Diaphragmatic weakness in hereditary motor and sensory neuropathy

We were interested to read a short report on diaphragmatic weakness in hereditary motor and sensory neuropathy. Whilst the case reports were highly suggestive of diaphragmatic weakness, we were disappointed that the authors relied on clinical and indirect assessments rather than on quantitative measurements of diaphragm and respiratory muscle strength. The techniques for quantifying respiratory muscle function are now well described, and can be used routinely in appropriate respiratory physiology laboratories. The measurements, which include those of maximum expiratory and inspiratory mouth pressures, of oesophageal and transdiaphragmatic pressures during maximal inspiratory manoeuvres such as an inspiratory sniff and during phrenic nerve stimulation, and of phrenic nerve conduction time, are widely available at a cost that is reasonable to both the patient and less than one hour.

Diaphragm weakness has already been reported in hereditary motor and sensory neuropathy,1 and we were surprised to find no mention of these reports in the paper by Hardie et al.2 In our paper, we reported two cases in which diaphragm dysfunction was proved and quantitated using a full range of appropriate tests. In a third report,3 a patient who had died from the condition was found at necropsy to have identical neuropathic changes in the phrenic nerves as in the other peripheral nerves.

It is important to distinguish between diffuse respiratory muscle weakness and isolated diaphragm dysfunction. Isolated diaphragm paralysis has not been shown to cause significant neuromuscular hypopontination or respiratory failure, as long as the patient does not sleep supine, or does not have other significant lung or chest wall disease.4 The majority of patients in the paper by Newsom Davies quoted by Hardie had diffuse neuromuscular disease, which would be expected to affect all the respiratory muscles as well as the diaphragm. It is interesting, however, that in the reported cases where respiratory muscle dysfunction has been quantitated, hereditary motor and sensory neuropathy appears to predominantly affect the diaphragm, perhaps because of the length of the phrenic nerves.

Only one of the patients described by Hardie et al had depression. Respiratory failure and the patient had smoked heavily. Use of discriminating tests of respiratory muscle function would elucidate whether there was involvement of the other respiratory muscles or co-existing lung disease due to smoking.


Hardie et al reply: The primary intention of our paper was to bring the possible development of diaphragmatic weakness in hereditary motor and sensory neuropathy (HMSN) to the attention of neurologists, to whom patients with these disorders are referred. We were of course aware of the other case reports on this subject, and indeed quoted the paper of Chan et al. in the report of Larche et al there were no clinical or electrodiagnostic data provided to substantiate the diagnosis of HMSN, and one of the patients had prominent urgency of micturition and defaecation, which would be most unusual. The paper of Gilchrist et al was published after our reply.

We also pointed out that involvement of the diaphragm in HMSN could be related to the length of the phrenic nerve, without proposing that diaphragmatic weakness was isolated. The relative importance of diaphragmatic involvement in our cases was illustrated by the nocturnal hyperventilation demonstrated in case 1, and the respiratory distress induced in the patients when lying supine, a position assumed by most of us at some time during sleep.

Whilst we agree that quantitative analysis of respiratory muscle weakness is important, there has to be a strong suspicion that these muscles are involved before the patient is referred to an "appropriate respiratory physiotherapy laboratory". Simple bedside tests of respiratory function, as described in our six cases, are useful in this context. Nocturnal oxygen saturation studies are also essential to exclude coexisting obstructive sleep apnoea.

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BOOK REVIEWS


This book is about 'fits' and 'faints' and not primarily about epilepsy. It discusses the many ways in which non-epileptic attacks may sound like epilepsy. It is mainly about paediatric practice although this does not detract from its overall interest. Dr Stephenson emphasises Jeavons data which suggests that 20%-30% of patients are erroneously labelled epileptic. None of us like the term 'known epileptic'; the dangers of a supposed positive family history of epilepsy are also stressed.

Most neurologists must have had doubts about their diagnosis of epilepsy in a few patients during follow-up. This may occur even after a confident initial diagnosis was made based on the patient's and witness's descriptions, cardiovascular and neurological examination, as well as EEG and ECG data. All are aware of the anoxic fit. However, my horizons about anoxic seizures have been widened as a result of reading this book. Dr Stephenson is to be congratulated on pursuing the art of history taking even further. The book is essentially clinical: it includes over 140 case histories which makes it fun for clinicians to decide what they would have diagnosed, although at times I think there may be unnecessary repetition.

Many neurologists will test for carotid hypersensitivity, especially in the elderly; few, I suspect, will use ocular compression with EEG and ECG monitoring in the assessment of anoxic seizures. This author clearly feels this is a useful test and it needs to be assessed further.

This book will be enjoyed by all neurologists who see blackouts!

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