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Myotonic Dystrophy and Traumatic Quadriplegia: Case Report

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Summary

A thirty-year-old man with traumatic quadriplegia, was also found to have weakness above the level of the injury. He had facial weakness, difficulty in swallowing, and recurrent respiratory problems. A diagnosis of myotonic dystrophy was supported by examination of his sister. The problems of diagnosis, and the implications of the diagnosis on the management of the patient with myotonic dystrophy and a spinal injury are discussed.
Key words: *Myotonia atrophica; Quadriplegia; Spinal injuries.*

Myotonic dystrophy is a myopathy, but also a systemic disorder, with a prevalence of around 5 per 100 000. It is inherited as an autosomal dominant, with variable manifestations within a family. The genetic abnormality has been localised on chromosome 19, and new mutations are extremely rare. The condition usually first becomes apparent between the ages of 20 and 50 years-of-age.

Myotonia is the delayed relaxation of a muscle or group of muscles after voluntary contraction or stimulation, manifesting typically as difficulty in relaxation of grip. The commonest presentation is with muscular weakness, especially distal, causing weakness of the hands, difficulty walking, and frequent falls. The facial appearance is characteristic with frontal balding, ptosis, and weakness and wasting of the facial muscles and sternomastoids (Harper, 1988, 1989).

There are special problems in managing quadriplegia from spinal cord injury (SCI) in patients with a muscular dystrophy, although rarely reported (Bloom and Freed, 1989). We describe a case of myotonic dystrophy, with special regard to its presentation, diagnosis and management, in association with SCI.

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Case History

An apparently healthy thirty-year-old man, working as a window cleaner, fell twelve feet from a ladder, and sustained a fracture dislocation of the cervical vertebrae C6 and C7. He was admitted to the Spinal Injuries Unit, where he was found to have a complete motor and sensory deficit below the C6 level. He was treated with cervical traction and halo brace immobilisation (Parry *et al.*, 1988).

On the second day, despite physiotherapy, his respiration deteriorated with right upper lobe collapse necessitating intubation and assisted ventilation. By 7 days he was able to breathe spontaneously, but at 10 days required tracheostomy and ventilation to correct hypoxia and poor respiratory effort. His respiratory difficulties were more marked during sleep. After 13 days he was again removed from the ventilator, but had an episode of apnoea with bradyarrhythmia, and ventilation was recommenced.

On the twentieth day he was breathing spontaneously and able to return to the ward. He had difficulty swallowing saliva, was fed by a nasogastric tube, and regurgitated the feeds. There was no abnormality of the oropharynx on clinical examination, but on barium swallow the orophayngeal movements were insufficient to propel the bolus into the oropharynx and upper oesophagus, and the majority passed into the larynx and trachea.

After 65 days he was still having difficulty in swallowing, with recurrent respiratory problems. He was noted to have some weakness of eye closure, and a clinical appearance suggestive of myotonic dystrophy. He remained paralysed below C6, with no clinical myotonia in the hands or tongue. Electrophysiological investigation demonstrated features of a motor neuropathy of demyelinating type in the arms and legs, with no activity at rest in the facial muscles, marked reduction in the interference pattern, but no myotonic discharges. Nerve biopsy of the superficial peroneal nerve showed axonal degeneration, with secondary myelin breakdown.

At 80 days the halo brace was removed, but his clinical condition remained unchanged. Previous enquiry had not revealed any family history of a neuromuscular disorder, but at 120 days his wife mentioned a possible facial weakness in his sister and her daughter. On interviewing his sister, aged 24 years, she mentioned that she had had difficulty in swallowing for many years, and had to bend her neck forwards to swallow bread. She had been told that she slept with the white of her eyes showing. On examination she was generally thin, with severe wasting of the temporalis muscle, and typical appearance of myotonic dystrophy. She had myotonia of grip and on thenar percussion. Electromyography demonstrated the typical 'dive bomber' discharge of myotonic dystrophy in the extensor digitorum communis muscle. (She had recently been investigated for weight loss, which had been attributed to an inadequate diet).

The positive electrophysiological diagnosis of myotonic dystrophy in his sister, confirmed the clinical suspicion of myotonic dystrophy in our patient, and the implications were explained to the family. At 140 days he had recurrent apnoeic episodes, with excessive secretions, and in view of the poor long term prognosis, it was decided not to recommence assisted ventilation. He died peacefully at 150 days after his injury.

