

Supplemental Table 2. Serum CK and clinical muscle biopsy findings of 15 subjects.

Immunostaining results are as reported in the clinical pathology report. Antibodies are Dys1 (rod domain), Dys2 (C-terminal domain), and Dys3 (N-terminal domain) (all from Novocastra); and Mab1645 (Chemicon).

Patient	Serum CK in IU/L (age)	Age at muscle biopsy (years)	Muscle general histopathology	Dystrophin immunostaining (IHC = immunohistochemistry; IB = immunoblot)
1	5603-22000 (16-17)	17	variation in size, necrosis and regeneration, endomysial fibrosis	IHC: patchy negative fibers (with Dys1, Dys2, and Dys3)
2	1208-1628 (43)	-	-	-
3	30444 (6)	-	-	-
4	283-8954 (12-20)	9	variation in size, internal nuclei, necrosis and regeneration, endomysial fibrosis, fat replacement	IHC: scattered negative/reduced fibers (with Dys2)
5	4239 (27)	27	variation in size, internal nuclei, necrosis and regeneration, endomysial fibrosis, fat replacement	IHC: patchy negative/reduced fibers (with Dys1, Dys2, and Dys3); IB: reduction (with Dys1), normal size
6	20000 (5)	5	variation in size, necrosis and regeneration, endomysial fibrosis	IHC: no staining with Dys1, Dys2, or Dys3
7	235-65945 (12)	12	inflammation, myopathic grouping, necrosis and regeneration	IHC: patchy reduction; IB: reduction (with Dys2 and Dys3)
8	-	7	a few focal chronic inflammatory cells, variation in size, necrosis and regeneration, endomysial fibrosis	IHC: mosaic (MAB 1645): most fibers absent, with clusters of positive fibers.
9	512-3180 (8-10)	9	variation in size, rare internal nuclei, necrosis and regeneration, endomysial fibrosis	-
10	2000-5889 (7)	8	inflammation, internal nuclei, necrosis and regeneration	IHC: Patchy reduction (with an anti-N-terminus antibody), normal reactivity for the rod and C-terminal epitopes
11	423-585 (65-66)	66	variation in size, internal nuclei, necrosis and regeneration, endomysial fibrosis	IHC: Patchy reduction (with Dys3); IB: reduction (with Dys2 and Dys3, two sizes of dystrophin with Dys1)
12	165 (34)	-	-	-
13	67-108 (19-34)	-	-	-
14	500 (40)	-	-	-
15	506 (54)	55	variation in size, endomysial fibrosis	IHC: scattered negative/reduced fibers