Paraplegia **29** (1991) 350–354

Paraplegia

Quadriparesis in the Laurence-Moon-Biedl-Bardet Syndrome: Case Report

M. Nyska, G. Mozes, C. Howard, J. Bar-Ziv, S. Dekel
Department of Orthopedic Surgery, Soroka Medical Center, The Ben-Gurion University of The Negev, Beer-Sheva, Israel.

Summary

A 36 year old patient known to suffer from the Laurence-Moon-Biedl-Bardet syndrome (LMBBS) developed spastic quadriparesis. The typical features of the syndrome, presented by this patient were polydactyly, obesity, hypogonadism, retinitis pigmentosa and relative mental retardation. Severe spinal cervical and lumbar canal stenosis imaged by plain X-rays and computerised tomography was found. Magnetic resonance imaging showed significant atrophy of the spinal cord, indicating that the cause of the quadriparesis was cervical myelopathy. The patient underwent laminoplasty with some improvement.
Key words: *Laurence-Moon-Biedl-Bardet syndrome; Cervical myelopathy; Quadriparesis.*

The cardinal features of the Laurence-Moon-Biedl-Bardet syndrome (LMBBS) are polydactyly, obesity, hypogonadism, retinitis pigmentosa and mental retardation.[6] In order to establish the diagnosis at least four out of the five cardinal clinical features must be present. However, there are at least seven hereditary syndromes with pigmentary retinopathy which are related with LMBBS.[1] These syndromes includes deafness, ataxia, diabetes mellitus, renal involvement, quadriparesis or paraplegia.

In 1866 Laurence and Moon described four members of a family with short stature, hypogonadism, mental retardation and spino-cerebellar ataxia minanting the clinical picture.[5] All 4 patients had nistagmus. These patients subsequently developed spastic paraplegia.[4] Gordon *et al*, reported 2 patients suffering from retinitis pigmentosa, mental retardation, hearing loss and progressive quadriparesis.

Correspondence to: M. Nyska, Department of Orthopedic Surgery, Soroka Medical Center, P.O.B. 151, Beer-Sheva, Israel 84101.

No definite pathology was reported in those cases as the cause for the quadriparesis or paraplegia.[3]

Recently we encountered a patient with LMBBS characterised by polydactyly, obesity, retinitis pigmentosa, hypogonadism and borderline mental retardation who late developed spastic quadriparesis. Further study of this patient revealed severe cervical and lumbar spinal canal stenosis. We have not encountered a similar case who had spinal canal stenosis associated with LMBBS. The patient and his treatment are presented and discussed.

Case report

A 36-year-old patient suffering from the Biedl-Bardet syndrome with postaxial polydactyly, retinitis pigmentosa, obesity, hypogonadism and borderline mental retardation was hospitalised for investigation because of difficulties in walking and micturition for the previous two years. He was unable to stand up even with assistance. Neurological examination disclosed lead pipe spastic quadriparesis with the lower extremities being more spastic than the upper extremities and the right side more spastic than the left. Patellar and ankle jerks were very brisk, with patellar and ankle clonus and bilateral Babinski signs. There was a symmetrical sensory hypoesthesis from the C–2 dermatome level. Hip joint movement was limited. The clinical grade of the patient was estimated to be one point according to the Japanese Orthopedic Association score.[2] All routine laboratory blood tests were normal. Urodynamic studies disclosed a spastic bladder sphincter which necessitated sphincterotomy. X-rays and computerised tomography demonstrated severe cervical and lumbar canal stenosis (Fig. 1 and 2). The narrowest interpedicular distance of cervical vertebra was 15 mm, and in the lumbar canal 14 mm. The smallest antero-posterior distance was 3 mm at the C–3 vertebra, and 10 mm at L–3 vertebra. There was also mild platyspondyly and Schmorl nodes of almost all thoracic vertebral bodies. Magnetic resonance imaging of the spine showed almost complete block of the cervical canal with atrophy of the cord at the C–3, C–4, and C–6 levels (Fig 3). Similar changes were imaged by magnetic resonance in the lumbar spine (Fig 4). The patient underwent laminoplasty from

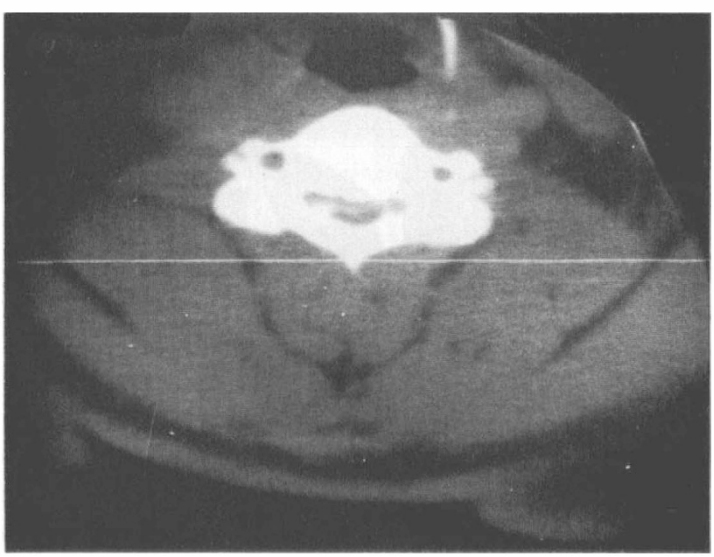

Figure 1 Computerized tomography of the cervical spine showing severe narrowing of the spinal canal.

