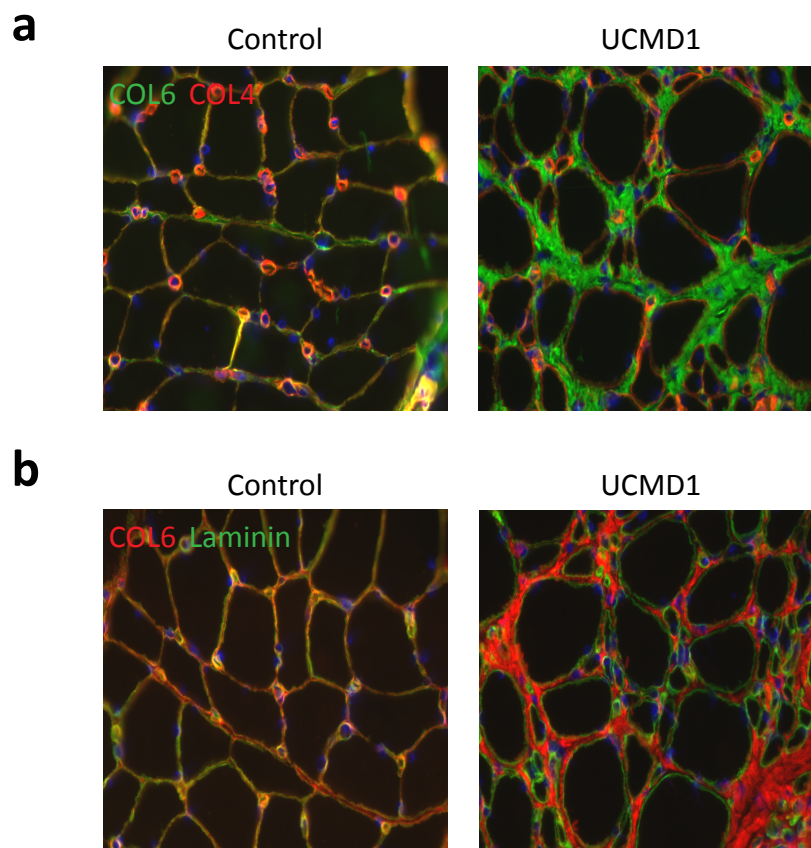


## Supplementary Material



**c** Mutations of UCMD1, UCMD2, UCMD3, UCMD4

	Nucleotide	RNA	Protein
UCMD1, UCMD3, UCMD4	c.6210+1G>A	r.spl_ex16	p.G2053_P2070del
UCMD2	c.6210+1G>T		

### Supplementary Figure S1

#### Genetic and histological description of the UCMD patients used in this study.

(a) Immunofluorescence imaging of muscle biopsy from patient UCMD1, co-stained with collagen VI (green) and basement membrane marker collagen IV (red); or (b) co-stained with collagen VI (red) and basement membrane marker laminin (green). (c) List of the patients used in this study and their mutations. Patient UCMD2 presents with a different substitution at the same nucleotide position compared to the three other patients. The two different mutations cause the skipping of exon 16 of COL6A3.