

Acute flaccid paraparesis (cauda equina syndrome) in a patient with Bardet–Biedl syndrome

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ABSTRACT

Bardet–Biedl syndrome (BBS) is a rare, autosomal-recessive, debilitating genetic disorder, which can present with multitudinous systemic clinical features including rod-cone dystrophy, polydactyly, Frohlich-like central obesity, mental retardation, hypogonadism, and renal anomalies. Diverse neuromuscular manifestations in patients afflicted by this heterogeneous disorder include ataxia, cervical, and thoracic canal stenoses, presenting as spastic quadriplegia and other gait disturbances. We report a young patient with BBS, who had presented with acute flaccid paraparesis due to severe primary lumbar canal stenosis. She underwent immediate lumbar decompression and discectomy following which she recovered significantly. Acute cauda equina syndrome due to primary lumbar canal stenosis has not been reported as a clinical feature of BBS previously.

Key words: Acute flaccid paraparesis, Bardet–Biedl syndrome, cauda equina syndrome, decompression, lumbar canal stenosis
MeSH terms: Paralysis, lower extremities, cauda equina syndrome, decompression, spinal stenosis

INTRODUCTION

Laurence-Moon and Bardet–Biedl syndrome (BBS) are heterogeneous genetic entities, whose diverse afflictions, have been discussed in the literature, ever since the initial description in 1866.^{1,2} Among the various neurological manifestations of the BBS, spinal canal stenosis at cervical or thoracic levels had been discussed in the past as a cause for spastic para- and quadriplegia.¹⁻³ The current case report involves a patient with BBS, who presented with severe primary lumbar canal stenosis and acute onset flaccid paraparesis due to cauda equina syndrome. The case is discussed in view of the extreme rarity of such a clinical presentation.

CASE REPORT

A 22-year-old female patient presented to us with acute onset radicular pain and weakness in both lower limbs.

She had sustained a trivial fall at home following which she developed the symptoms. She was a known case of BBS, with typical clinical features including night blindness, obesity, and polydactyly [Figure 1a-c]. Before the fall, she had been ambulatory despite these disabilities. On clinical examination, she had localized tenderness over the lumbosacral region. Neurological examination revealed spasticity in both upper limbs but reduced tone in lower limbs. Motor examination revealed weakness below L2 myotome bilaterally (right side L2-S1: 1/5, left side: 3/5). Sensations were grossly normal bilaterally in the upper limbs, but patchy hypoesthesia was present in lower limbs in L3, L4, L5, and S1 dermatomes. Deep tendon reflexes were exaggerated in the upper limbs, but absent in the patellar and Achilles tendons bilaterally. This pattern was suggestive of lower motor neuron type of pathology in the lower limbs. She was voiding per urethrally and had some urinary hesitancy. On per rectal examination, perianal sensation and anal tone were decreased.

Roentgenograms of the lumbar spine revealed loss of lordosis, without any obvious instability. Magnetic resonance

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Figure 1: Clinical photographs with typical clinical features of Bardet–Biedl syndrome in our patient including, (a) typical moon-shaped faces (obesity), (b) polydactyly in both lower limbs and (c) polydactyly in both upper limbs

imaging (MRI) scan of the lumbar spine showed severe primary canal stenosis of the lumbar canal (anteroposterior diameter – 6–7 mm), L1–L2 central disc protrusion, and L3–L4, L4–L5 annular disc bulge with secondary canal narrowing [Figure 2a–d]. MRI scan of dorsal and cervical spine revealed no significant abnormalities [Figure 3]. The Torg–Pavlov ratios at the cervical and lumbar levels were calculated, which revealed significantly narrowed bony canal [Figure 4a, b and Table 1].

She underwent immediate L3–L4 and L4–L5 midline decompression, with laminotomy and discectomy at L1–L2 level. Postoperatively, she had a good recovery and gradual improvement in the neurological status, noticed initially at the end of two weeks. She could be ambulated with support over the next 2 weeks. The neurological status improved significantly at 6 months postoperatively, and she was walking independently without support with a neurological grade of Frankel D [postoperative MRI scan – Figure 5a–c].

DISCUSSION

An unusual clinical presentation of acute flaccid paraparesis in a patient of BBS, secondary to severe primary lumbar canal stenosis has been described in this report. The rarity of such a clinical presentation, diagnostic dilemma involved in these scenarios, management options available, and a literature review of the condition are described. Though spastic parapareses due to cervical and thoracic stenoses have been described in BBS, acute flaccid paralysis due to lumbar canal stenosis has not been described in the literature.

