J. Neurol. 219, 133—137 (1978)

Journal of
Neurology

Short Communications

Myotonic Myopathy with Painful Muscle Contractions and Decrease of Symptoms by Cold

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Summary. Myotonic reaction and paresis accompanied by painful muscle contractions limited to the upper limbs, which decrease remarkably in the cold, were observed in a 29 year old man. The histological investigation revealed minimal non-specific signs of myopathy. The biochemical studies of muscular tissue contained a normal amount of myophosphorylase, acid maltase and glycogen. Ischemic work induced normal elevation of venous lactate. The activities of CPK, LDH and SGOT in the blood serum were occasionally increased. The EMG showed typical myotonic bursts and electrical silence during painful muscle contractions. Repetitive high frequency stimulation demonstrated a clear initial increase of the amplitude of action potentials followed by a decrease in the contracted muscle. The father of the patient suffered from dystrophia myotonica. This coincidence suggests that this myotonic myopathy is a variant of dystrophia myotonica.

Key words: Myotonic myopathy – Muscle contractions painful – EMG silent – Nerve stimulation – Dystrophia myotonica.

Zusammenfassung. Myotone Reaktionen, Paresen und schmerzhafte Muskelkontraktionen werden bei einem 29jährigen Patienten beobachtet. Die Symptome beschränken sich auf die oberen Extremitäten und sind in Kälte deutlich gebessert. Die Histologie zeigt uncharakteristische diskrete myopathische Zeichen. Der Gehalt an Phosphorylase, saurer Maltase und Glykogen im Muskel ist normal. Das EMG sichert die Myotonie. Die schmerzhaften Kontraktionen sind elektrisch stumm. Der Ischämie-Arbeitstest führt zu einem prompten Laktatanstieg. Die Serumenzyme CPK, SGPT und LDH sind intermittierend erhöht. Bei Stimulation mit hohen Reizfrequenzen wird ein deutlicher Anstieg der Summenpotentialamplitude zu Beginn, ein ausgeprägter Abfall der Amplitude im verkrampften Muskel beobachtet. Der Vater des vorgestellten Patienten ist an einer Dystrophia myotonica erkrankt gewesen. Dieses Zusammentreffen läßt in der beschriebenen myotonen Myopathie eine Variante einer Dystrophia myotonica vermuten.

0340-5354/78/0219/0133/$ 01.00

Case Report

A 29 year old man dated the onset of his myotonic muscular disfunction to his fourteenth year of life. Painful muscle contractions first appeared after severe manual work at the age of seventeen. There was marked increase in frequency and severity of muscle symptoms after this initial event. The painful muscle contractions were in the forearms and hands. The contractions appeared within a few minutes of severe manual work and receded within 10—30 min. Weakness subsided in 5—6 h. Occasionally myotonia involved the tongue, but was never seen in the face or jaw muscles.

An increase of painful muscle contractions was related to warm temperature, while a marked decrease of myotonia, weakness and painful contractions was observed in the cold. The patient was not handicaped during skiing and swimming in cold water.

Dystrophia myotonica was observed in the father of the patient. His disorder was worse in the cold but never was accompanied by painful muscle contractions. There was no further evidence in the history suggesting a muscle disorder in other members of the family.

Examinations

The neurological examination was normal except for active myotonia in the forearm and hand muscles. There were no dystrophic signs at all. Repeated muscle exertion of the hand brought about a painful contraction of the forehand flexor and intrinsic hand muscles (Fig. 1). Repeated forced manual pressure against a constant amount of resistance (frequency 28/min, skin surface temperature 34°C, cuff of a sphygmomanometer) evoked paresis after 14 s and painful muscle contractions after 17 s. The next pressure of the cuff was practicable after a 90 s rest. Using the same amount of exertion after exposure to cold—the hand and forearm having been placed in water up to the elbow of 10°C for 4 min (skin

