



## Images of a case with rippling muscle disease

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**Title: Images of a case with rippling muscle disease**

Running head: Images of RMD

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**Key words:** rippling muscle disease, percussion-induced rapid muscle contraction, muscle

hyperexcitability, caveolin-3, ultrasound

## **Case summary**

A 30-year-old man with rippling muscle disease (RMD) harboring an R27Q mutation in the caveolin-3 (*CAV3*) gene presented percussion-induced rapid muscle contractions and muscle rippling.<sup>1</sup> (Figure 1A, Video S1). The ultrasound revealed dynamic rolling muscle contractions spread diagonally or transversally to the axis of muscle fibers, showing a to-and-fro wavelike appearance not only in superficial parts but also in deep parts (Figure 1B, Video S2).

RMD is a myopathy characterized by mechanically-induced involuntary muscle contractions. The mislocalization of *CAV3* leads to increased activity of neuronal nitric oxide synthase in the skeletal muscle,<sup>2</sup> which may cause muscle hyperexcitability. The present study of visual inspection and ultrasound examination reveals a unique mode of muscle contraction in RMD in more details than previously reported.

## **Acknowledgments:**

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1. Maki T, Matsumoto R, Kohara N, et al. Rippling is not always electrically silent in rippling muscle disease: a case report. *Muscle Nerve* 2011;43:601-605.
2. Betz RC, Schoser BG, Kasper D, et al. Mutations in CAV3 cause mechanical hyperirritability of skeletal muscle in rippling muscle disease. *Nat Genet* 2001;28:218-219.

## **Figure legends**

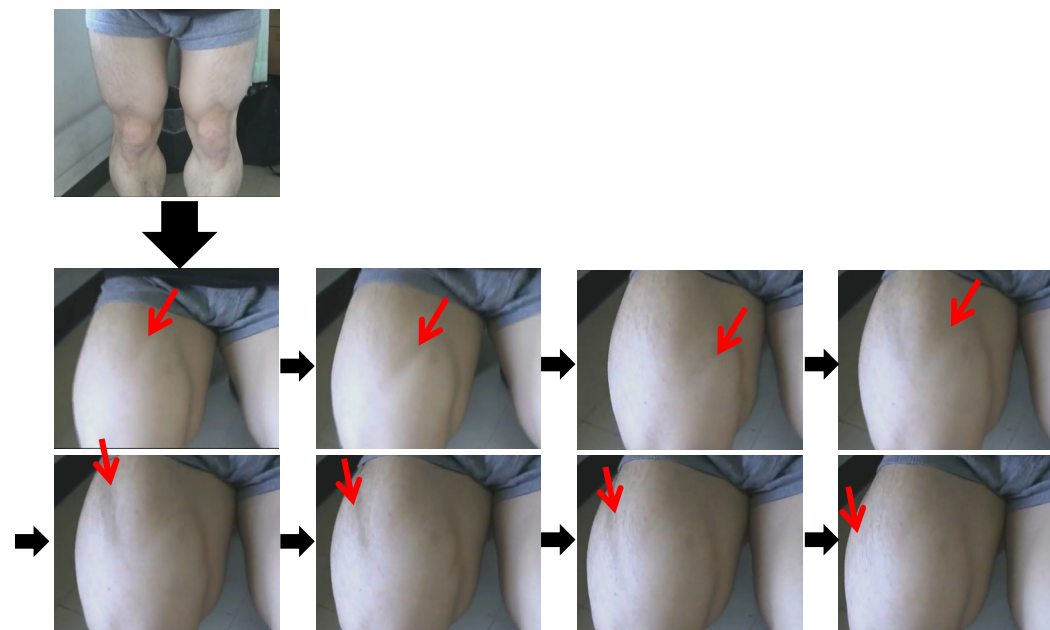
Figure 1 (A) Rippling muscle movement induced by a sudden stretch of the quadriceps femoris muscle, i.e., an abrupt knee flexion following its forceful extension for 15 seconds. Rippling muscle contractions (red arrows) spread diagonally or transversally to the axis of muscle fibers, occasionally showing a to-and-fro wavelike appearance. (B) The muscle ultrasound study revealed dynamic rippling muscle movement (red arrows) not only in superficial parts but also in deep parts. Recorded from the probe attached on the medial part of the muscle (blue arrow).

## **Supplemental video legends**

**Video S1:** Rippling muscle movement (arrows) induced by a sudden stretch of the quadriceps femoris muscle, i.e., an abrupt knee flexion following its forceful extension for 15 seconds.

**Video S2:** Ultrasound of the quadriceps femoris muscle during muscle rippling (arrows). Recorded from the probe attached on the medial part of the muscle.

**A**



**B**

