PLA2G6 Mutations Related to Distinct Phenotypes: A New Case with Early-onset Parkinsonism

Supplementary Figures

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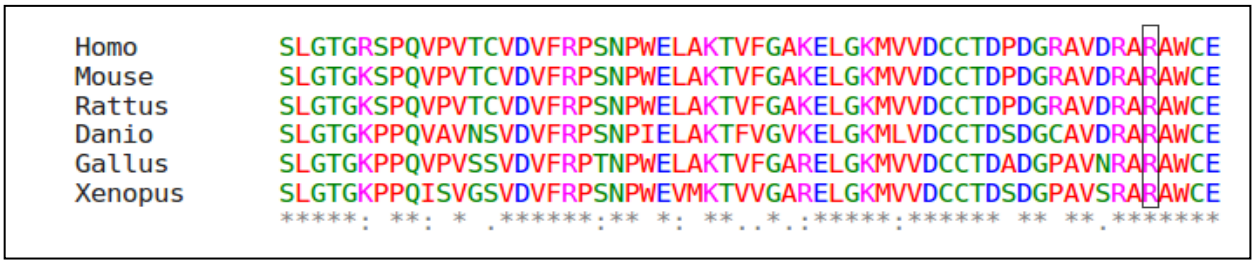

Figure S1. Multiple sequence alignment of pla2g6 protein showing that the region harboring p.R747W mutation (surrounded by a black square) is highly conserved across species.

