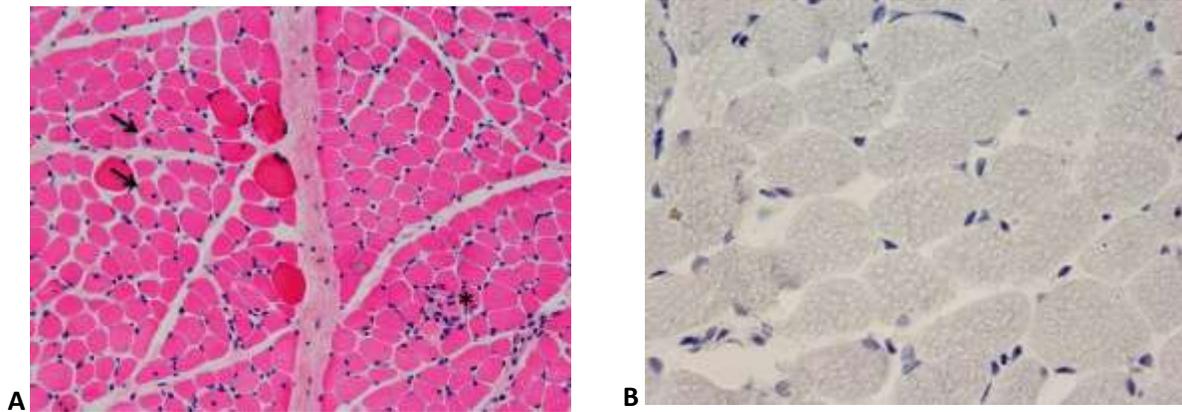


Supplementary Material I



Patient's muscle biopsy. A) The histological examination of slides stained with hematoxylin and eosin (H&E, 100x) revealed a slight to moderate change in muscle architecture. It was observed a diffuse variation in the size of muscle fibers, with hypertrophic and atrophic fibers with round and regular contours, and a slight increase in connective tissue. A moderate increase in internal nuclei (arrows) and occasional necrotic fibers were also identified (*). B) The immunohistochemical study revealed complete loss of expression of the DYS2 fraction of dystrophin (400x).