

**Supplementary Table 3.** Main features of muscle biopsies of individuals with homozygous variant c.1287+5G>A in *TRAPPC11*.

	Age at biopsy	Location	Fibrosis	Adipose tissue	Fibre size	Internal nuclei	Basophilic fibres	Regenerative fibres	Hpercontracted fibres	Necrosis	Intermyofibrillar pattern	Cox negative fibres	Lipids	Fibre type distribution	Sarcolemma protein staining	ADG (IIF6)	ADG (Via4)	Beta-DG
<b>Individual 4</b>	3	Left quadriceps	No	No	Moderate variability. Diameter 15-55 µm	<1%	5% (some in groups)	3%	1%	Yes	Intermyofibrillar pattern disruptions	No	No lipid increase	Normal mosaic pattern. Selective fast fibres atrophy.	Normal positive sarcolemma staining	Mosaic pattern with hyperintensity and hypointensity in some fibres	Mosaic pattern with hyperintensity and hypointensity in some fibres	Normal
<b>Individual 5</b>	3	Left quadriceps	No	No	Mild variability. Diameter 21-42 µm	<1%	No	<1%	5%	No	Intermyofibrillar pattern disruptions	No	No lipid increase	Normal mosaic distribution.	Normal positive sarcolemma staining	Normal	Normal	Normal
<b>Individual 8</b>	12	Left deltoids	No	No	Mild variability. Diameter 50-95 µm	5%	2%	<2%	5%	No	Intermyofibrillar pattern disruptions	No	No lipid increase	Normal mosaic distribution.	Normal positive sarcolemma staining	Mosaic pattern with hypointensity globally	Mosaic pattern with hypointensity globally	Normal
<b>Individual 11</b>	3	Right quadriceps	No	No	Moderate variability. Diameter 30-40 µm	2%	1%	<1%	5%	No	Intermyofibrillar pattern disruptions	No	No lipid increase	Normal mosaic distribution.	Normal positive sarcolemma staining	Mosaic pattern with hypointensity globally	Mosaic pattern with hypointensity globally	Normal
<b>Individual 13</b>	1,5	Quadriceps	No	No	Mild variability. Diameter 15-34 µm	1%	1%	<1%	7%	No	Intermyofibrillar pattern disruptions	No	No lipid increase	Normal mosaic distribution. Slow fibre type prevalence	Normal positive sarcolemma staining	Mosaic pattern with hyperintensity and hypointensity in some fibres	Mosaic pattern with hyperintensity and hypointensity in some fibres	Normal
<b>Individual 14</b>	12	Unknown	No	No	No variability	2%	No	No	No	No	Intermyofibrillar pattern disruptions	No	No lipid increase	Not assessable	Normal positive sarcolemma staining	Not assessable	Not assessable	Not assessable
<b>Individual 17</b>	4	Right quadriceps	No	No	Mild variability. Diameter 25-90 µm	<1%	No	1%	2%	No	Intermyofibrillar pattern disruptions	No	No lipid increase	Normal mosaic distribution. Selective fast fibres atrophy.	Normal positive sarcolemma staining	Mosaic pattern with hyperintensity and hypointensity in some fibres	Mosaic pattern with hyperintensity and hypointensity in some fibres	Normal
<b>Control individual without a neuromuscular disorder</b>	5	Right quadriceps	No	No	No variability	<1%	No	No	No	No	Normal	No	No lipid increase	Normal mosaic distribution.	Normal positive sarcolemma staining	Normal	Normal	Normal