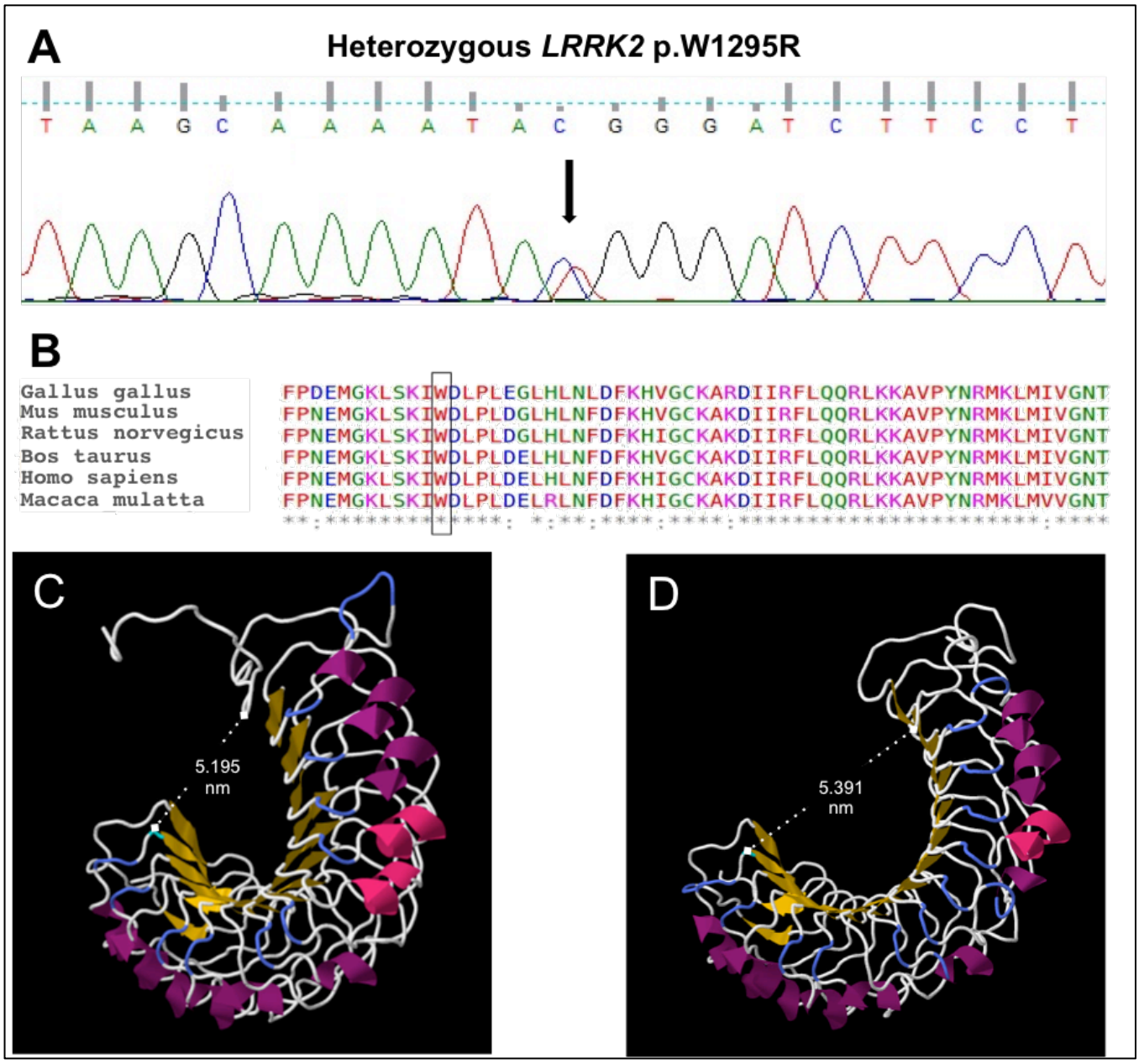

Figure S2. A) Electropherogram of *LRRK2* p.W1295R variant in the index case. The position of this variant is marked with a black arrow. **B)** Multiple sequence alignment of *LRRK2* homologues in the region targeted by p.W1295R variant (shown with a black square). **C-D)** Three dimensional conformation of wild-type (C) and mutated (D) LRR domain of lrrk2 protein. In the wild-type protein, the distance between positions 1295 and 985 is 5.195 nm while in the mutated protein it is 5.392 nm.

