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Myotonia Dystrophica in a Victim of Traumatic Brain Injury — A Case Report

Jeng-Yi Shieh, Chein-Wei Chang, I-Nan Lien

Myotonia dystrophica is one of the common adult-onset muscular dystrophies. It is characterized by its multisystemic involvement as compared with other myotonic disorders. We report here a 33-year-old man with myotonia dystrophica who was diagnosed after transfer to the rehabilitation department for the sequelae of traumatic brain injury, The clinical, electrophysiological and histopathological findings supported the existence of myotonia dystrophica. The presentation of myotonia did not change in association with his central paralysis.

Key words: myotonia dystrophica, traumatic brain injury

INTRODUCTION

Myotonia is defined as a delayed relaxation of a muscle after either voluntary contraction or stimulation. Myotonia dystrophica, one of the myotonic disorders, was well described by Dr. Steinert with its clinical course and manifestations in 1909 [1]. This disease is characterized and differentiated from other myotonic disorders by multisystemic involvement. Furthermore, the usual presentation includes distal and hand muscular weakness, walking difficulty, and frequent fall. In addition, the facial appearance is characteristic with frontal balding, ptosis, and weakness and wasting of the facial and neck muscles. It is thought to be a common adult-onset muscular dystrophy.

Electromyographic examination may present a typical feature of myotonic discharge with "diving-bomber" phenomenon. Histopathological findings of muscle, which can provide a good diagnostic feature, are internal nuclei, sarcolemmal masses, ring fibers and atrophy in type I muscle fibers.

We report here a case of myotonia dystrophica who met with a traumatic brain injury associated with hemiparesis. The change of muscle tone after head injury was discussed.

CASE REPORT

A 33-year-old man was brought to our emergency section with a progression of conscious disturbance on April 3, 1991. His brother stated that he met with a motorcycle accident about two hours ago. Emergent computerized tomography of the head was performed which revealed a well-defined, convex-shaped hyperdensity lesion over right frontal, temporal and parietal areas with shifting of the midline structures. Under the impression of epidural hematoma, emergent operation was performed and found an epidural hematoma over his right

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fronto-temporo-parietal areas. About 80cc blood was evacuated. After operation, his condition improved but neurological manifestation with left hemiplegia presented. Profuse sputum and poor oxygen saturation were observed frequently during the post-operative days. He had weak cough and was suffered from pneumonia once. On May 13, 1991, he was transferred to rehabilitation ward for ambulation training and other rehabilitation program.

Physical examination showed that he had a slightlypoor-nourished and weak stature. He had frontal baldness. Mild ptosis was observed (Fig. 1). His right leg appeared apparently thinner than the hemiplegic leg. During the neurological examination, short attention span was observed. His speech was dysarthric and fast in rhythm. The mentality was impaired. Cranial nerve examination revealed mild left central type facial palsy and deviation of tongue toward left in protruding. He could not raise his left arm but could achieve weak finger and leg movement. No spasticity was detected. The deep tendon reflex (DTR) was normal over left side and slightly decreased over right side. Needle electromyography (EMG) study showed a typical myotonic discharge over all examined muscles including right biceps brachii muscle, right first dorsal interosseous muscle, right tibialis anterior muscle and left first dorsal interosseous muscle.