The earliest formal description of Laurence–Moon–Biedl syndrome was made by Laurence and Moon⁴ in 1866. In 1920, George Bardet¹ and Artur Biedl independently published the syndromic presentation of hypothalamic obesity, retinitis pigmentosa, polydactyly, and mental retardation in a certain group of patients. Beales *et al.*,¹

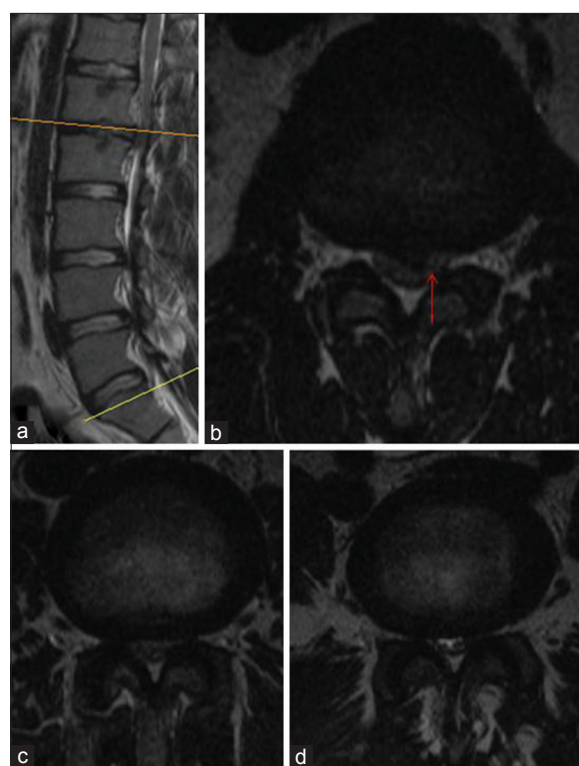


Figure 2: Preoperative T2-weighted magnetic resonance images (a) Mid-sagittal section showing severe primary canal stenosis of the lumbar canal with L1–L2 disc protrusion and L3–L4, L4–L5 annular disc bulge with secondary canal narrowing. (b) Corresponding axial section image at L1–L2 level showing L1–L2 left paracentral disc protrusion. (c) Corresponding axial section image at L3–L4 level showing L3–L4 central canal and foraminal stenoses. (d) Corresponding axial section image at L4–L5 level showing L4–L5 central canal and foraminal stenoses

in 1999, studied 109 BBS patients and their families, one of the largest series on this disorder until date and had comprehensively described the clinical criteria for the diagnosis of this syndrome. Typically, a patient with BBS is described to present with specific features of toe anomalies and obesity, in addition to the other cardinal manifestations of Laurence–Moon–Biedl syndrome.

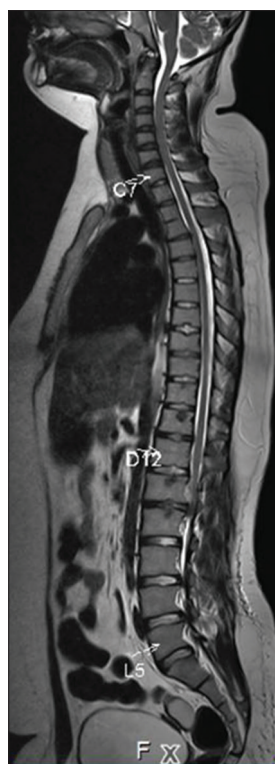


Figure 3: Preoperative T2-weighted mid-sagittal magnetic resonance image of whole spine showing no significant compression at the thoracic or cervical levels

Table 1: Preoperative mid-sagittal canal anteroposterior dimensions, vertebral body anteroposterior dimensions, and Torg–Pavlov ratios at C3–C7 and L1–L5 vertebral levels

Vertebral level	Canal AP diameter (mm) - A	Vertebral body dimension (mm) - B	Torg–Pavlov ratio - A/B
C3	9.4	17.5	0.53
C4	8.9	16.4	0.54
C5	9.1	16.8	0.6
C6	8.9	19.4	0.46
C7	10.1	18.1	0.56
L1	11.6	42.8	0.27
L2	13.8	43	0.32
L3	12.9	41.9	0.31
L4	10.5	42.5	0.25
L5	12.1	38.6	0.31

AP=Anteroposterior

Typical neurological manifestations of BBS include speech and hearing deficits, learning disabilities, behavioral problems, ataxia with poor coordination, gait abnormalities, and spastic motor deficiencies of the limbs.³ Among them,^{1,3} learning disabilities (62%) and developmental delay (50%) are the most frequently described. Spinal pathologies, including cervical and thoracic primary spinal canal stenoses, have been infrequently linked to gait disturbances observed in these patients. Nyska *et al.*⁵ described a 36-year-old patient suffering from Laurence–Moon–Biedl–Bardet syndrome, who had significant cervical canal myelopathy and had

