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 diaphragm 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 at a variety of levels 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, may be used to reach a confident initial diagnosis and to monitor the patient in less than one hour.

Diaphragm weakness has already been reported in hereditary motor and sensory neuropathy, and we were surprised to find no mention of these reports in the paper by Hardie et al. In our paper we reported two cases in which diaphragm dysfunction was proved and quantified using a full range of appropriate tests. In a third report, 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 neuraxial hypoposition or respiratory failure, as long as the patient does not sleep supine, or does not have other significant lung or chest wall disease. The majority of patients in the paper by Newsom Davis 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 quantified, 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 evidence of respiratory failure and she had smoked heavily. Use of discriminating tests of respiratory muscle function would have elucidated whether there was involvement of other respiratory muscles or co-existing lung disease due to smoking.

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