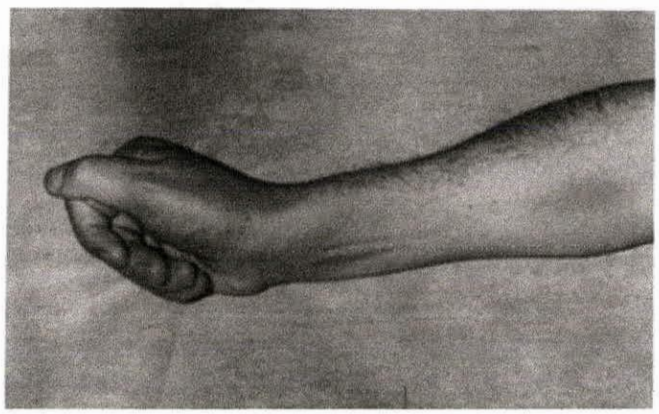

Fig. 1. Painful contraction of forearm and intrinsic hand muscles after exercise. The position of flexion not removable by voluntary effort

surface temperature 21°C)—the onset of paresis occurred after 30 s and of painful contractions after 35 s. The next manual pressure was practicable after a 40 s rest.

An exercise test under ischemic conditions induced normal elevation of venous lactate. Histochemical staining of a biopsy taken from the right biceps brachii muscle revealed normal amounts of glycogen, acid maltase and myophosphorylase. The histological investigation revealed minimal non-specific signs of myopathy.

The general physical examination was normal except for low blood pressure. There were no clinical signs of endocrine disturbance, especially testicular atrophy. Hearing was normal. The ophthalmological examination revealed cataracta punctata. X-ray studies of the abdominal viscera disclosed an incomplete mesenterioaxial volvolus. The ECG was marked by a prolonged P-Q interval. Serum enzyme levels were occasionally elevated (SGOT 27 U/l, CPK 134 mV/ml, LDH 288 mV/ml). Total serum protein was 5.9 g% with diminished gamma fraction 10.3%. ACTH load induced no elevation of 17-hydroxy- and 17-ketosteroids.

Electromyographic studies of the flexor digitorum superficialis and profundus, biceps brachii and opponens pollicis muscles were performed with a con-

