Supplementary Table 1. Studied genes in the proband.

A coverage of 98.38% of the genomic regions of interest were represented with at least 25 reads (25x), and 93.66% of the regions have been sequenced with an average coverage of at least 50 reads (50x).

Retinal Dystrophy associated genes:

ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADIPOR1, AHI1, AIPL1, ALMS1, ARL13B, ARL6, ATF6, ATXN7, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, C2orf71, C5orf42, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CC2D2A, CDH23, CDH3, CDHR1, CEP290, CEP41, CERKL, CHM, CIB2, CISD2, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, CRB1, CRX, CTSD, CYP4V2, DFNB31, DHDDS, DNAJC5, DYNC2H1, EFEMP1, ELOVL4, EYS, FAM161A, FLVCR1, FSCN2, FZD4, GDF6, GLIS2, GNAT1, GNAT2, GNB3, GNPTG, GPR179, GPR98, GRK1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, GYLTL1B, HARS, HGSNAT, HK1, HMX1, IDH3B, IFT140, IFT80, IMPDH1, IMPG2, INVS, IQCB1, ITM2B, JAG1, KCNJ13, KCNV2, KIF11, KIF7, KLHL7, LAMA1, LCA5, LRAT, LRP5, LZTFL1, MAK, MERTK, MFN2, MFRP, MFSD8, MKKS, MKS1, MTTP, MVK, MYO7A, NBAS, NDP, NEK8, NEUROD1 NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OFD1, OPA1, OPA3, OPN1SW, OTX2, PANK2, PAX2, PAX6, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX2, PEX6, PEX7, PGK1, PHYH, PITPNM3, PNPLA6, POMGNT1, PPT1, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, PRPS1, RAX2, RB1, RBP3, RBP4, RD3, RDH12, RDH5, RGR, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SDCCAG8, SEMA4A, SLC24A1, SLC41A1, SLC9A6, SNRNP200, SPATA7, TCTN1, TCTN2, TIMM8A, TIMP3, TMEM126A, TMEM138, TMEM216, TMEM237, TMEM67, TOPORS, TPP1, TREX1, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TTPA, TUBGCP6, TULP1, UNC119, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WFS1, XPNPEP3, ZNF513.

Associated ataxia genes

AAAS, ABCB7, ABHD12, ACO2, ADCK3, AFG3L2, ANO10, APTX, ATCAY, ATM, ATP8A2, BEAN1, C10orf2, CACNA1A, CACNB4, CASK, CCDC88C, CLCN2, CLN5, CYP27A1, DARS2, DNMT1, ELOVL4, FGF14, FLVCR1, FXN, GOSR2, GRM1, IFRD1,

ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIAA0226, LAMA1, MARS2, MRE11A, MTPAP, MTTP, NOP56, OPHN1, PDYN, PHYH, PIK3R5, PLEKHG4, PNKP, PNPLA6, POLG, PPP2R2B, PRKCG, PTF1A, SACS, SETX, SIL1, SLC1A3, SLC9A6, SPTBN2, SPTBN2, SYNE1, SYT14, TBP, TDP1, TGM6, TPP1, TTBK2, TTPA, VLDLR, ZNF592

Supplementary Table 2. Previously reported mutations in the gene *FLVCR1* in HGMD.

Nucleotide	Protein	Reported phenotype	Reference
Missense variants			
c.361A>G	p.N121D	Posterior column ataxia & retinitis pigmentosa	Rajadhyaksha (2010) Am J Hum Genet 87, 643
c.479T>C	p.L160P	Retinitis pigmentosa	Tiwari (2016) Sci Rep 6, 28755
c.574T>C	p.C192R	Posterior column ataxia & retinitis pigmentosa	Rajadhyaksha (2010) Am J Hum Genet 87, 643
c.661C>T	p.P221S	Sensory neurodegeneration with loss of pain perception	Chiabrando (2016) PLoS Genet 12, 1006461
c.721G>A	p.A241T	Posterior column ataxia & retinitis pigmentosa	Rajadhyaksha (2010) Am J Hum Genet 87, 643
c.847G>C	p.A283P	Retinitis pigmentosa	Stone(2017)Ophthalmology124,1314
c.1058C>T	p.T353M	Retinitis pigmentosa	Stone(2017)Ophthalmology124,1314
c.1150G>C	p.G384R	Retinitis pigmentosa	Liu (2015) JAMA Ophthalmol 133, 427
c.1477G>C	p.G493R	Posterior column ataxia & retinitis pigmentosa	Ishiura (2011) Neurogenetics 12, 117
c.1547G>A	p.R516Q	Posterior column ataxia & retinitis pigmentosa	Shaibani (2015) Int J Neurosci 125, 43
c.1158T>G	p.I386M	Retinitis pigmentosa without posterior column ataxia	Kuehlewein (2019) Graefes Arch Clin Exp Ophthalmol 257
c.202C>T	p.Q68*	Retinitis pigmentosa without posterior column ataxia	Kuehlewein (2019) Graefes Arch Clin Exp Ophthalmol 257
Splicing variants			
c.883+6T>C	p.?	Retinitis pigmentosa	Liu (2015) JAMA Ophthalmol 133, 427
c.1092+5G>A	p.Ile343Argfs*31	Retinitis pigmentosa	Glöckle (2014) Eur J Hum Genet 22, 99
Small deletions			
c.610delA	p.(Met204Cysfs*56)	Sensory neurodegeneration with loss of pain perception	Chiabrando (2016) PLoS Genet 12, 1006461
c.755delG	p.(Gly252Alafs*8)	Retinitis pigmentosa	Glöckle (2014) Eur J Hum Genet 22, 99
c.1593+5_1593+8delGTAA	p.?	Posterior column ataxia & retinitis pigmentosa	Shaibani (2015) Int J Neurosci 125, 43
c.1557_1561delCATAA	p.(Asn519Lysfs*8)	Retinal dystrophy	Jespersgaard (2019) Sci Rep 9,
Small insertions			
c.774_779dupTTTAGT	c.774_779dupTTTAGT	Retinal dystrophy	Sergouniotis (2016) Orphanet J Rare Dis 11, 125
c.1324dupT	p.Tyr442Leufs*7	Sensory neurodegeneration with loss of pain perception	Chiabrando (2016) PLoS Genet 12, 1006461