**Supplementary Table 1: Search Strategy (date of search: 05/07/2019)**

|  |  |  |  |
| --- | --- | --- | --- |
| Database (see URLs in Bibliography)  | Search expressions | Filters | Number of studies |
| European Clinical Trials Database (EudraCT)  | (neuropathy AND (inherited OR genetic)) OR (distal hereditary motor neuropathy OR dHMN OR motor neuropathy OR distal hereditary motor neuronopathy) OR (CMT OR charcot-marie-tooth OR hereditary motor and sensory neuropathy OR dejerine sottas disease) OR (hereditary sensory and autonomic neuropathy OR HSAN) | Trials with results | 17 |
| Centre for Reviews and Dissemination (CRD) | (neuropathy AND (inherited OR genetic)) OR (distal hereditary motor neuropathy OR dHMN OR motor neuropathy OR distal hereditary motor neuronopathy) OR (CMT OR charcot-marie-tooth OR hereditary motor and sensory neuropathy OR dejerine sottas disease) OR (hereditary sensory and autonomic neuropathy OR HSAN) | (Any field:) | 21 |
| ClinicalTrials.gov | (neuropathy AND (inherited OR genetic)) OR (distal hereditary motor neuropathy OR dHMN OR motor neuropathy OR distal hereditary motor neuronopathy) OR (CMT OR charcot-marie-tooth OR hereditary motor and sensory neuropathy OR dejerine sottas disease) | Neuropathy; Studies with Results; Interventional Studies | 47 |
| The Cochrane Central Register of Controlled Trials (CENTRAL) | (((neuropathy AND (inherited OR genetic)) OR (distal hereditary motor neuropathy OR dHMN OR motor neuropathy OR distal hereditary motor neuronopathy) OR (CMT OR charcot-marie-tooth OR hereditary motor and sensory neuropathy OR dejerine sottas disease) OR (hereditary sensory and autonomic neuropathy OR HSAN)) AND ((treatment OR intervention OR therapy) AND (RCT OR randomised controlled trial OR randomised-controlled-trial OR non-randomised OR case study OR case report OR observational))) NOT (chemotherapy-induced) NOT (diabetic) NOT (cancer) NOT (retina) NOT (optic) NOT (Guillain-Barré) | **(Title Abstract Key word:); Trials;** **Year First Published 1991** | 365 |
| PubMed | (((neuropathy AND (inherited OR genetic)) OR (distal hereditary motor neuropathy OR dHMN OR motor neuropathy OR distal hereditary motor neuronopathy) OR (CMT OR charcot-marie-tooth OR hereditary motor and sensory neuropathy OR dejerine sottas disease) OR (hereditary sensory and autonomic neuropathy OR HSAN)) AND ((treatment OR intervention OR therapy) AND (RCT OR randomised controlled trial OR randomised-controlled-trial OR non-randomised OR case study OR case report OR observational)))NOT(chemotherapy-induced) NOT (diabetic) NOT (cancer) NOT (retina) NOT (optic) NOT(Guillain-Barré) NOT (multifocal) NOT (chronic inflammatory) NOT (carpal) NOT (neuritis) NOT (HIV) NOT (carboplatin) NOT (mononeuritis multiplex) | Publication date from 01/01/1991;Species: Human | 1414 |
| The Cochrane Neuromuscular Disease Group Specialized Register (CNDGSR)  | N/A | Reviews by subtopic; Peripheral neuropathy; Hereditary neuropathy | 6 |
| WHO International Clinical Trials Registry Platform (WHO ICTRP) | charcot-marie-tooth | With results only | 1 |

**Supplementary Table 3: Full mutation information for Tables 1-3**

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| --- | --- |
| **First Author, Journal (Year)** | **Mutations (Gene, RefSeq, amino acid code)** |
| Berk, JAMA (2013) | *TTR* (NM\_000371.3):Val30Met, Leu58His, Thr60Ala, Ser50Arg, Phe64Leu, Asp38Ala, Ser77Tyr, Glu89Gln, Val122Ile, Val30Gly, Val32Ala, Lys35Asn, Lys35Thr, Gly42Gly, Phe44Ser, Thr49Pro, Glu54Gln, Val71Ala, Tyr78Phe, Ile84Asn, Ala97Ser, Ile107Phe |
| Cortese, J Neurol (2016) | *TTR* (NM\_000371.3):Val30Met, Phe64Leu, Glu89Gln, Thr49Ala, Tyr78Phe, Ala120Ser, Glu54Gln, Ile68Leu, Ser77Tyr, Ala45Thr |
| Barroso, Amyloid (2017) | *TTR* (NM\_000371.3): Val30Met, Asp38Ala, Gly47Ala, Leu58His, Thr60Ala, Phe64Leu, Ser77Phe, Ser77Tyr, Ile107Val |
| EudraCT number: 2007-006791-12 (2016)  | *TTR* (NM\_000371.3): Ser77Tyr, Thr60Ala, Tyr114Cys, Leu58His, Glu89Gln, Ser77Phe, Thr49Ala, Ile107Val, Val30Ala, Gly47Ala, Gly47Glu, Leu55Arg, Lys70Asn, Ile84Thr, Ile107Met |
| EudraCT number: 2013-002987-17 (2017)D. Adams, N Engl J Med (2018) | *TTR* (NM\_000371.3) Val30Met, Ala97Ser, Thr60Ala, Glu89Gln, Ser50Arg, Ser77Tyr, Asp38Ala, Phe64Leu, Ile107Val, Glu89Lys, Gly47Ala, Leu58His, Thr49Ala, Gly47Glu, Gly47Val, Lys35Asn, Ser77Phe, Val122Ile, Tyr114Cys, Ala36Pro, Ala45Thr, Asp38Val, Glu42Gly, Glu54Asp, Glu54Gln, Glu61Lys, Phe33Leu, Phe44Ser, Gly42Asp, His88Arg, Ile84Thr, Ile107Val, Pro24Ser, Ser50Ile, Ser52Pro, Thr49Ile, Thr59Lys, Val71Ala, Tyr78Phe |
| M. D. Benson, N Engl J Med (2018) | *TTR* (NM\_000371.3):Val30Met, Thr60Ala, Leu58His, Ser77Tyr, Phe64Leu, Ser50Arg, Glu89Gln, Val122Ile, Thr49Ala, Ala109Ser, Ala97Ser, Asp38Ala, Glu54Ser, Glu61Lys, Glu89Lys, Gly47Ala, Gly67Arg, Ile107Phe, Ile107Val, Ile84Ser, Lys35Thr, Lys70Asn, Phe33Leu, Pro24Ser, Ser77Phe, Thr59Lys,Tyr114Cys |
| Foley, Brain (2014) | SLC52A2 (NM\_001253815.2):p.Gly306Arg, p.Trp31Ser, p.Gln234, p.Ala284Asp, p.Tyr305Cys, p.Leu312Pro, p.Leu339Pro, p.Ala420Thr  |