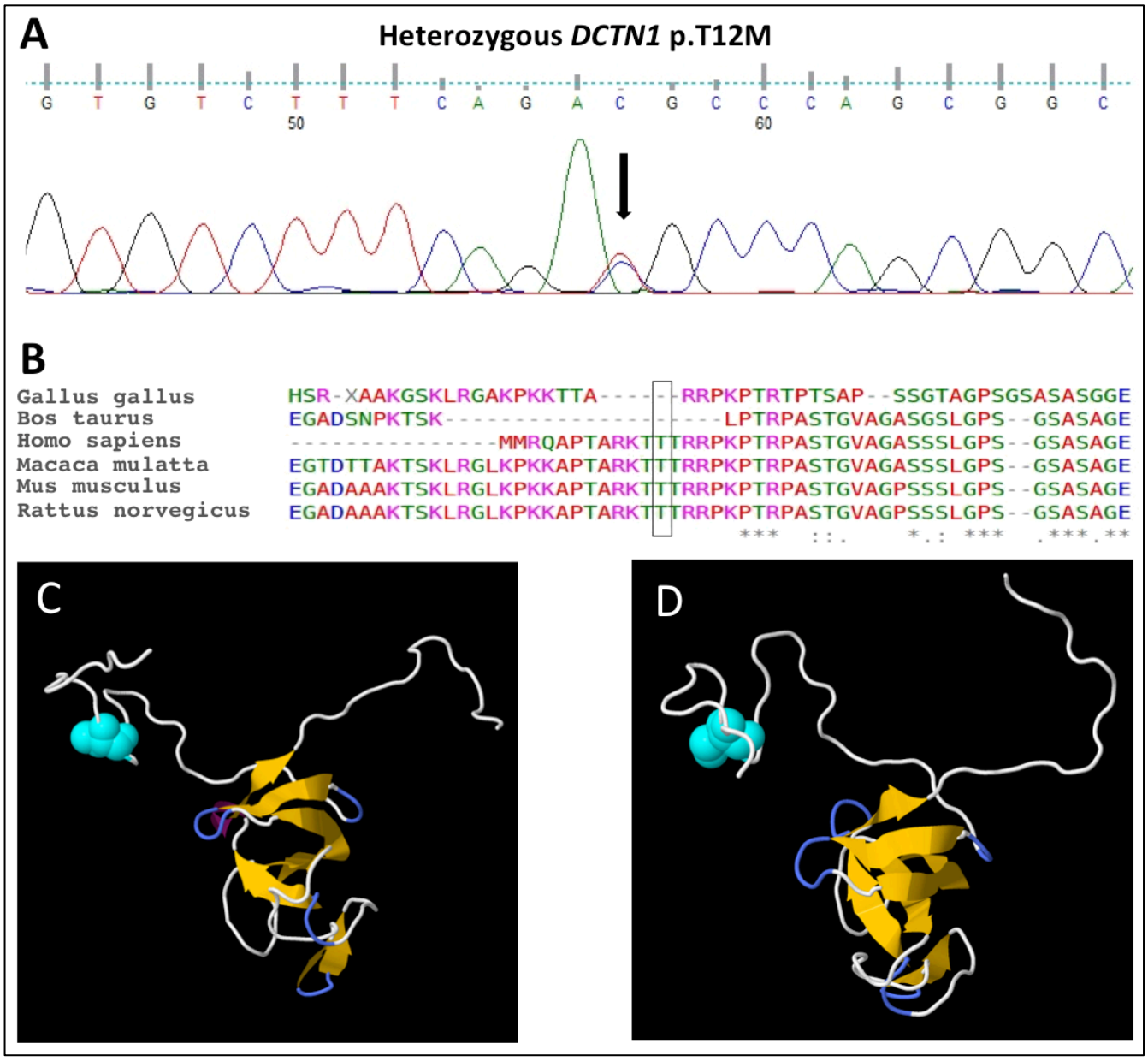

Figure S3. A) Electropherogram of *DCTN1* p.T12M variant in the index case. The position of this variant is marked with a black arrow. **B)** Multiple sequence alignment of dctn1 protein in the region targeted by p.T12M variant (shown with a black square). **C-D)** Three-dimensional conformation of (C) wild type and (D) mutated domain of dctn1.

