and it includes training and rehabilitation for blind patients and for those with specific learning disabilities, hearing aids for deafness, and diet and exercise for obesity.

Here, we are reporting the first case of BBS with hypokalaemic paralysis from India. The diagnosis of the Bardet Biedl syndrome should be considered in the patients with the characteristic phenotype of retinitis pigmentosa, postaxial polydactyly, and central obesity. Hypokalaemia with paralysis can be a rare complication.

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