**Supplementary table e-1:** Electrodiagnostic and genetic test results

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| Test (number of patients) | Results | Number of patients (percentage) |  Comments |
| Nerve conduction studies (27) | Normal |  17 (63%) |  |
| Low amplitude motor responses | 2 (7%) |  |
| Peripheral neuropathy | 3 (11%) | 2 axonal and 1 with demyelinating features |
| Superimposed mononeuropathy | 5 (19%) | Median (n=3), ulnar (n=2), trigeminal (n=1) |
| Needle electromyography (27) | Fibrillation potentials | 22 (82%) | Distribution as follows: axial muscles only in 7, axial and proximal in 6, proximal in 2, diffuse in 6 and random in 1.  |
|  fasciculation potentials | 5 (19%) | Only in 1 muscle in 1 patient, in 2 muscles in 2 patients, in 3 muscles in 1 patient, and in several muscles in 1 patient. |
| Myotonic discharges | 1 (4%) | In axial muscles and iliopsoas. |
| Short duration motor unit potential | 27 (100%) | Distribution as follows: axial muscles only in 4, axial and proximal in 11, proximal in 2, diffuse in 9 and random in 1.  |
| Long duration motor unit potential | 13 (48%) | Distribution as follows: random in 7, diffuse in 2, lower limb only in 1, cranial in 1, and distal in 3 (may be due to a coexisting peripheral neuropathy). |
| Genetic testing (9) | Normal myopathy gene panel by next-generation sequencing  | 5 | Included the following genes associated with a nemaline myopathy: *ACTA1, CFL2, KLHL41, KBTBD13, KLHL40, NEB, TNNT1, TPM2,*and *TPM*. In addition to *LMOD3* and *MYPN* in 2 patients. |
| Muscle mitochondrial DNA sequencing and deletion analysis\* | 3 | Performed because of concomitant mitochondrial histological abnormalities on muscle biopsy |
| Fascioscapulohumeral dystrophy type 1  | 2 | Performed because of the scapula winging and axial weakness |
| Muscle biopsy (30) | Diagnostic  | 30 | Biopsied muscle: biceps (n=9), cervical paraspinal (4 including one from the splenius capitis), deltoid (4), gluteus medius (3), quadriceps (3), tensor fascia lata (1), trapezius (1), triceps (1), thigh muscle (1\*) and biopsy site not specified (3\*). |
|  | Non diagnostic | 10 | Biopsied muscle: deltoid (n=2), quadriceps (2), biceps (1), lumbar paraspinals (1), and thoracic paraspinals (1), thigh muscle (1\*), arm muscle (1\*) and site not specified (1\*).  |

Diffuse: majority of muscles showed the abnormality; random: scattered muscles showed the abnormality but not following a specific pattern.

\*Obtained at an outside facility