A Case of Myotonic Dystrophy with Psychic Problems

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INTRODUCTION

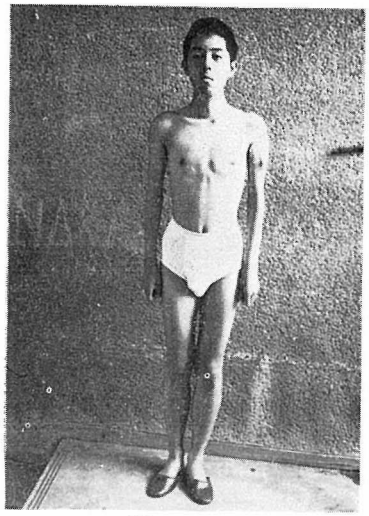
The clinical and histopathological features of myotonic dystrophy have already been well described by many authors[1-4].

Myotonic dystrophy is assumed to be an inherited disease which is transmitted by an autosomal dominant gene. It is characterized by myotonic symptoms, muscle weakness, muscle wasting of the face and neck, cataracts, early baldness and testicular atrophy, in association with other endocrine disorders[5]. The main pathological features are hypertrophy and fragmentation of the muscle fibers which, in the final stages of the disease, are followed by muscular atrophy and its replacement by fat and connective tissue[6].

This paper presents the case report and the findings obtained by muscle biopsy of a patient who exhibited myotonic dystrophy in association with unusual psychiatric symptoms.

CASE REPORT

The patient, T.Y., a male, was 18 years old when he first came under our observation in May of 1974. History taking revealed that one month earlier, after graduation from high school, he had entered the Air Branch of the Japanese Self Defense Forces. Shortly thereafter, in May of 1974, he had begun to suffer from sleeplessness and unusual experiences during the night. These strange experiences were characterized by optic hallucinations that occurred within a short period of time after awakening in the middle of the night. The hallucinations, which were associated with a sensation of fear, consisted of the faces of known frie-



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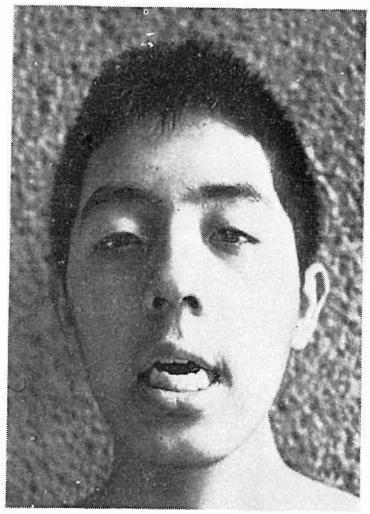

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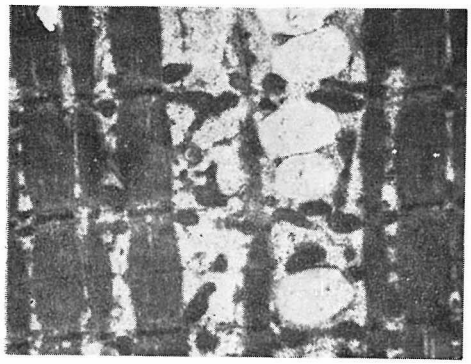

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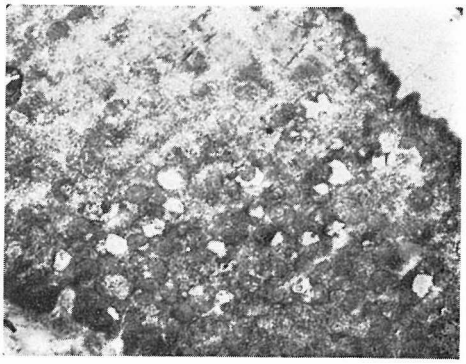

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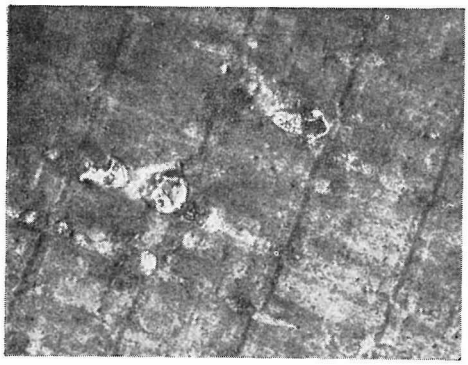

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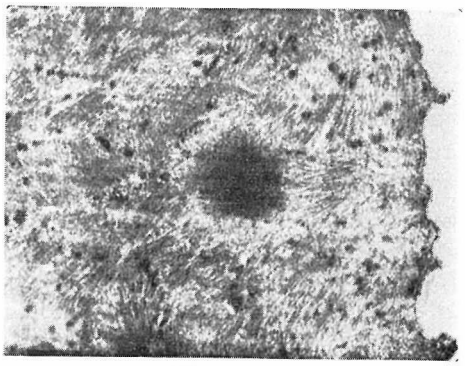

Photo. 6.