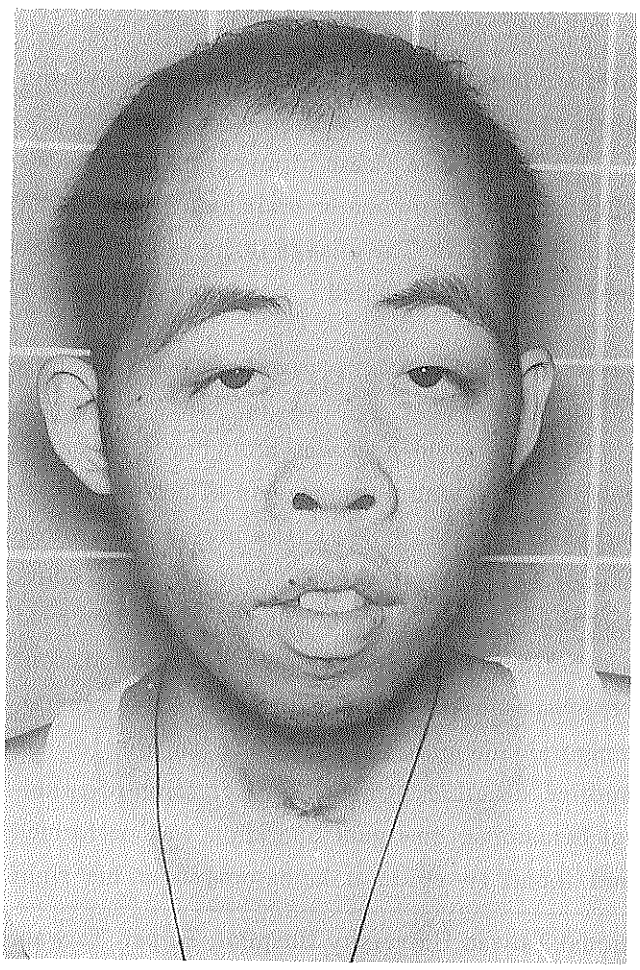

Fig. 1. The facial appearance of the patient. Long face, alopecia, ptosis, parted lips, and temporalis atrophy.

Subsequent clinical evaluation revealed that he had weak facial muscles and weak neck flexors. The temporalis muscle was weak and atrophic. His mouth was kept open at all times and looked like an inverted "V" (Fig. 1). The right lower leg was markedly atrophic. Ophthalmological examination revealed no cataract. Audiometric examination revealed moderate sensorineural hearing loss over right ear and mixed type hearing loss over left ear. Electrocardiography (EKG) showed normal sinus rhythm with occasional premature supraventricular complexes with abberation, left axis deviation with left anterior hemiblock and non-specific ST-T changes. Blood chemistry showed normal GOT and LDH but slightly elevated creatine phosphokinase (CPK) up to 180 IU/L, mainly of MM type (100%). Serum protein electrophoresis revealed low albumin (3.3 g/dl), low gamma-globulin (0.46 g/dl) and decreased immunoglobulin G (IgG 0.6 g/dl). Examinations of his endocrine functions showed normal glucose tolerance, normal thyroid function but decreased testosterone level (1.66 ng/ml) with elevated FSH (24.8 mIu/ml). The plain skull X-ray showed sclerotic change of the cranial bones. The pituitary fossa was unusually small (6 mm in diameter). Muscle biopsy taken from his left quadriceps femoris muscle showed prominent internal nuclei (Fig. 2) and necrosis of some muscle cells with a mild increased

