Supplementary Table 1 The 113 genes associated with peripheral nerves disturbances, analyzed firstly in the WES data from the patient.

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| **Gene symbol** | **Gene name** | **Reference sequence number** |
| *AARS* | alanyl-tRNA synthetase | NG\_023191.1; NM\_001605.2 |
| *ABHD12* | abhydrolase domain containing 12 | NG\_028119.1; NM\_001042472.2  |
| *AIFM1* | apoptosis inducing factor, mitochondria associated 1 | NG\_013217.1; NM\_004208.3 |
| *ARHGEF10* | Rho guanine nucleotide exchange factor 10 | NG\_008480.1; NM\_014629.3 |
| *ARSA* | arylsulfatase A | NG\_009260.2; NM\_000487.5 |
| *ASAH1* | N-acylsphingosine amidohydrolase 1 | NG\_008985.1; NM\_177924.4 |
| *ATL1* | atlastin GTPase 1 | NG\_009028.1; NM\_001127713.1  |
| *ATL3* | atlastin GTPase 3 | NG\_033985.1; NM\_015459.4 |
| *ATP7A* | ATPase copper transporting alpha | NG\_013224.2; NM\_000052.6 |
| *BICD2* | BICD cargo adaptor 2 | NG\_033908.1; NM\_000052.6 |
| *BSCL2* | BSCL2, seipin lipid droplet biogenesis associated | NG\_008461.1; NM\_032667.6 |
| *C12ORF65* | chromosome 12 open reading frame 65 | NG\_027517.1 ; NM\_001143905.2  |
| *CCT5* | chaperonin containing TCP1 subunit 5 | NG\_012160.1 ; NM\_012073.4  |
| *CLTCL1* | clathrin heavy chain like 1 | NG\_033805.1; NM\_007098.3 |
| *COX6A1* | cytochrome c oxidase subunit 6A1 | NG\_034299.1; NM\_004373.3 |
| *CTDP1* | CTD phosphatase subunit 1 | NG\_007988.1; NM\_004715.4 |
| *DARS* | aspartyl-tRNA synthetase | NG\_034149.1; NM\_001349.3  |
| *DCAF8* | DDB1 and CUL4 associated factor 8 | NG\_034154.1; NM\_015726.3 |
| *DCTN1* | dynactin subunit 1 | NG\_008735.2; NM\_004082.4 |
| *DHH* | desert hedgehog | NG\_008973.2; NM\_021044.3  |
| *DHTKD1* | dehydrogenase E1 and transketolase domain containing 1 | NG\_033248.1; NM\_018706.6 |
| *DNAJB2* | DnaJ heat shock protein family (Hsp40) member B2 | NG\_029553.1; NM\_001039550.1 |
| *DNAJC3* | DnaJ heat shock protein family (Hsp40) member C3 | NG\_041830.1; NM\_006260.4 |
| *DNM2* | dynamin 2 | NG\_008792.1; NM\_001005360.2 |
| *DNMT1* | DNA methyltransferase 1 | NG\_028016.3; NM\_001130823.2 |
| *DRP2* | dystrophin related protein 2 | NG\_016403.1; NM\_001939.2 |
| *DST* | dystonin | NG\_029322.2; NM\_001723.5 |
| *DYNC1H1* | dynein cytoplasmic 1 heavy chain 1 | NG\_008777.1; NM\_001376.4 |
| *EGR2* | early growth response 2 | NG\_008936.2; NM\_000399.4 |
| *FAM134B* | family with sequence similarity 134 member B | NG\_016644.2; NM\_001034850.2 |
| *FBLN5* | fibulin 5 | NG\_008254.1; NM\_006329.3 |
| *FBXO38* | F-box protein 38 | NG\_033871.1; NM\_030793.4 |
| *FGD4* | FYVE, RhoGEF and PH domain containing 4 | NG\_008626.2; NM\_139241.3 |
| *FIG4* | FIG4 phosphoinositide 5-phosphatase | NG\_007977.1; NM\_014845.5 |
| *FLVCR1* | feline leukemia virus subgroup C cellular receptor 1 | NG\_028131.1; NM\_014053.3 |
| *GALC* | galactosylceramidase | NG\_011853.2; NM\_000153.3 |
