



Central and Peripheral Demyelination with Immune Polymyositis in a Patient with Bardet Biedl Syndrome

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Abstract

We report extremely rare combination of central and peripheral demyelination with immune polymyositis in a patient with Bardet – Biedl syndrome. The inciting factor postulated was mycoplasma pneumonia infection.

Introduction

Clinical scenarios, at times can be so intriguing that the treating physician may find it difficult to arrive at a single diagnosis. This is especially true in immune mediated disorders where multiple systems or different components of the same system can be involved simultaneously. We report one such extremely rare combination of central, optic nerve and peripheral demyelination with immune polymyositis in a patient with Bardet – Biedl syndrome.

Case report

The patient was a 23 year old male, born of a non consanguineous marriage. His illness started with an episode of lower respiratory infection which was treated with azithromycin. A week later he developed severe pain in the left thigh followed by the right thigh. Following the day he developed weakness involving all the four extremities. Weakness was more in the lower limbs and predominantly proximal than distal. For this he received indigenous treatment,¹ Five days later he developed weakness with neck and trunk muscle weakness with retention of urine. He was catheterized and referred to us.

On examination, pulse was 80 per minute, Blood pressure was 130/80 mm Hg in the right upper limb. He weighed 85 kg, was 165 cms tall Body mass index was 31.2. He had postaxial polydactyly of all limbs (fig 1). Secondary sexual characters were normal. There were no neurocutaneous markers.

Neurological examination revealed a MMSE score of 15/30, impaired attention span, calculation, memory, and cognition. Fundus examination showed bilateral retinitis pigmentosa (fig 2). Other cranial nerves were normal. There was severe muscle tenderness involving both the thighs with hypotonia, hyporeflexia and predominantly proximal quadriparesis. Plantars were bilaterally flexor. Rest of the examination was normal.

His hemogram was normal. ESR was 34 mm in 1 hr. Renal functions, urine microscopy and ultrasound of the abdomen were normal. He was euthyroid and semen analysis was normal. Spinal fluid examination showed albuminocytological dissociation (protein 66 mg/dL; sugar 74 mg/dL; no cells). The CSF culture was negative. No oligoclonal bands were seen. C reactive protein was positive, creatinine kinase was 18,175 u/L

MB fraction u/L 184. Nerve conduction study was suggestive of peripheral axonal demyelination with prolonged F wave latencies.

He was treated with iv methylprednisolone 1 gm/day for 5 days followed by close and oral prednisolone. While on treatment, he developed sudden painless loss of vision with drowsiness which lasted for 5 hours. On examination vision was limited to perception of light with relative afferent papillary defect. Fundoscopic examination showed bilateral optic neuritis..

A repeat CSF examination showed details. Creatinine kinase was 3896 u/L with MB fraction 84 u/L. Repeat nerve conduction study showed a pattern of axonal demyelination. Electromyography was suggestive of myopathy. MRI of brain showed focal T2/FLAIR hyper intensities in the sub cortical white matter of right occipital lobe and bilateral posterior temporal region suggestive of cerebral demyelination (fig 3). Biopsy of the left quadriceps muscle revealed preserved architecture with polygonal to round myofibres, lymphocytic infiltration, internalization of nuclei, polyfocal necrosis, myophagocytosis, rhabdomyolysis and regenerating fibers. The features were compatible with immune polymyositis.

A diagnosis of recurrent demyelination (cerebral, optic and peripheral) with immune polymyositis secondary to mycoplasma pneumonia infection was made. Though the serological test for mycoplasma was not performed, the manifestations were thought to be an immune response to mycoplasma pneumoniae since the patient had a preceding lower respiratory infection. A few similar cases have been reported in the literature.

Discussion

The combination of immune demyelination with polymyositis is extremely rare and very few cases have been reported in the literature. Most were due to mycoplasma pneumoniae.^{3,4} One was due to hepatitis A infection,² while no cause could be found for one case.¹ This occurs due to antibodies produced against normal human antigens like serum anti-galactocerebroside in Guillian Barre syndrome and optic neuritis, anti-I and Sia-b1 auto antibodies in autoimmune hemolysis.

The CNS manifestations of mycoplasma pneumoniae infection include meningoencephalitis, aseptic meningitis, encephalitis, ascending paralysis and acute transverse myelitis. Musculoskeletal involvement is in the form of immune polymyositis and arthritis.

Though, our case could not establish the etiology, considering the history and clinical presentation mycoplasma pneumonia appears the most likely cause. The case has been reported due to

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Fig. 1 : Postaxial polydactyly of all the four limbs



Fig. 2 : Retinitis pigmentosa

the extreme rarity of the combination of immune demyelination with polymyositis.

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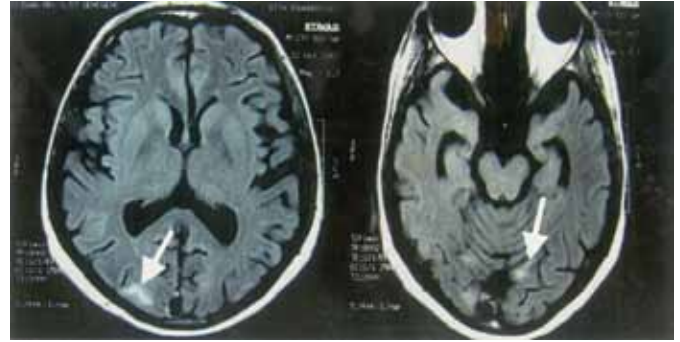


Fig. 3 : MRI brain showing areas of demyelination

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