Alterations of excitation-contraction coupling and excitation coupled Ca(2+) entry in human myotubes carrying CAV3 mutations linked to rippling muscle

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Rippling muscle disease is caused by mutations in the gene encoding caveolin-3 (CAV3), the muscle-specific isoform of the scaffolding protein caveolin, a protein involved in the formation of caveolae. In healthy muscle, caveolin-3 is responsible for the formation of caveolae, which are highly organized sarcolemmal clusters influencing early muscle differentiation, signalling and Ca(2+) homeostasis. In the present study we examined Ca(2+) homeostasis and excitation-contraction (E-C) coupling in cultured myotubes derived from two patients with Rippling muscle disease with severe reduction in caveolin-3 expression; one patient harboured the heterozygous c.84C
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