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# Dyspnoea-fasciculation syndrome: 'the clue is in the title'

James Rigby,[1] Andrew Holton,[2] Jonathan Partridge,[3] Duwarakan Satchithananda[1]

[1]Department of Cardiology, University Hospital of North Staffordshire, Stoke-on-Trent, UK
[2]Department of Neurophysiology, University Hospital of North Staffordshire, Stoke-on-Trent, UK
[3]Department of Neurology, University Hospital of North Staffordshire, Stoke-on-Trent, UK

**Correspondence to** Dr James Rigby, jamesrigby1@hotmail.co.uk

## DESCRIPTION

A non-smoking 71-year-old man had suffered myocardial infarction 6 years back. After a circumflex angioplasty, he remained asymptomatic with moderately impaired left ventricular function. This was unchanged when he presented with 6 months' progressive breathlessness and orthopnoea, but no chest pain. There was insufficient evidence on CT to explain his restrictive spirometry. A diagnostic coronary angiography was arranged, but soon abandoned when he developed pulmonary oedema while recumbent. Frequent fasciculations were noted (video 1). The pulmonary oedema resolved with treatment, but his type II respiratory failure and orthopnoea persisted. Troponin was not raised.

**Video 1** Fasciculations in the latissimus dorsi muscle when the patient was lying supine during cardiac catheterisation.

Retaking the history revealed 3 months' weight loss alongside general weakness. He had noticed he was not able to lift his arms. Over the last 4 months there had been progressive loss of speech articulation, cramps in hands and difficulty in arising from a chair. Ankle jerks were absent.

Clinical neurophysiology revealed sensory responses generally absent or reduced, as were the lower limb motor responses. Motor response in his right hand was not augmented after a 10 s maximal contraction (against Lambert-Eaton myasthenia). There was no decrement in repetitive nerve stimulation. A concentric needle electromyography (EMG) (table 1) identified an active denervation in upper and lower limbs, extending into the cranially innervated territory. Notwithstanding the axonal-type polyneuropathy, disproportionate diffuse motor axonal loss supported amyotrophic lateral sclerosis.

Symptoms improved with non-invasive ventilation, suggesting pulmonary oedema had arisen from insufficient respiratory excursion while recumbent. Dyspnoea–fasciculation syndrome is a presentation of Amyotrophic Lateral Sclerosis.[1] Type II respiratory failure from weakness can underlie orthopnoea, in our case exacerbated when supine for cardiac catheterisation.

**Table 1** Findings of concentric needle electromyography that supports the diagnosis of motor neuron disease

| | | | Maximum effort | |
|---|---|---|---|---|
| **Muscle** | **Spontaneous activity** | **Motor units** | **Number** | **Maximum size (mV)** |
| Right tibialis anterior | Fasciculations 3+<br>Positive sharp waves 3+<br>fibrillations 2+ | Long-duration polyphasic units | Discrete | 4 |
| Left tibialis anterior | Fasciculations 3+<br>Positive sharp waves 1+<br>Fibrillations 2+ | Long-duration polyphasic units | Discrete | 4 |
| Right vastus lateralis | Fasciculations 3+<br>Fibrillations 2+ | Long-duration polyphasic units | Reduced | 2.5 |
| Right first dorsal interosseous | Fasciculations 4+ | Long-duration polyphasic units | Discrete | 4 |
| Right biceps brachii | Fasciculations 3+ | Long-duration polyphasic units | Reduced | 4 |
| Right trapezius | Fasciculations 2+ | Long-duration polyphasic units | Discrete | 1 |

## Learning points

- Though breathlessness is a single reported symptom it can be caused by many individual processes.
- Orthopnoea should not be assumed to be due to a heart failure without a detailed clinical evaluation. Orthopnoea associated with worsening type II respiratory failure might be neuromuscular or mechanical until proven otherwise.
- Multiple (and often invasive) investigations are an unacceptable substitute for clinical re-evaluation—including a comprehensive history and examination—irrespective of the perceived time constraints of increasingly busy medical clinics.

**Competing interests** None.

**Patient consent** Obtained.

## REFERENCE

1. **Scelsa SN,** Yakubov B, Salzman SH. Dyspnea-fasciculation syndrome; early respiratory failure in ALS with minimal motor signs. *Amyotroph Lateral Scler Other Motor Neuron Disord* 2002;**3**:239–43.