AARS, AARS2, ABCB7, ABCD1, ABHD12, ACMSD, ADAR, ADCK3, ADD3, ADH1C, AFG3L2, AHI1, AIMP1, AIRE, ALAS2, ALG6, ALS2, ALS3, ALS7, AMACR, AMPD2, ANG, ANO10, ANO3, AOA1, AOA2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOE, APP, APTX, ARHGEF28, ARL13B, ARL6IP1, ARSA, ARSI, ASAH1, ASPA, ATCAY, ATL1, ATM, ATN1, ATP13A2, ATP1A3, ATP6AP2, ATP7A, ATP7B, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, AUH, B4GALNT1 , BCAP31, BCKDK, BEAN1, BICD2, BRAF, BSCL2, BST1, BTD, C10orf2, C10ORF2, C12ORF65, C19ORF12, C9orf72, C9ORF72, CA2, CA8, CACNA1A, CACNB4, CC2D2A, CCDC62, CCDC88C, CCT5, CEP290, CHCHD10, CHMP2B, CIZ1, CLCN2, CLN3, CLN5, CLN6, CLN8, COASY, COL4A1, COMT, COX20, CP, CRAT, CSF1R, CSTB, CTSD, CTSF, CYP27A1, CYP2U1, CYP7B1, DAO, DARS, DARS2, DCAF17, DCTN1, DDB2, DDHD1, DDHD2, DDRGK1, DGKQ, DLAT, DLG2, DNAJB2, DNAJC13, DNAJC19, DNAJC5, DNAJC6, DNMT1, DYNC1H1, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ELOVL4 , ELOVL5, ELP3, ENTPD1, EPM2A, ERBB4, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERLIN1, ERLIN2, FA2H, FAM126A, FAM47E, FBXO38, FBXO7, FGF14, FGF20, FIG4, FLRT1, FLVCR1, FOLR1, FTL, FUS, FXN, GAD1, GAK, GALC, GAN, GARS, GATA3, GBA, GBA2, GBE1, GCDH, GCH1, GCLC, GFAP, GIGYF2, GJB1, GJC2, GLA, GLB1, GNAL, GNAQ, GOSR2, GPNMB, GPR56, GRID2, GRN, Gaolf, HEPACAM, HEXA, HEXB, HLA-DQB1, HLA-DRB5, HNRNPA1, HNRNPA2B1, HPRT1, HSPB1, HSPB3, HSPB8, HSPD1, HTRA1, HTRA2, HTT, IGHMBP2, INPP5E, INPP5F, ITGA8, ITM2B, ITPR1, JPH3, KANK1, KCNA1, KCNC3, KCND3, KCNJ10, KCNMA1, KCTD7, KIAA0196, KIAA0226, KIF1A, KIF1C, KIF5A, KIF5C, L1CAM, L2HGDH, LMNB1, LRRK2, LYST, MAG, MAPT, MARS, MARS2, MATR3, MCCC1, MECP2, MFSD8, MIR4697, MIR4697HG, MLC1, MMP16, MRE11A, MTHFR, MTPAP, MTTP, NDUFV1, NEFH, NEU1, NHLRC1, NIPA1, NKX2-1, NMD3, NOP56, NOTCH3, NPC1, NPC2, NPHP1, NR4A2, NT5C2, NUCKS1, NUPL2, OPA1, OPA3, OPTN, PANK2, PARK2, PARK7, PARK7/DJ1, PAX6, PDE8B, PDGFB, PDGFRB, PDHX, PDSS1, PDSS2, PDYN, PEX10, PEX2, PEX7, PFN1, PGAP1, PGK1, PHYH, PINK1, PLA2G6, PLEKHG4, PLEKHG5, PLP1, PM20D1, PMM2, PNKD, PNPLA6, POLG, POLH, POLR3A, POLR3B, PPP2R2B, PPT1, PQBP1, PRICKLE1, PRICKLE2, PRKAR1B, PRKCG, PRKRA, PRNP, PRPH, PRRT2, PSAP, PSEN1, PSEN2, PTPN11, QDPR, RAB3GAP1, RAB3GAP2, RAB7l1, RAB7L1, RAF1, RAI1, RARS2, REEP1, REEP2, RELN, REPS1, RIT2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPGRIP1L, RTN2, SACS, SAMHD1, SCARB2, SCO2, SCP2, SETX, SGCE, SIGMAR1, SIL1, SIPA1L2, SLC16A2, SLC17A5, SLC1A3, SLC20A2, SLC25A15, SLC25A4, SLC2A1, SLC30A10, SLC33A1, SLC5A2, SLC5A7, SLC6A3, SMPD1, SNCA, SOD1, SOX10, SPAST, SPG11, SPG20, SPG21, SPG7, SPPL2B, SPR, SPTBN2, SQSTM1, SREBF1, STK39, STUB1, STX1B, SUMF1, SYNE1, SYNJ1, SYT11, SYT14, TAF1, TAF15, TARDBP, TBP, TDP1, TECPR2, TFG, TGM6, TH, THAP1, TIMM8A, TMEM163, TMEM175, TMEM216, TMEM67, TOMM40, TOR1A, TPP1, TREM2, TREX1, TRPV4, TSEN2, TSEN34, TSEN54, TTBK2, TTPA, TUBB4A, TUBB4A , TYROBP, UBA1, UBQLN2, UCHL1, UNC13A, USP25, USP8, VAMP1, VAPB, VCP, VEGFA, VLDLR, VPS13A, VPS13C, VPS35, VPS37A, VPS54, VRK1, WDR45, WDR48, WFS1, WWOX, XK, XPA, XPC, ZFR, ZFYVE26, ZFYVE27, ZNF592

Box S1. List of genes with different implications in neurodegeneration (NDD). Variation within all these genes was investigated in the WGS data generated.