| *GAN* | gigaxonin | NG\_009007.1; NM\_022041.3 |
| *GARS* | glycyl-tRNA synthetase | NG\_007942.1; NM\_002047.3 |
| *GDAP1* | ganglioside induced differentiation associated protein 1 | NG\_008787.3; NM\_018972.2 |
| *GJB1* | gap junction protein beta 1 | NG\_008357.1; NM\_000166.5 |
| *GJB3* | gap junction protein beta 3 | NG\_008309.1; NM\_024009.2 |
| *GNB4* | G protein subunit beta 4 | NG\_033163.1; NM\_021629.3 |
| *HADHB* | hydroxyacyl-CoA dehydrogenase/3-ketoacyl-CoA thiolase/enoyl-CoA hydratase (trifunctional protein), beta subunit | NG\_007294.1; NM\_000183.2 |
| *HARS* | histidyl-tRNA synthetase | NG\_032158.1; NM\_002109.5 |
| *HINT1* | histidine triad nucleotide binding protein 1 | NG\_032998.1; NM\_005340.6 |
| *HK1* | hexokinase 1 | NG\_012077.1; NM\_033498.2 |
| *HOXD10* | homeobox D10 | NG\_008133.2; NM\_002148.3 |
| *HNRNPA1* | heterogeneous nuclear ribonucleoprotein A1 | NG\_033830.1; NM\_002136.3 |
| *HSPB1* | heat shock protein family B (small) member 1 | NG\_008995.1; NM\_001540.3 |
| *HSPB3* | heat shock protein family B (small) member 3 | NG\_027758.1; NM\_006308.2 |
| *HSPB8* | heat shock protein family B (small) member 8 | NG\_007953.2; NM\_014365.2 |
| *IGHMBP2* | immunoglobulin mu binding protein 2 | NG\_007976.1; NM\_002180.2 |
| *IKBKAP* | inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein | NG\_008788.1; NM\_003640.4 |
| *INF2* | inverted formin, FH2 and WH2 domain containing | NG\_027684.1; NM\_022489.3  |
| *IFRD1* | interferon related developmental regulator 1 | NG\_027799.1; NM\_001007245.2 |
| *KARS* | lysyl-tRNA synthetase | NG\_028025.1; NM\_001130089.1 |
| *KCC3/SLC12A6* | solute carrier family 12 member 6 | NG\_007951.1; NM\_133647.1 |
| *KIF1A/ATSV* | kinesin family member 1A | NG\_029724.1; NM\_001244008.1 |
| *KIF1B* | kinesin family member 1B | NG\_008069.1; NM\_015074.3 |
| *KIF5A* | kinesin family member 5A | NG\_008155.1; NM\_004984.2 |
| *LITAF* | lipopolysaccharide induced TNF factor | NG\_009008.1; NM\_004862.3 |
| *LMNA* | lamin A/C | NG\_008692.2; NM\_170707.3 |
| *LRSAM1* | leucine rich repeat and sterile alpha motif containing 1 | NG\_032008.1; NM\_138361.5 |
| *MARS* | methionyl-tRNA synthetase | NG\_034077.1; NM\_004990.3 |
| *MED25* | mediator complex subunit 25 | NG\_017091.1; NM\_030973.3 |
| *MEN2B/RET* | ret proto-oncogene | NG\_007489.1; NM\_020975.4 |
| *MFN2* | mitofusin 2 | NG\_007945.1; NM\_014874.3 |
| *MORC2* | MORC family CW-type zinc finger 2 | NG\_046752.1; NM\_001303256.2 |
| *MPZ* | myelin protein zero | NG\_008055.1; NM\_000530.7 |
| *MTMR2* | myotubularin related protein 2 | NG\_008333.1; NM\_016156.5 |
| *NAGLU* | N-acetyl-alpha-glucosaminidase | NG\_011552.1; NM\_000263.3 |
| *NDRG1* | N-myc downstream regulated 1 | NG\_007943.1; NM\_006096.3 |
| *NEFL* | neurofilament, light polypeptide | NG\_008492.1; NM\_006158.4 |
| *NGFB* | nerve growth factor | NG\_007944.1; NM\_002506.2 |