Photo. 1. The patient shows relative muscular atrophy of the upper extremities.

Photo. 2. Myopathic face with open bite.

Photo. 3. Electron micrograph showing lipid droplet accumulation in the muscle. Gastrocunaemius muscle.

Photo. 4. Intramyofibrillar vacuole formation with myelin like figure. Gastrocunaemius muscle.

Photo. 5. Subsarcolemmal accumulation of mitochondria, glycogen granules and vacuoles. Gastrocunaemius muscle.

Photo. 6. Subsarcolemmal cytoplasmic body. Gastrocunaemius muscle.

nds that could be seen vividly even in complete darkness. These hallucinations usually lasted several minutes to half an hour. Though earlier he had experienced a weakness in his hands at age 16 after slightly injuring both wrists by fighting. He visited our clinic for treatment of his sleeplessness and his hallucinations and not because of any muscle wasting or weakness.

Past history: One febrile convulsion was noted at 5 years of age. Otherwise prior to his visit to the clinic, he had suffered from no acute illness and had been healthy.

Family history: The patient's father died of accidental causes at the age of 46. He was said to have been in excellent physical condition and showed no signs of baldness or muscle wasting. The mother is still alive and is physically healthy and well. The sister, aged 13, is also well. None of the patient's relatives showed any sign of myotonia or muscle weakeness.

Present illness: The patient was a slender man. His face was dull and expressionless (Photo. 1). The eyes looked sleepy and the mouth always hung slightly open. Ocular movement was unremarkable. No trace of cataracts were seen. His jaw was long and thin. He had a moderate degree of open bite (Photo. 2). The trait of baldness was not yet seen. Other than some weakness of the orbiculares palpebrarum and orbicularis oris muscles, no remarkable muscle wasting or weakness was seen except in the muscles of the forearms and hands. The thenar and hypothenar muscles were slightly atrophied. The gait was normal. If the patient was asked to make grasping movement repeatedly, he usually experienced difficulty in opening the palm and stretching the fingers; in particular, the thumb stretched only jerkily and with effort. The grasping power was 6 kilograms on the right and 7 on the left. No marked decrease of the grasping power was noted after hand exercises were performed. Deep tendon reflexes were all hypoactive and no pathological reflexes were elicited. The jaw articulation seemed to be slightly impaired due not only to the open bite but presumably also because of muscle weakness. The serum test for syphilis was negative. No sensory disturbances were noted. The WAIS intelligence scale indicated a score of 98. EEG showed high voltage slow activity during hyperventilation (Fig. 1) and 4-6 per second theta waves, seen dominantly in the right occipital area during drowsy state (Fig. 2). Activation by bemegride failed to show any tendency towards convulsive disorder. The profuse alpha component suggested of diffuse alpha activity. The electromyogram was silent in all muscles tested when the muscles were at rest. Prolonged duration of discharges during voluntary movement and diminished spike discharges

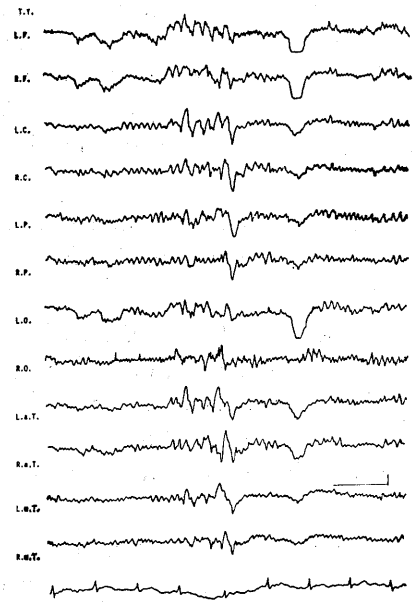

Fig. 1. Paroxysmal high voltage slow activity observed during hyperventilation.

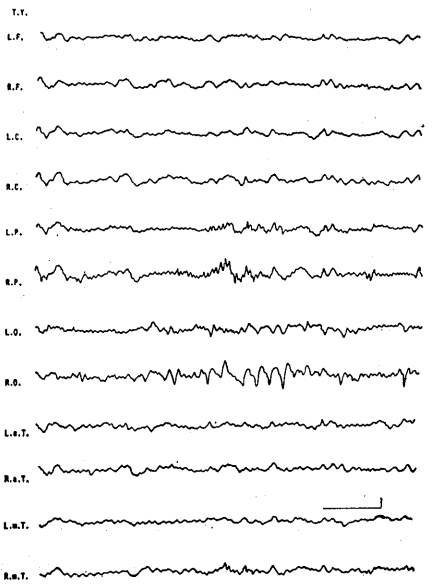

Fig .2. Right occipital slow waves seen in drowsy state.

associated with frequent dive bomber sounds in the flexura carpi were suggestive of myogenic change of EMG. Cerebrospinal fluid examination was not remarkable. The serum creatine phosphokinase level was slightly elevated at 39.5 U. The serum creatine level was normal as well as urinary creatine excretion. The T_3 resin uptake test was normal and serum aldolase activity was normal. Serum immunoglobulin value showed a slight decrease in Ig A. Urinary 17 KS and 17 OHCS excretion per day were within normal range.