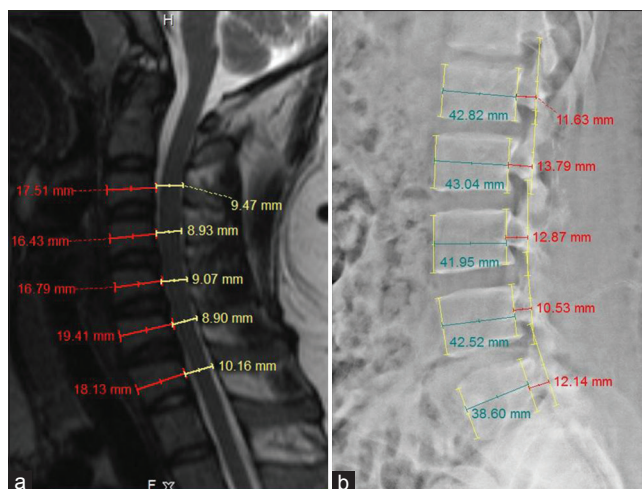


Figure 4: Preoperative Torg–Pavlov ratios (a) sagittal section image at the mid-sagittal plane showing mid-sagittal anteroposterior spinal canal and anteroposterior vertebral body dimensions at C3, C4, C5, C6, and C7 vertebral levels. (b) Lateral Roentgenogram showing the mid-sagittal anterior-posterior spinal canal and anteroposterior vertebral body dimensions at L1, L2, L3, L4, and L5 vertebral levels

recovered following cervical laminoplasty. Similar reports of spastic tetraparesis following cervical and thoracic canal stenoses have been made by Moses *et al.*⁶ and Uçar *et al.*⁷ However, no previous description of severe lumbar canal stenosis presenting precipitously as cauda equina syndrome in a patient with BBS has been made in the literature, until date.

Our patient was unique in her clinical presentation with significant lumbar spinal canal stenosis, and acute onset cauda equina syndrome. In a case of BBS presenting with neurological manifestations, one may expect cervical or thoracic canal stenoses. However, the present patient had a rare presentation of acute, rapidly progressing lower motor neuron type of neurological dysfunction in both lower limbs. Early diagnosis and timely treatment in such situations are extremely important, as immediate decompression is the best way to facilitate neurological recovery in these patients.⁸

Primary bony canal stenosis in patients with skeletal dysplasias is generally managed with wide laminectomy and instrumented fusion. We, however, performed midline fenestration and decompression at L3–L4 and L4–L5 levels, with laminotomy and discectomy at L1–L2 level in our patient. Since the stress radiographs in our patient did not reveal a significant instability, we did not perform any additional instrumentation or fusion procedures. She had a significant neurological recovery following the surgery.

Patients with congenital skeletal dysplastic syndromes including achondroplasia⁹ and spondyloepiphyseal

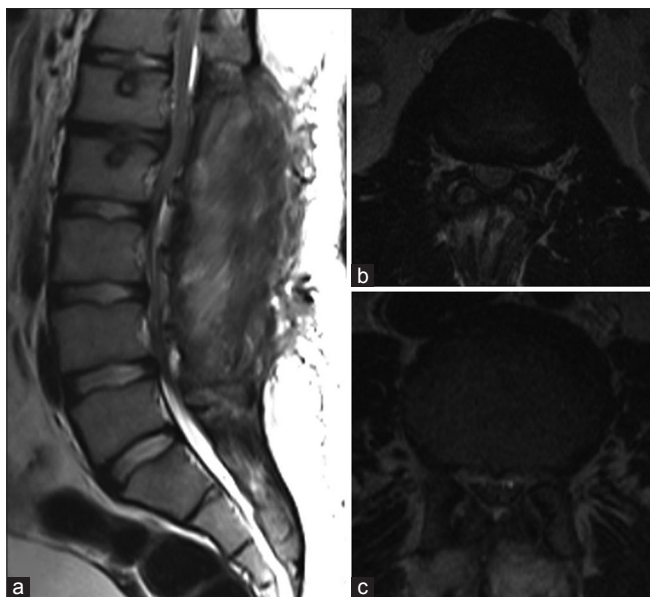


Figure 5: Postoperative T2-weighted magnetic resonance imaging scans (a) Mid-sagittal section showing a well-decompressed spinal canal following L1-L2, L3-L4, and L4-L5 surgical decompression. (b) Corresponding axial section image at L1-L2 level showing a well-decompressed spinal canal. (c) Corresponding axial section image at L3-L4 level showing a well-decompressed spinal canal

dysplasia have been reported to present with severe, primary bony canal stenosis. Early spine and brain MRI screening has been recommended as a routine protocol in these patients with subtle neurological manifestations to rule out compressive craniospinal pathologies. A similar approach in patients with BBS may be advocated for reasons, thus discussed.

This rare case highlights an extremely rare scenario, which may be encountered in BBS patients. The primary objective of the article is to discuss the rarity of the clinical presentation, the need to obtain MRI of all regions of spine in BBS and the good results obtained with early surgery. Although quadri- and paraparesis are known to occur in BBS, this case report describes an unreported occurrence of cauda equina syndrome due to compression at the lumbar spine. Timely diagnosis and immediate surgical intervention are clearly advocated in these patients. Simple, noninstrumented decompression surgeries performed at

the earliest, as in our patient, can provide excellent results in such situations.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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