

Supplementary table 1: Copy number variations (CNVs) in the patient

CNV	Chromosomal position	Genomic position ^a	Affected gene
Copy-number gain	4q34.3-q35.2	max: chr4: 179034540-qter min: chr4: 179094770-qter	<i>MGC45800, MIR1305, ODZ3, DCTD, FAM92A3, C4orf38, WWC2, CLDN22, CDKN2AIP, ING2, RWDD4A, C4orf41, STOX2, ENPP6, IRF2, CASP3, CCDC111, MLF1IP, ACSL1, SLED1, HELT, SLC25A4, KIAA1430, SNX25, LRP2BP, ANKRD37, UFSP2, C4orf47, CCDC110, PDLIM3, SORBS2, TLR3, FAM149A, CYP4V2, KLKB1, F11, MTNR1A, FAT1, ZFP42, TRIML2, TRIML1</i>
Copy-number loss	7q35-q36.3	max: chr7:145612170-qter min: chr7:145619934-qter	<i>CNTNAP2, MIR548I4, MIR548F4, MIR548F3, C7orf33, CUL1, EZH2, PDIA4, ZNF786, ZNF425, ZNF398, ZNF282, ZNF212, ZNF783, ZNF777, ZNF746, ZNF767, KRBA1, ZNF467, SSPO, ZNF862, LOC401431, ATP6V0E2, ACTR3C, LRRC61, C7orf29, RARRES2, REPIN1, ZNF775, LOC728743, GIMAP8, GIMAP7, GIMAP4, GIMAP6, GIMAP2, GIMAP1, GIMAP5, LOC100128542, TMEM176B, TMEM176A, ABPI, KCNH2, NOS3, ATG9B, ABCB8, ACCN3, CDK5, SLC4A2, FASTK, TMUB1, AGAP3, GBX1, ASB10, ABCF2, CHPF2, MIR671, SMARCD3, NUB1, WDR86, CRYGN, RHEB, PRKAG2, GALNTL5, GALNT11, MLL3, FABP5L3, LOC100128822, XRCC2, ACTR3B, DPP6, LOC100132707, PAXIP1, LOC202781, HTR5A, INSIG1, EN2, CNPY1, RBM33, SHH, C7orf4, C7orf13, RNF32, LMBR1, NOM1, MNX1, UBE3C, DNAJB6, PTPRN2, MIR153-2, MIR595, NCAPG2, ESYT2, WDR60, LOC154822, VIPR2</i>
Copy-number gain	Xp22.31	max: chrX: 6430569-8164744 min: chrX: 6445321-8152876	<i>HDHD1A, STS, VCX, PNPLA4, MIR651</i>

^a Genomic positions are referred to Human Genome Database (hg19, build 37).

max: maximum interval of the CNV (the interval between the first copy-number neutral probes at both sides of the CNV)

min: minimum interval of the CNV (the interval between the two most distant probes within the CNV)