|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Table S1** The mode of inheritance and electrophysiological data of the 36 unrelated patients with Charcot-Marie-Tooth disease type 2 | | | | | | | | | | | | | |
| Patient | Gender | Age at NCS (yrs) | Mode of inheritance | Motor NCS | | | | Sensory NCS | | | | | |
| Median nerve | | Peroneal nerve | | Median nerve | | Ulnar nerve | | Sural nerve | |
|  |  |  | NCV, m/s | CMAP, mV | NCV, m/s | CMAP, mV | DL,  ms | SNAP, uV | DL, ms | SNAP, uV | DL, ms | SNAP, uV |
| Normal value1🡪 | |  |  | ≧51.92 | ≧6.42 | ≧422 | ≧2.12 | ≦2.93 | ≧172 | ≦3.03 | ≧172 | ≦3.33 | ≧122 |
| 14 | M | 51 | AD | 38.1 | 6.3 | NR | NR | NR | NR | NR | NR | NR | NR |
| 24 | F | 5 | AS | 35.1 | 2.3 | NR | NR | 2.7 | 7.4 | 1.7 | 7.8 | NR | NR |
| 34 | M | 34 | AD | 32.1 | 1.8 | NR | NR | NR | NR | NR | NR | NR | NR |
| 44 | F | 26 | AD | 40.8 | 0.5 | NR | NR | NR | NR | NR | NR | NR | NR |
| 54 | M | 47 | AS | 38.2 | 1.5 | NR | NR | NR | NR | NR | NR | NR | NR |
| 64 | F | 45 | AD | 33 | 0.3 | NR | NR | NR | NR | NR | NR | NR | NR |
| 74 | F | 43 | AD | 47.9 | 2.6 | 33.5 | 0.2 | NR | NR | NR | NR | NR | NR |
| 84 | M | 27 | AR | 41.3 | 1.6 | NR | NR | NR | NR | NR | NR | NR | NR |
| 94 | F | 60 | AD | 40.3 | 7.5 | 27.1 | 1.2 | NR | NR | 4.4 | 1.2 | NR | NR |
| 104 | M | 20 | AD | 56.5 | 9.8 | 38.8 | 3.4 | 3.3 | 9 | 2.9 | 9 | NR | NR |
| 114 | M | 41 | AR | 65.3 | 9 | 44 | 6.5 | 2.5 | 17 | 2.6 | 11.6 | 3.1 | 8.8 |
| 124 | M | 6 | AS | 37.2 | 4 | NR | NR | NR | NR | NR | NR | NR | NR |
| 134 | M | 13 | AS | 40.2 | 0.2 | NR | NR | NR | NR | NR | NR | NR | NR |
| 144 | M | 32 | AD | 49 | 5.2 | NR | NR | NR | NR | NR | NR | NR | NR |
| 15 | F | 46 | AD | 41.1 | 7.3 | 39 | 0.5 | 3.9 | 10 | 2.6 | 7 | NR | NR |
| 16 | F | 14 | AD | 44.2 | 0.3 | NR | NR | NR | NR | NR | NR | NR | NR |
| 17 | M | 33 | AD | 44.7 | 2.2 | NR | NR | NR | NR | NR | NR | NR | NR |
| 18 | M | 50 | AD | NR | NR | 45 | 0.5 | 2 | 11.4 | 2.3 | 12.2 | 2.6 | 8.5 |
| 19 | M | 51 | AD | 46.6 | 5.5 | 30.7 | 0.3 | 3.1 | 16.8 | 2.9 | 14.4 | NR | NR |
| 20 | F | 21 | AS | 56.1 | 4 | NR | NR | NR | NR | NR | NR | NR | NR |
| 21 | M | 62 | AR | 60.2 | 4.5 | 42.1 | 1 | 2.44 | 9 | 3.74 | 7.2 | NR | NR |
| 22 | M | 25 | AS | 38.5 | 6.2 | NR | NR | 3.92 | 8.6 | NR | NR | NR | NR |
| 23 | M | 59 | AR | 43.6 | 2.4 | 27.6 | 0.4 | NR | NR | NR | NR | NR | NR |
| 24 | M | 57 | AD | 56 | 9 | 35.3 | 1.3 | NR | NR | NR | NR | NR | NR |
| 25 | F | 38 | AS | 49.4 | 5.6 | NR | NR | 3.5 | 4.4 | 4 | 6.8 | NR | NR |
| 26 | M | 42 | AR | 55.2 | 7.4 | 33.8 | 0.2 | 3.12 | 8.8 | 3.1 | 7.6 | NR | NR |
| 27 | M | 30 | AD | 48 | 1.2 | 38.3 | 0.1 | 3.5 | 7.6 | 3.6 | 11.6 | 3.7 | 8 |
| 28 | F | 36 | AR | 60.3 | 13.3 | NR | NR | 2.8 | 26 | 2.7 | 16 | 2.9 | 8.4 |
| 29 | M | 55 | AR | 41.8 | 0.6 | NR | NR | 2.6 | 17 | 2.4 | 9.5 | 2.9 | 5 |
| 30 | F | 35 | AD | 56.7 | 5.7 | 48.5 | 3.1 | 2.8 | 8.4 | 2.7 | 8.4 | NR | NR |
| 31 | M | 31 | AD | NR | NR | NR | NR | NR | NR | NR | NR | NR | NR |
| 32 | F | 22 | AS | 61.4 | 8.8 | NR | NR | 4.2 | 6.8 | 3.2 | 6.8 | NR | NR |
| 33 | M | 18 | AD | 39.8 | 7.4 | 31.4 | 0.4 | 3.8 | 7.3 | NR | NR | NR | NR |
| 34 | M | 62 | AD | 44.2 | 6.1 | NR | NR | 3.4 | 6.8 | NR | NR | NR | NR |
| 35 | M | 22 | AS | 60.7 | 7.6 | 50.7 | 1.5 | 3.4 | 18.7 | 2.9 | 13.2 | 2.7 | 8.3 |
| 36 | M | 36 | AD | 56.8 | 1.9 | 48.5 | 2 | 2.4 | 10.1 | 2.2 | 10.4 | 3.2 | 4.1 |

NCS: nerve conduction study; NCV: nerve conduction velocity; CMAP: compound muscle action potential; SNAP: sensory nerve action potential; DL: distal latency; yrs: years; m/s: meter per second; mV: millivolt; ms: millisecond; uV: microvolt; AD: autosomal dominant; AR: autosomal recessive; AS: apparently sporadic; NR: no response.

1The normal values in 100 nerves from 50 Han Chinese individuals, 20 to 64 years of age (average, 38), with no apparent disease of the peripheral nerves.

2Lower limits of normal, calculated as the mean – 2 standard deviation (SD).

3Upper limits of normal, calculated as the mean + 2 SD.

4The genetic and clinical information of these patients were also listed in Table 1.