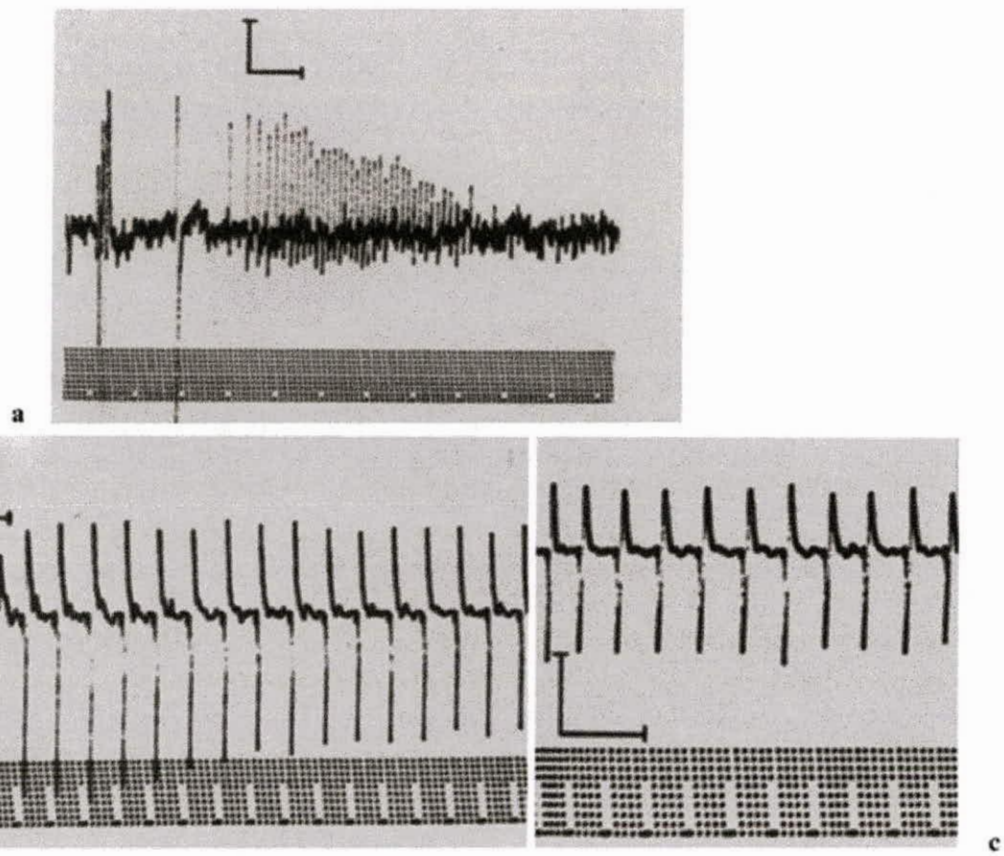

Fig. 2. **a** Myotonic discharge after needle movement in right opponens pollicis muscle (200 μV/Div.; 100 ms/Div.). **b** Repetitive supramaximal stimuli applied to right median nerve at elbow, frequency 20/s; action potentials registered from flexor digitorum superficialis. Initial increase of amplitude and beginning decline within the first second (2 mV/Div.; 100 ms/Div.). **c** After 15 s of stimulation clear decline of amplitude in contracted flexor digitorum superficialis muscle (2 mV/Div.; 100 ms/Div.; medelec EMG)

centric needle electrode. Myotonic discharges were seen in all sites and consisted of bursts of repetitive discharges following needle insertion or movement (Fig. 2a), voluntary contraction or muscle percussion. The discharges waxed and waned in amplitude (divebomber noise) and lasted 3 s on the average. After exposure to cold a decrease in frequency and duration of myotonic bursts was observed. Voluntary muscle action potentials were of normal wave form and duration and there was maximum recruitment. The onset of paresis was accompanied by a mixed interference pattern, finally with reduction to single action potentials. The EMG was silent during painful muscle contractions. During the remission from paresis a few fibrillations were observed.

Electroneurographic studies revealed normal motor nerve conduction velocities of median and ulnar nerve. The evoked potentials were normal. The myoneural junction was examined according to Stöhr et al. [6]. Repetitive supramaximal stimuli of various frequencies and 2 s duration were applied to the median nerve at the elbow and wrist. The muscle action potentials were registered with surface electrodes from the flexor digitorum superficialis and abductor pollicis brevis muscles. No change of the amplitude of the action potential was seen during stimulation with frequencies of 5 and 10/s. With continuous stimulation for 20/s frequency an increase of amplitude of 57% was observed within 0.3 s. After the first second the amplitude of the action potentials still showed an increase of 12% (Fig. 2b). After 15 s of stimulation a clear decline of amplitude (28%) was seen in the now painfully contracted muscles (Fig. 2c).

Discussion

The electromyographic and electroneurographic findings in our patient corresponded on the whole to the results of Stöhr et al. [6]. They have reported this findings in a patient of a family having true myotonia with painful, electrically silent muscle contractions following exercise. The nature of the underlying metabolic defect was unknown. The most remarkable difference between the case of Stöhr and our case was decrease of myotonia, delay of the development of painful muscle contractions and the subsidence of weakness after exposure to cold.

The myoneural junction was examined in patients with myotonia congenita, dystrophia myotonica [1] and with paramyotonia congenita [3]. The results of high frequency stimulation differed from our patient in any case. Beside electroneurographic investigations the clinical picture of our patient was incomparable to paramyotonia congenita, myotonia congenita and to some extent to dystrophia myotonica [2].

Except for the electrically silent muscles during painful contractions and for the delay of the development of painful contractions and paresis by cooling [5] no other symptoms of Mc Ardle disease [4] were observed.

The coincidence of dystrophia myotonica in the father of our patient and the myopathy described above suggests that this myotonic myopathy is a variant of dystrophia myotonica, in which symptoms are decreased by cold and dystrophic

signs are not marked [2]. This hypothesis is supported by findings of other clinical manifestations apart from muscle lesions, such as cataracta punctata, abnormalities of the gastrointestinal tract and partial endocrine disturbances.

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Received May 24, 1978