

## Severe diaphragm weakness in spinocerebellar degeneration

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Spinocerebellar degeneration is characterised by weakness and spasticity of the legs with early onset in childhood or adolescence and slow progression.<sup>1</sup> The pathological changes are those of a chronic degenerative process affecting the spinal cord and cerebellum. Other areas of the nervous system may be affected but diaphragmatic weakness has not previously been reported.

### Case report

A 23 year old housewife presented with dysarthria, which she had had since the age of 11, and mild dyspnoea and difficulty in swallowing of recent onset. Her symptoms showed little deterioration until eight years later, when at the age of 31 she complained of increasing breathlessness and mild orthopnoea. She had also become unsteady with difficulty walking because of stiffness of her legs, she had occasional clonus, and her writing had deteriorated. There was no parental consanguinity and no relevant family history.

On examination she was mildly breathless and she had paradoxical inward inspiratory motion of the anterior abdominal wall when supine. Otherwise there was no abnormality of the respiratory or cardiovascular system. She had mild bilateral facial weakness, volitional difficulty in keeping her eyes closed, and little palatal movement on phonation; but bilateral facial jerks were present and the gag reflex was brisk. Tendon jerks were exaggerated with extensor plantar responses and there was spasticity of all limbs with upper motor neurone weakness.

Investigations showed the following to be normal: chest radiographs; arterial blood gas tensions; cerebrospinal fluid; visual evoked responses; electromyograms of the right deltoid, triceps, and vastus medialis; single fibre electromyograms, and a quadriceps muscle biopsy specimen. The vital capacity in the sitting position was 2.65 l (81% predicted normal) and this fell by 28% to 1.9 l when the patient was supine (normal postural fall < 25%).

Oesophageal and gastric pressures were measured with balloon catheters, placed in the mid oesophagus and stomach respectively, and transdiaphragmatic pressure (Pdi) was obtained by electronic subtraction of oesophageal pressure from gastric pressure. The change in transdiaphragmatic pressure during a slow inspiration to total lung capacity was 17.5 cm H<sub>2</sub>O (normal > 25 cm H<sub>2</sub>O<sup>3</sup>), and Pdi during a maximal sniff was 29 cm H<sub>2</sub>O (normal > 70 cm H<sub>2</sub>O<sup>4</sup>),

indicating bilateral diaphragm weakness during voluntary manoeuvres. By contrast, the transdiaphragmatic pressure recorded during bilateral transcutaneous stimulation of the phrenic nerves at 1 Hz (twitch) was normal (22.8 (SD 1.8) cm H<sub>2</sub>O: normal 8-33 cm H<sub>2</sub>O<sup>5</sup>). The ratio twitch: sniff Pdi was 79% (normal 11-26%). The amplitude of diaphragm muscle action potentials measured with surface electrodes was normal (1.2 mv). Phrenic nerve conduction time<sup>6</sup> was 7 ms (normal < 9.2 ms).

Computed tomography showed generalised cerebellar atrophy. Lower limb somatosensory evoked responses were abnormal and showed absent lumbar and bilaterally delayed cortical responses.

### Discussion

Both the clinical picture and the investigatory findings in this patient were consistent with a diagnosis of spinocerebellar degeneration of familial spastic paraplegia type. In cases where there is no family history it is postulated that either inheritance is autosomal recessive or the patient has a new dominant mutation.

The clinical features of exertional dyspnoea, orthopnoea and paradoxical inward abdominal motion during inspiration are found in patients with severe diaphragmatic weakness. Diaphragm dysfunction is recognised in various neuromuscular conditions; the principal causes generally quoted in these circumstances include anterior horn cell lesions, neuropathies of the phrenic nerve, and myopathies. Both phrenic nerve function and diaphragm muscle contractility during phrenic stimulation were, however, normal in this patient. By contrast, clear evidence of prominent bulbar disease and of cerebellar atrophy were found. Accordingly it appears possible that the diaphragmatic dysfunction was secondary either to an associated upper motor neurone lesion or, more likely, to a cerebellar lesion, as this was the abnormality shown by computed tomography. Such cerebellar lesions could produce failure of coordination during voluntary diaphragmatic contractions.

While cerebellar incoordination of limb muscles is known to manifest itself as ataxia during voluntary effort, this mechanism has not previously been described for the diaphragm. Such failure of coordination might be of clinical importance, since the diaphragm is the principal muscle of inspiration<sup>7</sup> and is mainly responsible both for the tidal volume during quiet breathing and for maximal inspiratory voluntary manoeuvres. We suggest that diaphragmatic dysfunction should be considered in patients with a brainstem or cerebellar lesion who complain of breathlessness or orthopnoea.