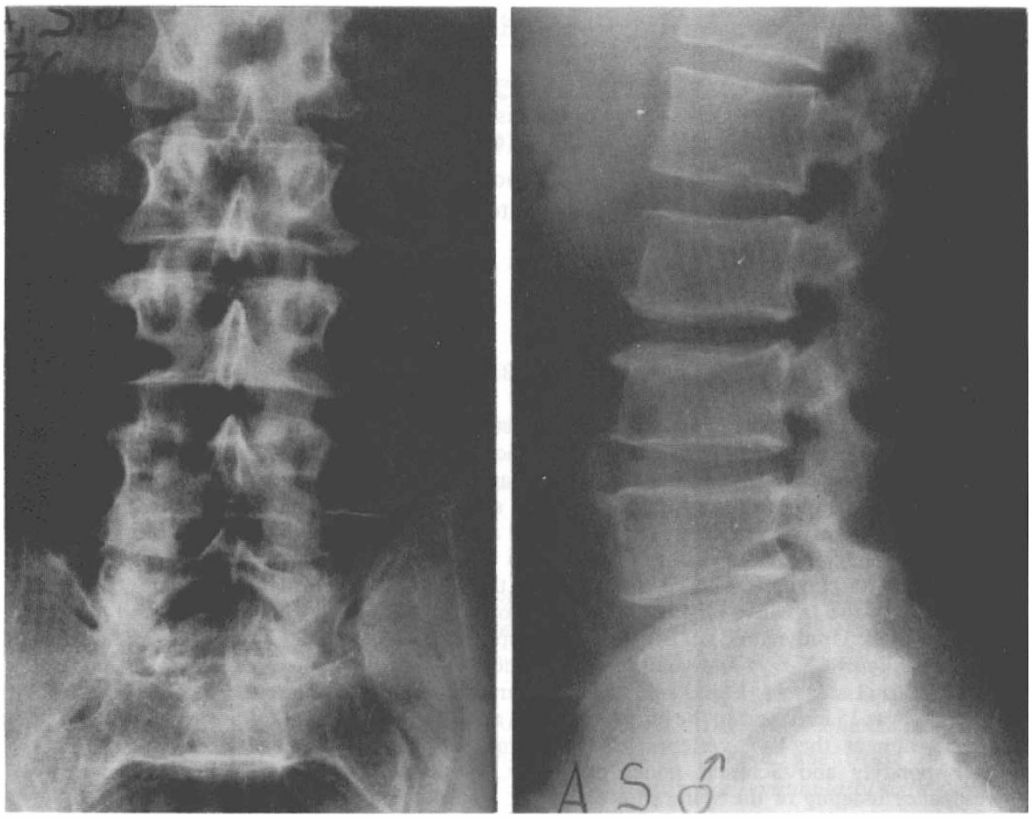

Figure 2 Lumbar spine X-rays demonstrating spinal canal stenosis.

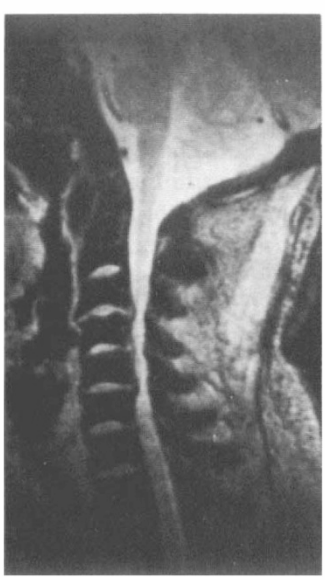

Figure 3 Magnetic resonance imaging of the cervical spine showing almost complete block with severe cord atrophy.

