MEDICAL MEMORANDA

Dystrophia Myotonica Presenting with Dysphagia

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Dysphagia is an uncommon manifestation of dystrophia myotonica and has been considered a terminal symptom of the disease (Caughy and Myrianthopoulos, 1963). We report the case of a patient presenting with dysphagia in whom electromyographic sampling of the pharyngeal musculature confirmed a diagnosis of dystrophia myotonica, and in whom dysphagia improved on treatment with procainamide.

Case History

A 35-year-old housewife was admitted to hospital in May 1968 complaining of difficulty in swallowing, weakness of limbs, and loss of weight. Eighteen months earlier she began to cough and splutter during meals; food seemed "to lodge above the breast bone," and regurgitation often occurred. As her dysphagia progressed she developed weakness of the legs and noticed that they

FIG. 1—Barium swallow. Contrast material seen in the larynx and trachea. A = Vallecula. B = Pyriform fossa. C = Barium in ventricle of larynx. D = Vocal cords. E = Oesophagus. F = Trachea. FIG. 2—Barium swallow. Normal appearance. (On treatment with procainamide.)

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were becoming thinner. Weakness of the hands occurred about six months later. She had lost 3 stone (19 kg) over the previous 18 months owing to inadequate food intake secondary to her dysphagia.

On examination she had bilateral early posterior cortical cataracts and a myopathic facies. Her speech was dysarthric with a nasal quality. Weakness of the muscles of mastication and weakness and wasting of the sternomastoids and trapezii were present. The gag reflex was intact. Percussion myotonia was present in the tongue. She had difficulty in swallowing water, and accomplished this by maximally extending the neck and "tossing the fluid back." Muscles in the upper and lower limb girdles, hands, and legs showed weakness and wasting. Myotonia was pronounced, and she had difficulty in relaxing hand grip. There were no sensory or reflex changes.

Routine blood investigations were within normal limits, and so were x-ray pictures of the skull and chest. Serum creatine phosphokinase level was raised at 87 IU/l. (normal range 0-40 IU/l.). Lung function tests showed reduction of the maximum breathing capacity to one-third of the predicted value, and there was widening of the QRS complex on E.C.G. A barium cine swallow showed incoordination of swallowing and no appreciable rise of the larynx; the pharynx was not cleared each time, and contrast passed into the larynx and trachea (Fig. 1). These appearances were not altered after the intravenous administration of edrophonium 10 mg.

Electromyographic examination of the right superior or middle constrictor muscles of the pharynx was undertaken. A concentric needle electrode was inserted percutaneously at the upper posterior border of the hyoid bone while the sternomastoid muscle and underlying vessels were retracted laterally to reduce the risk of damage to adjacent structures. The site of sampling was identified by the presence of motor unit activity on deglutition. In two areas a spontaneous repetitive discharge of motor units had a high initial frequency which declined rapidly and thereby produced a "dive-bomber" sound in the electromyograph loudspeaker. On sampling skeletal muscle groups a full interference pattern was found, but motor units were reduced in amplitude and were of short duration; these findings were suggestive of a myopathy. A myotonic "dive-bomber" phenomenon was also present.

A diagnosis of dystrophia myotonica was made and she was given procainamide 500 mg thrice daily. The myotonia and dysphagia improved, and two years later there was no evidence of myotonia on clinical examination, though she complained of some difficulty in releasing hand grip in the morning. A repeat barium cine swallow showed no abnormality. (Fig. 2). The patient refused further electromyographic studies.

Comment

This case is of interest because the dysphagia, which was one of the patient's presenting complaints, was reversible with procainamide and was shown to be associated with myotonia of the pharyngeal musculature. Electromyographic sampling of pharyngeal muscles may be valuable in establishing a cause of dysphagia in other organic diseases if the radiological findings are not diagnostic; it is also helpful in determining whether the difficulty in swallowing in dystrophia myotonica is due to myotonia or myopathy.

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Reference

Caughy, J. E., and Myrianthopoulos, N. C. (1963). *Dystrophia Myotonica and Related Disorders*, Springfield, Illinois, Thomas, p.29.