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## References

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## Book notices

*Surgical Pathology of Diffuse Infiltrative Lung*. A Flint, TV Colby. (Pp 234; \$49.50.) Orlando: Grune and Stratton, 1987. ISBN 0-8089-1867-2.

Now that relatively safe and non-invasive methods of biopsy are available, a firm diagnosis can be made in many cases of diffuse lung disease. At the same time histopathologists are increasingly faced with a bewildering range of changes, often in tiny samples of tissue. This book deals specifically with the pathology of disorders characterised by diffuse infiltration of the lung parenchyma—disorders which are frequently investigated by transbronchial, needle, or open biopsy. In the opening chapter the general principles of processing and interpretation are covered, and there is a short but highly relevant section on the "abnormal, non-diagnostic" specimen. The second chapter is devoted to infective processes, with emphasis on viral and fungal disease, tuberculosis, and pneumocystis pneumonia. Subsequent chapters deal with pulmonary eosinophilia, hypersensitivity pneumonitis and drug induced disease, diffuse alveolar damage, interstitial pneumonitis, vascular disease, and sarcoidosis. In the section on malignant neoplasms the emphasis is on diffuse infiltrative neoplasms, including lymphomas, metastatic disease, alveolar carcinoma, and Kaposi's sarcoma. The final chapter describes a miscellany of disorders such as histiocytosis X, alveolar proteinosis, and amyloidosis. The text is well written and copiously illustrated by high quality photomicrographs. References are comprehensive and up to date. This book is not meant to be exhaustive. The authors have wisely omitted the exotic, concentrating instead on problems most commonly met with in contemporary practice. Inevitably, some topics could have been covered in more detail. Amiodarone toxicity, for instance, is dismissed in two sentences and one incomplete reference, and paraquat poisoning seems to have been forgotten. Nevertheless, this book ranks among the best of the currently available monographs on pulmonary pathology. Although primarily a bench guide for pathologists, it will also be a useful reference work for clinicians concerned with the management of chest disease—CWE.

*Manual of Chest Medicine*. JE Stark, JM Shneerson, T Higenbottom, CDR Flower. (Pp 256; £9.95.) Edinburgh: Churchill Livingstone, 1986. ISBN 0-443-02737-4.

This pocket manual has been written by a group of experienced Cambridge respiratory physicians with a radiological colleague, Dr CDR Flower. It is designed for the young doctor on a short term attachment to a respiratory unit and lays particular emphasis on dealing with practical problems. The first chapter covers a range of common respiratory symptoms and signs and is followed by a useful chapter on the radiology of the lung, including helpful comments about the place of specialist techniques such as computed tomography. A further chapter is devoted to respiratory function tests. Although simple tests are well described, the doctor working in a specialised unit might be helped by a fuller description of such "less widely available" tests as flow-volume loops and exercise testing. Subsequent chapters provide useful and often not readily obtained details of practical procedures. They deal concisely with the management of clinical problems and give useful practical hints, such as the need to alert the pathology laboratory of the arrival of samples from procedures such as transbronchial biopsy. Nearly all the clinical advice is very sound, as expected, though some would disagree with the statement that in Asian patients with suspected cervical tuberculosis aspiration or biopsy may be unnecessary. The chapter on pulmonary eosinophilia puts this disease spectrum into context with comments that the association with systematic vasculitis is very confusing, both in classification and in terminology—an observation which may take some time to dawn on the inexperienced doctor. This book will be particularly helpful to the junior doctor at senior house officer and registrar level. The senior registrar specialising in respiratory medicine may require additional detail.

## Notices

### SEPCR meeting 1988

The 23rd annual meeting of SEPCR (Societas Europaea Physiologiae Clinicae Respiratoriae) will take place in Athens, Greece, on 20-24 June 1988. The topic will be respiratory failure. There will be invited lectures, free communications, seminars, and satellite symposia. Information from Dr NM Siafakas, 23rd Annual Congress of SEPCR, Organising Secretariat, 23 Asklipiou Street, PO Box 30365, 10680 Athens, Greece.

### Clinical respiratory physiology course

A course on clinical respiratory physiology will be held at Hammersmith Hospital, London W12 0HS, on 15-18 March, 1988, for doctors and pulmonary function technicians, emphasising practical aspects and clinical applications. Details from the organisers (Drs JMB Hughes and Dr NB Pride, Department of Medicine).

## Correction

### Drill biopsy in the diagnosis of lung lesions

In the paper by Professor P Shatpathy and others (November 1987;42:858) line 3 of the second paragraph of column 2, p 858, should read "The hollow Steel's trephine."