Lafora Disease Diagnosed by Skin Biopsy

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Among progressive myoclonus epilepsy, Lafora disease is characterized by the presence of Lafora body (polyglucosan body) in the central neurons. A similar substance is found in other organs (Schwarz et al. 1965). Lafora disease has been so far diagnosed by liver (Inoue et al. 1974) or muscle biopsy (Carpenter et al. 1974). Recently however, skin biopsy is found to be very useful (Carpenter et al. 1981).

We would like to describe here a case report of Lafora disease diagnosed by skin biopsy.

Case Report

H. M., female, was 14 years old. The family and past history revealed nothing of note except for her parent’s consanguineous marriage.

She was considered to be a normal girl until 9 years old. At the age of 10, visual seizures (twinkling stars similar to a soap bubble) with myoclonus of eyelids started occurring and became progressive. When she was 12 years old, she began to present generalized seizures with impairment of consciousness, progressive mental retardation, gait disturbance and personality change. Since the age of 13, she was hypersensitive to light, by which visual seizures were induced. Other prominent symptoms included impediment in speech and memory disturbance. Myoclonus of the upper extremities and facial musculature also occurred and became gradually worse. When she was admitted to Kurume University Hospital at the age of 14, finger tremor, the above mentioned myoclonus and generalized convulsive fits were more progressive.

Mental symptoms such as progressive dementia and personality change were noted. Neurological abnormalities such as visual seizures, generalized seizures, myoclonic jerks (in the upper extremities and facial musculature), finger tremor, hypotonia, scanning speech and cerebellar symptoms were found. The spleen and liver were not palpable.

There were no abnormalities in laboratory findings. Blood and urine tests, liver and kidney functions and fundus were normal. No vacuoles of lymphocytes were observed. An EEG showed that basic activity was dominated by slow waves, showing diffusely polyspike and wave complex increased by photic stimulation (Fig. 1). CT scan showed slight dilatation of gyri. She scored 55 at the age of 13, and below 35 at the age of 14 in the intelli-
Skin biopsy of this patient was performed. The specimen showed numerous 5-7 μm oval or round PAS-positive materials in most of the glandular cells of eccrine glands. These materials were almost digested by α-amylase, but not by β-amylase. These were not stained with H.E. and alcian blue. Electronmicroscopic observation showed that the materials were made of numerous glycogen granules (Fig. 2, 3-A, B). The data obtained in this skin biopsy were in agreement with those reported by Carpenter et al. (1980) and by Kusunose et al (1984).

PAS-positive materials are not found in degeneration type of myoclonus epilepsy (diagnosed by clinical signs and liver biopsy).

Therefore, this case was diagnosed as Lafora disease.

References


SHORT COMMUNICATION

Fig. 1. EEG showing polyspike and wave complex.

Fig. 2. Section of skin biopsy showing PAS-positive substance in the granular cells (PAS staining 400×).

Fig. 3-A. PAS-positive materials were constituted of numerous glycogen granules.

Fig. 3-B. These are magnified in the inset.