| *NTRK1* | neurotrophic receptor tyrosine kinase 1 | NG\_007493.1; NM\_002529.3 |
| *PDK3* | pyruvate dehydrogenase kinase 3 | NG\_016762.1; NM\_001142386.2 |
| *PHYH* | phytanoyl-CoA 2-hydroxylase | NG\_012862.1; NM\_006214.3 |
| *PLA2G6* | phospholipase A2 group VI | NG\_007094.2; NM\_003560.2 |
| *PLEKHG5* | pleckstrin homology and RhoGEF domain containing G5 | NG\_007978.1; NM\_020631.4 |
| *PMM2* | phosphomannomutase 2 | NG\_009209.1; NM\_000303.2 |
| *PMP22* | peripheral myelin protein 22 | NG\_007949.1; NM\_000304.3 |
| *POLG* | DNA polymerase gamma, catalytic subunit | NG\_008218.2; NM\_002693.2 |
| *PRDM12* | PR/SET domain 12 | NC\_000009.12; NM\_021619.2  |
| *PRNP* | prion protein | NG\_009087.1; NM\_000311.3 |
| *PRPS1* | phosphoribosyl pyrophosphate synthetase 1p | NG\_008407.1; NM\_002764.3 |
| *PRX* | periaxin | NG\_007979.1; NM\_181882.2 |
| *RAB7* | RAB7A, member RAS oncogene family | NG\_008070.1; NM\_004637.5 |
| *RARS* | arginyl-tRNA synthetase | NG\_041809.1; NM\_002887.3 |
| *REEP1* | receptor accessory protein 1 | NG\_013037.1; NM\_022912.2  |
| *RFVT2/SLC52A2* | solute carrier family 52 member 2 | NG\_032872.1; NM\_024531.4 |
| *RNF170* | ring finger protein 170 | NG\_032868.1; NM\_001160223.1 |
| *RYR1* | ryanodine receptor 1 | NG\_008866.1; NM\_000540.2 |
| *SBF1* | SET binding factor 1 | NG\_041810.1; NM\_002972.3 |
| *SBF2* | SET binding factor 2 | NG\_008074.1; NM\_030962.3 |
| *SCN11A* | sodium voltage-gated channel alpha subunit 11 | NG\_033859.1; NM\_014139.2 |
| *SCN9A* | sodium voltage-gated channel alpha subunit 9 | NG\_012798.1; NM\_002977.3 |
| *SETX* | senataxin | NG\_007946.1; NM\_015046.5 |
| *SH3TC2* | SH3 domain and tetratricopeptide repeats 2 | NG\_007947.2; NM\_024577.3  |
| *SLC25A19* | solute carrier family 25 member 19 | NG\_008274.1; NM\_001126121.1 |
| *SLC5A7* | solute carrier family 5 member 7 | NG\_042267.1; NM\_021815.4 |
| *SOX10* | SRY-box 10 | NG\_007948.1; NM\_006941.3 |
| *SPTLC1* | serine palmitoyltransferase long chain base subunit 1 | NG\_007950.1; NM\_006415.3 |
| *SPTLC2* | serine palmitoyltransferase long chain base subunit 2 | NG\_028282.1; NM\_004863.3 |
| *SPTLC3* | serine palmitoyltransferase long chain base subunit 3 | NC\_000020.11; NM\_018327.2 |
| *SURF1* | SURF1, cytochrome c oxidase assembly factor | NG\_008477.1; NM\_003172.3 |
| *TDP1* | tyrosyl-DNA phosphodiesterase 1 | NG\_009164.1; NM\_018319.3  |
| *TFG* | TRK-fused gene | NG\_027821.1; NM\_006070.5  |
| *TRIM2* | tripartite motif containing 2 | NG\_041788.1; NM\_015271.4  |
| *TRPV4* | transient receptor potential cation channel subfamily V member 4 | NG\_017090.1; NM\_021625.4 |
| *VCP* | valosin containing protein | NG\_007887.1; NM\_007126.3 |
| *VRK1* | vaccinia related kinase 1 | NG\_016293.1; NM\_003384.2 |
| *WNK1* | WNK lysine deficient protein kinase 1 | NG\_007984.3; NM\_213655.4 |
| *YARS* | tyrosyl-tRNA synthetase | NG\_008408.1; NM\_003680.3 |