



主要研究テーマ

• Distal Myopathy with Rimmed Vacuoles (DMRV) 🔅 Chloroquine Myopathy などの縁取り空胞ミオパチーにおいて、ubiquitin-proteasome system、および autophagy-lysosome system を検討することにより、これらの蛋白質分解経路が 筋線維の崩壊に関与していることを解明した。

Distal Myopathy Histochemical and Ultrastructural Studies

Toshihide Kumamoto, MD; Nobuyoshi Fukuhara, MD; Masaru Nagashima, MD; Takemasa Kanda, MD; Masatoshi Wakabayashi, MD

 $\mathbf{D}^{ ext{istal myopathy is characterized by}}$ In three familial cases and one spovery slowly progressive weakness radic case of late-onset distal myopathy, and wasting of the pretibial muscles muscle wasting started in the distal portions of the lower extremities. The most and the muscles of the hands and striking change seen by light microscopy forearms. Hereditary late-onset distal myopathy was clearly defined as a was the appearance of rimmed vacuoles. separate entity by Welander in 1951, These were presumed to be autophagic, because they were found by electron on the basis of 249 cases in 72 pediicroscopy to contain membranous grees from Sweden.¹ A number of amellar structures and other heterogecases, mostly familial, have since been nous materials enclosed by a limiting reported in many countries. nembrane. On the other hand, lysosomal Histologic features of this disease activity was markedly increased in skeleare similar to those of other types of tal muscle. In 6% to 22% of affected progressive muscular dystrophy.^{1,2} muscle fibers there were acid phospha-The disease was referred to as "distal tase-positive granules deep in the sarcomuscular dystrophy" by Walton and plasm, whereas control muscles had no Gardner-Medwin.³ Apart from a few such granules. The degenerative process reports,45 histochemical and ultrain distal myopathy may be different from structural changes have not been adethat in other muscular dystrophies. quately studied. (Arch Neurol 1982;39:367-371) This report presents histologic, his-

(case 2) and a sister (case 3) described in this report, had a similar neuromuscular disease

General examination findings were unremarkable. The patient had no mental abnormalities. Cranial nerves were normal, as were the facial and sternomastoid muscles. The muscles of the arms and lower extremities were wasted and weak The patient could walk on his toes but not on his heels. Gowers' sign and mild waddling gait were also observed. The distal muscles of the upper limbs were markedly affected, but the proximal muscles were almost normal (Fig 1, left). The scapulae were not winged. There were no fasciculations, pseudohypertrophy, contracture, or myotonia. Muscle stretch reflexes were diminished in the upper limbs, and normal in the lower limbs. Sensation and coordination were normal. Laboratory data were normal, except for the levels of SGOT and creatine phosphokinase (CPK), which were elevated on some determinations to a maxtochemical, and ultrastructural findimum of 48 units/dL (normal, < 35 units/ ings in four cases of late-onset distal dL) and 216 mU/mL (normal, <80 myopathy, three familial and one spomU/mL), respectively. Needle electromyo graphy (EMG) showed short-duration (5 ms) and low-amplitude (1 mV) potentials in all muscles examined. However, in the right rectus femoris and hamstring, we CASE 1.-A 25-year-old man had first occasionally observed slightly long-duranoted weakness in his legs when he was 23 tion (12 ms) and high-amplitude (8 mV) years old. He subsequently became unable potentials mixed with low-amplitude to run or hop and had increasing difficulty potentials. There were no fibrillation or in climbing stairs. At the age of 24 years, fasciculation potentials at rest, and no he noted slight bilateral weakness in the myotonic discharges. Motor and sensory hands and forearms. He had no paresthenerve conduction velocities were normal. His parents were first cousins. Three of CASE 2.- A 32-year-old man, an older brother of the first patient, had been his five siblings, including an older brother healthy until the age of 26 years, when he noted foot-drop and steppage gait. He sub-Accepted for publication June 11, 1981. From the Department of Neurology, Brain sequently became aware of weakness and wasting of the muscles in his lower limbs, Research Institute, Niigata (Japan) University. hands, and forearms. Reprint requests to Department of Neurology, Results of a general examination were Brain Research Institute, Niigata University, 1 normal. Mentation and cranial nerve func-Asahimachi-dori, Niigata 951, Japan (Dr Kuma-

tion were also normal, with no facial diple-



sia, pain, or cramps.

radic.

REPORT OF CASES

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●筋サルコイドーシス患者の生検筋の検討から、腫瘤型筋サルコイドーシスでは、 筋線維内に浸潤した肉芽腫性炎症細胞が肉芽腫を形成する過程で筋線維が崩壊 し、高度の線維化が引き起こされることを明らかにし、病因・病態解明に貢献した。

