



Brachial Amyotrophic Diplegia: A Rare Variant of Motor Neuron Disease- Case Report

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ABSTRACT

BRACHIAL AMYOTROPHIC DIPLEGIA (BAD) is a syndrome of proximal weakness and wasting of the upper limbs. The syndrome typically presents with progressive upper limb weakness and wasting that is often symmetric and proximal, without significant functional involvement of lower limbs or bulbar muscles. Here we presented a patient with complaints of difficulty in lifting his right arm in his medical history. Brachial amyotrophic diplegia was diagnosed with neurological examination and EMG findings. It is presented because of its extreme rarity and rare clinical manifestations.

INTRODUCTION

Motor neuron diseases (MND) are a heterogeneous group of disorders affecting the upper motor neuron, lower motor neuron or both. Amyotrophic lateral sclerosis is the most common type of MND, focal MNDs are relatively rare.

Brachial amyotrophic diplegia (BAD) and leg amyotrophic diplegia (LAD) are variants of progressive muscular atrophy with disease confined to one spinal region.

Isolated bulbar ALS have symptoms confined to speech and swallowing, and can be comprised of both upper and lower motor neuron involvement. Brachial amyotrophic diplegia is a sporadic disorder confined to upper limb for long periods. Proximal limb is involved, bulbar muscles usually

spared with predominant LMN signs in the affected limb.

CASE REPORT

A 64 year old male patient, fisherman by profession with no significant past history or family history presented with insidious onset slowly progressive bilateral symmetrical weakness of both shoulder girdle for five years. He ends up with severe symmetrical thinning of muscles of both shoulder. Patient was not able to raise his right arm and minimal movement was possible with the left shoulder. The weakness was painless without any sensory disturbances. There was no history of lower limb weakness, cranial nerve involvement or disturbances of higher mental function.

On examination his vitals were normal. General physical examination was normal.

Nervous system examination showed normal higher mental function and cranial nerves.

There was bilateral severe wasting of supra spinatus, infra spinatus, deltoid, biceps and rhomboids without winging of scapula.

Tone was normal for both the upper limb and lower limb.

Power was grade 2+ for the right shoulder abduction, adduction and rotator movements (medical research council grading) 4/5 grade for wrist and finger movements. Power was grade 3+ for the left shoulder movements. Forearm muscle power was near normal with bilateral strong hand grip. Facial muscle strength, neck flexion and extension, lower limb power was normal. Superficial reflexes were normal.

Deep tendon reflexes were sluggish in both upper limb but the reflexes were well preserved in lower limb. Plantar was B/L flexor.

Fasciculations were noted in right shoulder. Sensory system examination was normal. Gait was normal. Skull and spine normal including full movement of spine

Other system examination does not reveal any abnormality

Investigations revealed normal routine blood results.

BRE, RFT, LFT, RBS, FBS, PPBS, HBA1C, VDRL and viral markers was normal. Peripheral smear-normal

TFT, PTH, S.Electrophoresis, Ach and Musk Antibody, S.calcium, S.phosphorous - normal.

Serum immunoglobulin -normal

Creatine kinase (CK) level was slightly elevated - 240 IU.

ANA including ANA profile -negative,

P ANCA, C ANCA -negative

GM1 Ab Titers were normal.

X-RAY both Shoulder – Mild Osteoporotic changes were present

MRI spine showed mild atrophy of cord with degenerative changes in cervical region.

Nerve conduction studies (NCV) showed low amplitude median and ulnar compound muscle action potentials (CMAP).

There was no electrodiagnostic evidence of conduction block or demyelination.

Median, ulnar and radial sensory nerve action potentials (SNAP) were normal.

EMG: Positive sharp waves and fibrillation

Lower limb electro physiological study was normal

IN view of pure motor weakness, wasting of proximal muscles of upper limb without any sensory involvement, without cranial nerve involvement or higher function and electrophysiological evidence of focal motor neuron disease, we arrived at a diagnosis of motor neuron disease (Brachial Amyotrophic Diplegia).