BIOPSY FINDINGS

Deltoid muscle: No remarkable findings were observed under conventional photomicroscopy. No degeneration or inflammation were observed.

Gastrocunaemius muscle: Focal subsarcolemmal nuclei proliferation was observed by photomicroscopy. Occasional centrally located nuclei[7] and myofibrillar degeneration were also seen. Rarely were the nuclei arranged in a single, straight row.

Electron microscopic examination: Examination by electron microscope revealed a frequent lipid droplet accumulation (Photo. 3), subsarcolemmaly stored glycogen granules, mitochondria and lysosome-like bodies, all of which appeared to compress the myofibrils. Vacuoles were seen mostly around the mitochondria (Photo. 4). Intravacuolar myelin-like bodies were observed on occasion (Photo 5). T system proliferation was not decidedly demonstrable. There was no typical lattice-like figure reported by Schotland[8] or Z disc fragmentation[9]. Rarely were subsarcolemmal cytoplasmic bodies found (Photo. 6). As far as our investigation could reveal, ring fiber formation was not found.

COMMENT

One of the unusual clinical characteristics of this case was the maldevelopement or malformation of maxillar bone which appeared to be a rare complication of myotonic dystrophy; similar maldevelopement in association with myotonic dystropy has never been reported. On the other hand typical myotonic dystrophy symptoms of baldness and cataracts were not featured in this case, but the patient is still only 18 years old, so in all likelihood these features will become manifest in the future.

Myotonic phenomenon was observed in the tongue, hypothenar and thenar muscles.

The other characteristic features of this case were the psychiatric symptoms of character deviation. He was very ill-tempered. He was easily provoked into struggles against others. Oddly enough, he was

quite uninterested in his marked muscle weakness and wasting. His intelligence score was 98, though his verbal test was especially poor.

Electroencephalogram results disclosed a peculiar abnormality; i.e. occipital slow run appeared during the drowsy state. As he usually complained of visual hallucinations shortly after awakening during the night, perhaps the peculiar EEG changes observed might have some relation with these clinical psychic phenomenon. However, it still remains an open question whether these psychic problems have any causal relation with the pathogenesis of the myotonic disease or not. Bemegride activation failed to show that there was any tendency towards convulsive disorder.

Histologically, the findings were almost identical with previous reports on this disease[10)11)]. But the pathological features of this case were not especially marked, possibly because the patient is still quite young and thus has not yet progressed sufficiently. In other words, the findings we observed here seemed to be indicative of the early stage of this type of disease.

Under the electron microscope, some characteristic features of the myotonic dystrophy were seen. The most characteristic features of the initial stage of the disease are likely to be vacuolar changes in association with the appearance of special intramitochondrial myelin-like structures in the vacuoles[10)] and subsarcolemmal cytoplasmic bodies[12)]. The other associative findings were: lipid droplets, possible overproduction of T system, Z disc fragmentation and subsarcolemmal accumulation of mitochondria, although these findings are thought to be nonspecific degenerative findings. Interestingly enough, the ultrastructural findings were unexpectedly marked in comparison with those obtained by photomicroscopy. The electron microscope proved to be a very effective tool for detecting initial change of myopathic degeneration. Especially the gastrocunaemius muscle, which appeared to be healthy under clinical examination, but showed moderate change under electron microscopic examination.

Because none of these findings are very specific, a more precise examination of these and related disorders will be performed subsequently.

SUMMARY

The patient reported here, an 18-year-old boy, was suffering from myotonic dystrophy which was associated with psychic symptoms. Pertinent characteristics observed in this patient included personality change,

visual hallucinations during the night, muscle weakness of the upper extremities and an unusual malformation in the maxillary bone. Except for a slight muscle weakness, wasting and myotonic symptoms of the arm, hand and tongue muscles, clinical examination revealed no abnormalities in the musculature. An abnormality was observed by the EEG that was characterized by occipital high voltage slow run during the drowsy state.

The muscle biopsy specimen was examined by both photo and electron microscope. The findings were identical with previous reports on myotonic dystrophy but the course of the disease did not appear to have progressed as far as most previously described cases. The results of the muscle biopsy performed during the early stage of the disease were discussed.

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