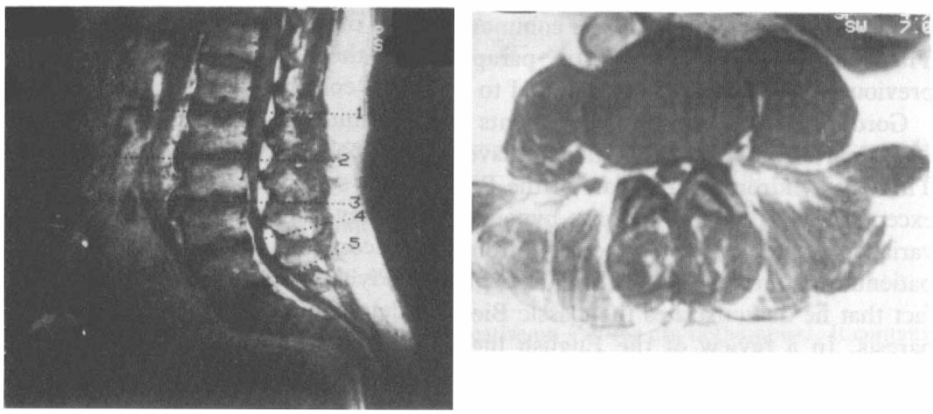

Figure 4 Magnetic resonance imaging of the lumbar spine demonstrating severe spinal stenosis with cord atrophy.

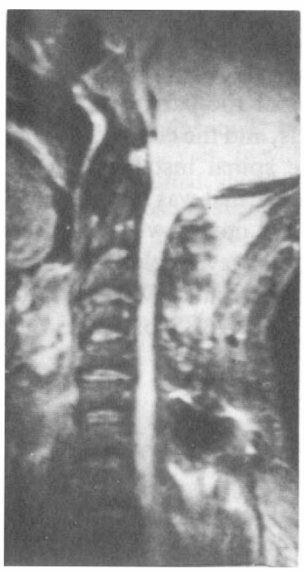

Figure 5 Post-operative magnetic resonance imaging of the cervical spine. The laminoplasty from C–3 to C–6 relieved the pressure on the cord.

C–3 to C–6. Magnetic resonance imaging 3 months after the operation showed widening of the cervical spinal canal with no direct pressure on the cord itself (Fig 5). Six months following the operation he still had spastic quadriparesis, but some improvement was noticed as he was able to stand up for a short time with help.

Discussion

The cardinal symptoms of our patient are typical of the Biedl-Bardet syndrome. He had polydactyly, obesity, hypogonadism, retinitis pigmentosa and slight mental retardation.[6] The patients published by Laurence and Moon did not have obesity and polydactyly, but they did have spastic paraplegia.[5] Our patient had spastic

quadriparesis in addition to the common features of the Biedl-Bardet syndrome. The description of the spastic paraplegia in the Laurence-Moon patients, previously described, was not related to the spinal cord myelopathy.

Gordon *et al.* found in 2 patients with retinitis pigmentosa, progressive quadriparesis.[3] His patients did not have obesity, polydactyly or hypogenitalism. Thus his syndrome is not part of the Biedl-Bardet syndrome. However with the exception that they did not have hypogenitalism they can be considered as a close variant of the Laurence-Moon syndrome. The common spinal features of his patients and ours is that they both have platyspondyly. Our patient is unique in the fact that he demonstrates the classic Biedl-Bardet syndrome with spastic quadriparesis. In a review of the English literature we were unable to find such an association.

The diagnosis of cervical and lumbar spinal canal stenosis, was obvious in plain X-rays, as the interpedicular and antero-posterior distance of the spinal canal were significantly decreased. Computerised tomography confirmed the presence of severe spinal canal stenosis, but only magnetic resonance imaging demonstrated accurately the severe atrophy of the spinal cord itself indicating that cervical myelopathy was the main reason for the spastic quadriparesis.

The poor functional status of the patient may be explained by the significant stenotic canal at multiple levels, and the chronicity of the symptoms.[2] Laminectomy at multiple levels may cause spinal instability, and therefore laminoplasty was chosen. However, although the canal was wider after the operation, post-operative magnetic resonance imaging did not show any improvement in the cross sectional area of the cord. Presumably irreversible cord atrophy had occurred prior to surgery, leading to poor clinical results. We conclude that spinal stenosis may be the cause for quadriparesis in LMBBS patients. Early operative treatment may result in a better clinical result.

References

1. Cantany A, Belioni P, Bamonte G, Salvinelli F, Bamonte MT. Seven hereditary syndromes with pigmentary retinopathy. A review and differential diagnosis. Clin Pediatr (Phila) 1985:**24**:578–583.
2. Fujiwara F, Yonenolu K. The prognosis of surgery for cervical compression myelopathy. J Bone and Joint Surg.
3. Gordon AM, Capute AJ, Konigsmark BW. Progressive Quadriparesis, Mental Retardation, Retinitis Pigmentosa, and Hearing Loss: Report of Two Sibs. The Johns Hopkins Medical Journal 1976:**138**:142–145.
4. Hutchinson J. Retinitis Pigmentosa and Allied Affections. As Illustrating The Laws Of Heredity. Ophtalmol Rev 1882:2–30.
5. Laurence JZ, Moon RC. Four cases of retinitis pigmentosa occurring in the same family and accompanied by general imperfections of development. Rev Ophtal 1866:**2**:33.
6. Schachat AP, Maumene JH. Bardet-Biedle syndrome and related disorders. Arch Ophthalmol 1982:**100**:285–288.