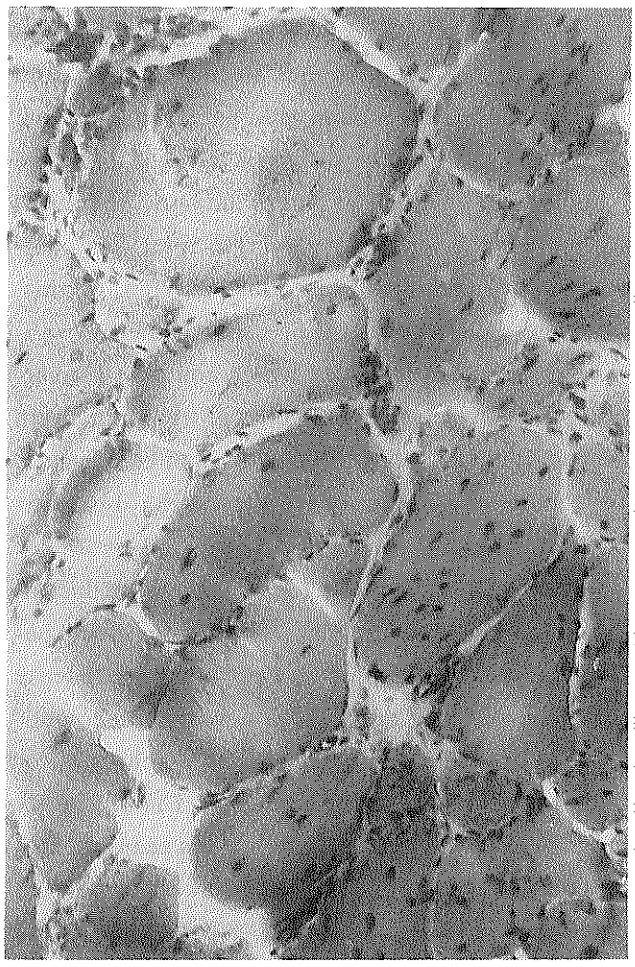

Fig. 2. The findings of muscle biopsy. Notice the internal nuclei.

mononuclear cells infiltration. There were ring fibers and sarcolemmal masses. All of the above findings are compatible with the diagnosis of myotonia dystrophica.

This patient underwent physical and occupational therapies in our ward. Aleviatin (10 mg, twice daily) was prescribed after the diagnosis was sustained. The main problems in his rehabilitation were the presence of short attention span, impaired cognitive function including left hemineglect, poor motor control of left upper extremity, poor physical endurance and poor muscle power. The ability to be aware of environmental dangers was also impaired. Psychological evaluation revealed that his verbal IQ and performance IQ were 76 and 55 respectively. He was discharged on July 15, 1991. By the time of discharge, he could walk independently without assistive device and perform most of the activities of daily living under supervision. As for myotonia, no apparent improvement could be detected either clinically or electrophysiologically.

DISCUSSION

This patient showed the following typical presentations of myotonia dystrophica: facial appearance, limb atrophy, abnormal electrophysiological recording and pathological findings. Moreover, a lot of associated anomalies in myotonia dystrophica have been well-documented. Wright et al. found 17 of 25 patients with myotonia dystrophica had moderate to severe hearing loss which was usually the sensorineural type. Therefore, they suggested that audiometric screening should be a routine in evaluating patients with myotonia dystrophica [2]. Abnormal EKG is common and represents myocardial involvement. The most common finding in ambulatory EKG is first degree A-V block and this abnomality can even occur in the early stage of the disease [3]. In addition, cataract formation is of diagnostic significance between the age of 10 to 60. The initial and most specific abnomality consists of numerous small, closely packed white and multicolor opacities particularly of the posterior cortex of the lens while the nucleus of the lens remains free [4]. Our patient had hearing loss and arrhythmia but had no cataract. The patient's presentation might have been a variant but we should include ophthalmological examination during his follow-up care. Endocrine and metabolic disturbances are frequently encountered such as hypogonadism and abnormal glucose tolerance. Although the sexual hormone profile was abnormal, our patient did not show apparent testicular atrophy which has been mentioned in other studies [1].

There is also much evidence for the widespread involvement of myotonia dystrophica. Affected oropharyngeal musculature results in defective speech,

dysarthria and nasality of speech. Some patients may have an explosive speech which is barely intelligible, particularly when they speak fast [5]. This picture was shown in our patient. Besides, Hannon and associate ever reported a case with post-anesthetic aspiration pneumonia and was then confirmed as a case of myotonia dystrophica after clinical investigation [6]. Such cases were sensitive to various anesthetic drugs, including depolarizing muscle relaxants, barbiturates, opioids, benzodiazepines and volatile agents [7]. Excessive respiratory depression is due to an additive effect to the already diminished respiratory and cardiac reserve in these patients. This might explain why our patient appeared cyanotic with profuse sputum at surgical ICU frequently. Some authors stated that abnormal uterine contraction resulting in abnormal labor progress could be found in pregnant patient with myotonia dystrophica while others did not agree [8].