Discussion

This case, of a patient with SCI quadriplegia, subsequently diagnosed as having myotonic dystrophy, illustrates several points relating to the problems of diagnosis and management, and the importance of examining other members of the family.

It is possible that muscular weakness or myotonia contributed to the initial fall and injury. Most patients with myotonic dystrophy have myotonia which can be detected on electromyography. It is difficult to say if the SCI affected any myotonic response in the muscles of the limbs, but this seems unlikely as the myotonia is thought to be primarily a muscular phenomenon. His features of facial nerve weakness and bulbar involvement, without myotonic features on electromyography, are those of a 'partial syndrome' reported in families with myotonic

dystrophy (Pryse-Phillips *et al.*, 1982). Electrophysiological evidence of peripheral nerve involvement has been described in myotonic dystrophy, and is thought to be independent of the muscular abnormality (Panayiotopoulos and Scarpalezos, 1976).

Dysphagia is often a presenting feature of myotonic dystrophy. Aspiration is common and causes recurrent pneumonia, especially of the right lung. Weakness and myotonia of the striated muscle of the pharynx and upper oesophagus, and disordered peristalsis with involvement of the smooth muscle of the lower oesophagus occur (Goldberg *et al.*, 1972). The dysphagia may be influenced by neck position (Bosma and Brodie, 1969). This was a specific symptom of his sister, and for the patient with a neck injury requiring traction, the neck movement is abolished and the dysphagia exacerbated.

Respiratory problems are common in the later stages of myotonic dystrophy, and death is usually due to respiratory infection or cardiac failure. Recurrent aspiration contributes to the problem, but other components of the respiratory failure are diaphragmatic muscle weakness or myotonia, chest wall muscle weakness or myotonia, and decreased central neurogenic drive (Carroll *et al.*, 1977). The patient with a cervical cord injury of C6 would normally rely on diaphragmatic and accessory muscles of respiration, but in myotonic dystrophy both these groups of muscle may be affected. There is a reduced hypoxic drive suggesting involvement of brainstem neuroregulatory mechanisms, and causing difficulties with maintaining adequate ventilation especially during sleep.

There is no specific treatment which will alter the course of myotonic dystrophy, although several drugs have limited symptomatic effect on the myotonia. For the patient with myotonic dystrophy and cervical cord injury, complications of recurrent chest infections, respiratory failure, and difficulty swallowing may be anticipated and minimised by careful nursing, physiotherapy, dietary advice, and medical treatment. Additional attention must be given to the possibility of cardiac arrhythmias, and caution in the use of anaesthetic agents, above the problems otherwise associated with autonomic dysregulation in quadriplegia. For the patient who recovers from the injury, rehabilitation may be more prolonged and require additional aids. For those with weakness of the neck muscles a cervical collar, and head rest for the car and chair, may reduce the chance of recurrent cervical cord injury.

In retrospect the persistent problems of difficulty swallowing and respiratory insufficiency, may have pointed at an earlier stage to the possibility of myotonic dystrophy. The rate of progression of the disease is variable, usually over decades, but is sometimes more rapid. As a result of the difficulties of clinical examination in the presence of a traumatic quadriplegia, and the absence of myotonia on electromyography, only the personal examination of the relatives may lead to the diagnosis, as the symptoms are often not noticed, or accepted as a minor inconvenience by those affected. Having made the diagnosis in the family, genetic counselling should be offered. As an autosomal dominant condition, there is a 50% chance of a child being affected, and there is the possibility of anticipation, where the condition occurs earlier and is more severe in successive generations. The children of affected women are at risk of a severe congenital myotonic dystrophy. Subclinical cases may be detected by investigation. There are increasing opportunities for prenatal diagnosis, and if linkage studies are appropriate, it is

important to have DNA from affected members of the family. Where death is imminent, as in the case described, the blood could be taken and stored for future use.

Although myotonic dystrophy is rare, patients are at risk of falls, may have weak neck musculature, and are probably at higher risk of cervical cord injury. The possibility should be considered in patients with recurrent aspiration, respiratory problems, and weakness above the level of the spinal cord injury. Other members of the family should be offered genetic counselling.

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