

Supplement to “The most common European HINT1 neuropathy variant phenotype and its case studies”

Supplement Table 1. Allele frequency of variant c.110G>C (p.Arg37Pro) in European populations.

| Population | Allele frequency |
|------------------|------------------|
| Latvia | 0.0100 |
| Bulgaria | 0.0037 |
| Estonia | 0.0031 |
| Russia | 0.0020 |
| Finland | 0.0009 |
| Sweden | 0.0005 |
| Northwest Europe | 0.0001 |
| South Europe | 0.0000 |

Supplement Table 2. Nerve ultrasonography of patients with *HINT1* neuropathy

| | Median nerve cross-section area (CSA) dx (cm ²) | | | | | | | | Ulnar nerve cross-section area (CSA) dx (cm ²) | | | | | | | |
|---|---|-----------------|-----------------|-----------------|-----------------|-----------------|------------|-----------|--|-----------------|-----------------|-----------------|-----------------|-----------------|------------|-----------|
| | P1 HINT 1 | P2 HINT 1 | P3 HINT 1 | P4 HINT 1 | P5 CMT1 A | P6 CMT1 A | P7 AGel | P8 ALS | P1 HINT 1 | P2 HINT 1 | P3 HINT 1 | P4 HINT 1 | P5 CMT1 A | P6 CMT1 A | P7 AGel | P8 ALS |
| 1 | 0.07 | 0.09 | 0.10 | 0.09 | 0.08 | 0.19 | 0.1 5 | 0.1 1 | 0.06 | 0.04 | 0.04 | 0.05 | 0.06 | 0.10 | 0.0 5 | 0.0 6 |
| 2 | 0.06 | 0.04 | 0.06 | 0.06 | 0.18 | 0.23 | 0.1 1 | 0.0 9 | 0.04 | 0.05 | 0.07 | 0.05 | 0.07 | 0.15 | 0.0 6 | 0.0 6 |
| 3 | 0.06 | 0.05 | 0.11 | 0.05 | 0.27 | 0.38 | 0.1 1 | 0.0 7 | 0.05 | 0.06 | 0.06 | 0.05 | 0.14 | 0.25 | 0.0 8 | 0.1 1 |
| 4 | 0.07 | 0.06 | 0.11 | 0.08 | 0.12 | 0.32 | 0.1 6 | 0.1 0 | 0.07 | 0/06 | 0.07 | 0.08 | 0.08 | 0.13 | 0.1 4 | 0.0 8 |
| 5 | 0.10 | 0.05 | 0.10 | 0.09 | 0.58 | 0.33 | 0.1 6 | 0.2 0 | 0.06 | 0.06 | 0.06 | 0.04 | 0.17 | 0.25 | 0.1 4 | 0.1 2 |
| 6 | 0.10 | 0.05 | 0.07 | 0.06 | 0.44 | 0.28 | 0.2 9 | 0.1 1 | 0.05 | 0.05 | 0.07 | 0.08 | 0.32 | 0.33 | 0.2 1 | 0.1 1 |

Median nerve positions: 1) on the level of the carpal canal, 2) proximal to the carpal canal, 3) middle 1/3 of the forearm, 4) proximal to cubital fossa, 5) in the cubital fossa, 6) on the level of armpit region (axillary region);

Ulnar nerve positions: 1) on the level of the Guyons canal, 2) proximal to the Guyons canal, 3) middle 1/3 of the forearm, 4) below elbow 6) above elbow