三好氏遠端肌肉病變

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Miyoshi distal myopathy

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INTRODUCTION Miyoshi distal myopathy is a rare muscular dystrophy. It is inherited by autosomal recessive pattern, including homozygous and compound heterozygous mutation. The clinical manifestation demonstrates childhood onset, predominant involvement of calf muscles with relative spared anterior compartment muscle group and elevated serum creatine kinase level. Immunostain of muscle specimen reveals absence of dysferlin and the genetic mutation is currently mapped to chromosome 2p13, sharing the same locus with LGMD 2B.
CASE Mr. H was a 17-year-old male student, a person of mixed-blood from Taiwanese and Thai, without prominent medical history. Three years prior to admission, he noticed of difficulty with riding a bicycle or jumping high insidiously but could still perform activities of daily living and play the basketball, soccer and run. One year later, his right leg weakness became prominent which kept him from running and kicking a ball. The symptoms progressed slowly in the following years that he hardly managed with daily activities, such as climbing up- or down-stairs and no longer participated in sport activities. Upon examine, he demonstrated thin stature, prominent distal muscle atrophy and weakness of the leg, especially posterior compartment. Gower's sign was negative, but he failed to walk on-toe. Blood test showed elevation of serum CPK level (26455 IU/L). Electrophysiology studies demonstrated normal nerve conduction velocity, but increased positive sharp wave and fibrillation with brief and small-amplitude potentials of left gastrocnemius were noted. Leg MRI revealed increased signal intensity in the posterior compartment on T2WI sequence, especially the soleuses. A muscle biopsy from right soleus muscle showed dystrophic muscle fibers of variable fiber size in irregular distribution. Immunostain of dysferlin demonstrated markedly reduced with focal area of total loss. The genetic analysis of dysferlin revealed Exon 10 mutation (1310+1 G>A; intron position 1), while no significant mutation found in exons 2/13/54 (wild type). Reviewing his family history, his brother had similar clinical presentation, but further confirmation has not been estimated yet.
DISCUSSION Miyoshi myopathy is a rare distal myopathy, and it is different from neuropathy due to spared sensory component and electrophysiology study demonstrate myopathy pattern. In our case, we present a case of a mixed-blood of Taiwanese and Thai. Reviewing previous reports, 1310+1G>A mutation has been discovered in exon 10 among Chinese families and Japanese families which blocks the dysferlin expression. Currently, no cross-racial cases have been reported yet.

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