

Supplementary Table S1. Demographic features and pathogenic mutations in patients with LGMD R2.

Patients	1	2	3	4	5	6	7	8
Age of onset (years)	32	30	31	26	57	32	26	24
Gender	Female	Male	Male	Male	Male	Male	Female	Female
Family history	No	No	No	No	Yes	No	No	No
MSAs or MAAs	(-)	Unknown	(-)	(-)	(-)	(-)	(-)	Unknown
Misdiagnosed	Yes	Yes	Yes	Yes	No	No	No	No
<i>DYSF</i> mutations	c.2646-1 G>A	c.2977 T>C	c.3113 G>A c.4863 C>A	c.5512 G>A	c.4714 C>T, c.1393 G>C	c.5626 G>A	c.2125 C>T c.5626 G>A	c.1141_1142insG

Abbreviations: LGMD R2, limb-girdle muscular dystrophy R2; MSAs, myositis specific antibodies; MAAs, myositis associated antibodies; IHC, Immunohistochemistry.