Our patient also had the specific features in radiographic and laboratory aspects. The skull often appears hyperostotic with large sinuses and a small sella turcica. Enlargement of the ventricles was also reported [1]. Skeletal deformities such as scoliosis and deformed foot were ever mentioned [9,10]. Computerized tomography may reveal a decrease in muscle volume, irregular hypodensity of the muscles, especially in the distal muscles, and spinal muscle damage [11]. The laboratory findings often include normal or slightly elevated CPK, hypogammaglobulinemia, increased catabolism of IgG, and abnomalities of blood cell about their membrane function.

In our patient, right lower leg atrophy and easy fatigue were the only findings that his family could recall regarding his disease. He withdrew from the primary school and became a laborer because of poor intellectual performance. This could be expected because mental defect is a usual finding. He lived with his father and was quite independent of the activities of daily living. Traumatic brain injury can result in many sequelae including motor and sensory deficits as well as impaired cortical function. The main problems in his rehabilitation performance included poor endurance and abnormal cognitive function. The former is thought to be due to underlying myotonia, whereas the latter is thought to be a sequela of traumatic brain injury as compared with his premorbid status. In view of this, we suggested that he must be accompanied by others and avoid the activities which could be dangerous. The recovery from sequelae of traumatic brain injury might be lasting even after months or years. But recovery from underlying myotonia dystrophica seemed dim.

The change of muscle tone in myotonia dystrophica after brain injury was never mentioned before. Although myotonia dystrophica is a disease primarily affecting the muscle, neuropathic component was ever reported [1]. DTR in myotonia dystrophica was stated to be diminished. The spasticity in our patient was not as prominent as seen in usual victims of head injury. He appeared generalized hypotonic with decreased DTR, especially over the sound side. We presumed that the discrepancy of DTR between bilateral limbs was due to traumatic brain injury which caused disinhibition over affected side, while the DTR was not totally absent.

Treatment in the myotonic disorders remains controversial. Drugs of proven effectiveness are procainamide, quinine, phenytoin, mexiletine and tocainide. Imipramine, prednisone, acetazolamide, verapamil and nifedipine were also ever reported [12]. Phenytoin was thought to stabilize the excitable membrane because of its ability to block sodium influx, calcium influx or both [13]. Lithium was also shown effective because it may affect the kinetics of the sodium channel in skeletal muscle [14]. However, the treatment of myotonia dystrophica is not as effective as that of myotonia congenita. Because there is no convincing evidence that amelioration of the myotonia influences the dystrophic features. For now, treatment of myotonia should consist of advice about keeping warm and a

class I antiarrhythmic agent such as phenytoin or procainamide in the first instance [12].

The patient's mother died at the age of forties without definite diagnosis. As his family stated, she also had frontal baldness, parted lips and hallow cheeks. The patient has three siblings. All of them did not show any significant finding after clinical examination. We have obtained their blood for chromosome study. After these data are available, genetic counselling should be given to his siblings.

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萎縮性肌強直症合併頭部外傷—病例報告

謝正宜　張權維　連倚南

肌強直是一種肌肉持續收縮的狀態，而萎縮性肌強直是肌強直疾病中較常見的一種。它在1909年首次由Steinert加以詳述，所以又稱為Steinert氏病。它的特色是除了肌強直外，有全身各系統的症狀。包括肌肉萎縮，禿頭，眼瞼下垂，白內障，聽力障礙，心律不整，性腺萎縮，代謝障礙及智障等。

本篇病例報告描述一個33歲因頭部外傷而轉入復健病房的男性萎縮性肌強直症的病人。鄰床上有禿頭，眼瞼下垂，咬肌萎縮，右小腿肌肉萎縮，聽力障礙，心律不整，性腺萎縮及智障。肌電圖檢查呈現典型肌強直連續肌電波，類似轟炸機俯衝的聲音。肌肉病理組織學撿查發現肌細胞內有明顯內核(internal nuclei)，漿團(sarcolemmal mass)，及環狀纖維(ring fiber)。病人因腦傷雖導致右側肢體麻痺，但無明顯肌痙攣性(spasticity)，其原肌強直性也無明顯改變。

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