Reply to Sabatelli et al.: Detecting Collagen VI in Bethlem Myopathy

This is a response to a letter by Sabatelli et al. (1). In their letter, Sabatelli et al. (1) refer to our recent paper on a Bethlem myopathy patient with a homozygous COL6A2 p.D871N mutation. The authors are concerned that our data are inconsistent because we reported that collagen VI staining was absent in muscle when using monoclonal antibody VI-26 but present in very reduced amounts in cultured fibroblast extracellular matrix when detected with monoclonal antibody 3C4 (2). We agree that based on our in vitro fibroblast data it is likely that this patient’s muscle contains some collagen VI, and we believe that our staining conditions were not sufficiently sensitive to detect the small amount present even though collagen VI staining was strong in the control muscle. We had this in mind when we said “collagen VI was not detected in UCMD65 muscle” and used the phrase “absent collagen VI staining” under “Results.” Unfortunately we used the term “absent collagen VI” in the subheading, and this has resulted in the inadvertent apparent inconsistency. The collagen VI staining was actually done on sections of paraffin-embedded muscle, not frozen sections as suggested under “Experimental Procedures,” and this is why we used an antigen retrieval protocol, which resulted in strong staining in the control muscle (2). We apologize for omitting this important detail under “Experimental Procedures.”

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