Rimmed vacuolar distal myopathy in two Taiwanese siblings

陳啓仁

Wu YR;Ro LS;Chen ST;Chen CJ;

摘要

Abstract

Distal myopathy with rimmed vacuoles (DMRV) is a rare disease that has not previously been reported in Taiwan. This paper reports two siblings with DMRV. Each showed a different pattern of disease progression, one being slowly and the other rapidly progressive. Both patients' initial symptoms appeared in early adulthood, manifesting as muscular wasting and weakness of the legs, especially in the distal muscles. Severe generalized involvement of skeletal muscles, with sparing of the facial, extraocular, bulbar, intercostal and diaphragm muscles was recognized in the advanced stage. The striking finding in their muscle biopsy specimens was the presence of "rimmed" vacuoles. Magnetic resonance imaging showed more severe involvement of the anterior compartment muscles of the lower legs. DMRV is thought to be inherited as an autosomal recessive trait and is distinguishable from other types of distal myopathy on the basis of clinical and pathologic findings. The literature on the subject is reviewed with emphasis on the differences between distal myopathic syndromes.