Fig 1,2,3 – Wasting of shoulder girdle

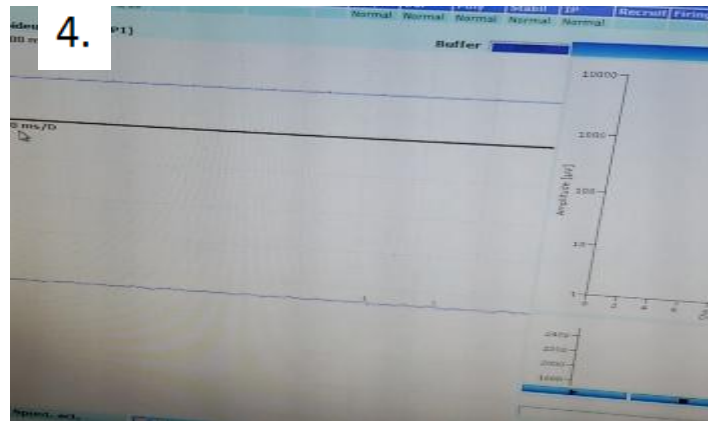


Fig 4 – Fibrillatory waves of right posterior deltoid in EMG showing spontaneous activity



Fig 5 – Positive sharp wave of left deltoid in EMG showing spontaneous activity

DISCUSSION

Brachial Amyotrophic Diplegia is a segmental motor neuron disease. It is also known as “men in barrel syndrome” or “flail arm syndrome”. It results in severe lower motor weakness and atrophy of muscles of upper extremities in the absence of bulbar or lower extremity involvement. The disease usually does not show pyramidal signs, bowel and bladder involvement. The disease shows sporadic occurrence pattern and benign course without affection of bulbar and lower extremity. The disease has been found to be associated with patients with retro viral infection. Classic clinical presentations for sporadic ALS include limb-onset (about 2/3), bulbar onset (about 1/3), and pure lower motor neuron (~5%)

Some patients have disease isolated to single spinal regions for years

These potentially slower progressing regional variants include:

1. Brachial amyotrophic diplegia (BAD)

2. Leg amyotrophic diplegia (LAD)

3. Isolated bulbar ALS (IBALS)

The differential diagnosis for BAD includes bilateral cortical watershed infarcts, spinal cord infarction etc. The main differential for BAD in the absence of other clinical or diagnostic abnormalities is regional presentation of classic ALS. Patients are more likely to be male than the general ALS population. Symptoms can begin asymmetrically but usually progress to include both arms. Unlike arm-onset ALS which usually presents with distal weakness, the majority of BAD patients have proximal weakness at presentation. Most of the patients with BAD have only lower motor neuron involvement at presentation, with decreased or absent reflexes.

Common features of diagnosing BAD include:

- Insidious onset of weakness in the proximal arm muscles
- Decreased or absent reflex

- Symptoms confined to one spinal region for 12–18 month.
- In the absence of sensory symptoms or signs

Diagnostic testing should include:

Normal MRI of the cervical spinal cord

Negative GM1 antibody testing

Consideration for genetic testing for spinal muscular atrophy and/or spinobulbar muscular atrophy

Prognosis: Overall the prognosis for BAD is better than classic ALS. There is increased 5 and 10 year survival in patients with BAD compared to classic ALS

Leg Amyotrophic Diplegia

LAD patients present with a pelviperoneal pattern of weakness, or diffuse or distal weakness

Reflexes are reduced or absent

May be more common in male patients

About 25% progress to include a second spinal cord region at 2 years of follow up

Diagnosis: The definitions for LAD differ by case series but common features include:

- Insidious onset of weakness isolated to the legs
- Decreased or absent reflexes at presentation
- Symptoms confined to one spinal region for 12–24 months
- In the absence of Sensory symptoms or signs

Diagnostic testing should include:

MRI of the spine

Negative GM1 antibody testing

Prognosis: LAD have a better overall prognosis than classic ALS.

Isolated Bulbar ALS

IBALS have weakness confined to the bulbar region. Patients may have difficulty with speech or swallowing but respiration is usually preserved. Patients may show flaccid or spastic dysarthria.

Possibly more common in female patients

Diagnosis: The diagnosis of IBALS is suggested by the presence of:

- Insidious onset of symptoms isolated to the bulbar region for at least 6 months
- Spastic or flaccid dysarthria or mixed dysarthria
- Preserved respiration at presentation
- In the absence of Sensory symptoms or signs

Diagnostic testing should include

MRI of the brain

Consider auto antibody screening (acetylcholine receptor antibodies, LEMS antibody possibly)

Clinically may show pathologically brisk reflexes, including positive jaw jerk or Hoffman's sign.

Electromyography should show denervating changes isolated to the bulbar region

Prognosis: IBALS also appears to have a more benign prognosis compared to classic ALS

CONCLUSIONS

Amyotrophic lateral sclerosis is a rapidly progressive, invariably fatal disease, comprised of mixed upper and lower motor neuron involvement in different spinal cord regions. Regional variants have been described where disease is restricted to one spinal region at presentation including Brachial amyotrophic diplegia.

These regional variants do not meet clinical diagnostic criteria for definite ALS and usually carry a better prognosis than classical ALS.

A better understanding of the natural history of regional variants is essential for diagnosis and treatment of these rare diseases.