**Figure S1** - Analysis of the *CEP85L* transcript in patient 1. (A) Graphical representation of *CEP85L* exons 1-3 (not drawn to scale); the position of the primers used on cDNA is indicated by red arrows. (B) cDNA amplification of exon 1-3 region in cDNA from two wild-type controls (proband's father, f, and mother, m) and the proband, p; nt=no-template control, mw=molecular weight marker (Gel Pilot 100 bp ladder, QIAGEN, Maryland, USA). The proband's lane presents both the expected 424 bp amplicon and a band at lower molecular weight. (C) Sequencing of the cDNA from the proband's low-weight band demonstrates the formation of a 1/3 exon junction.
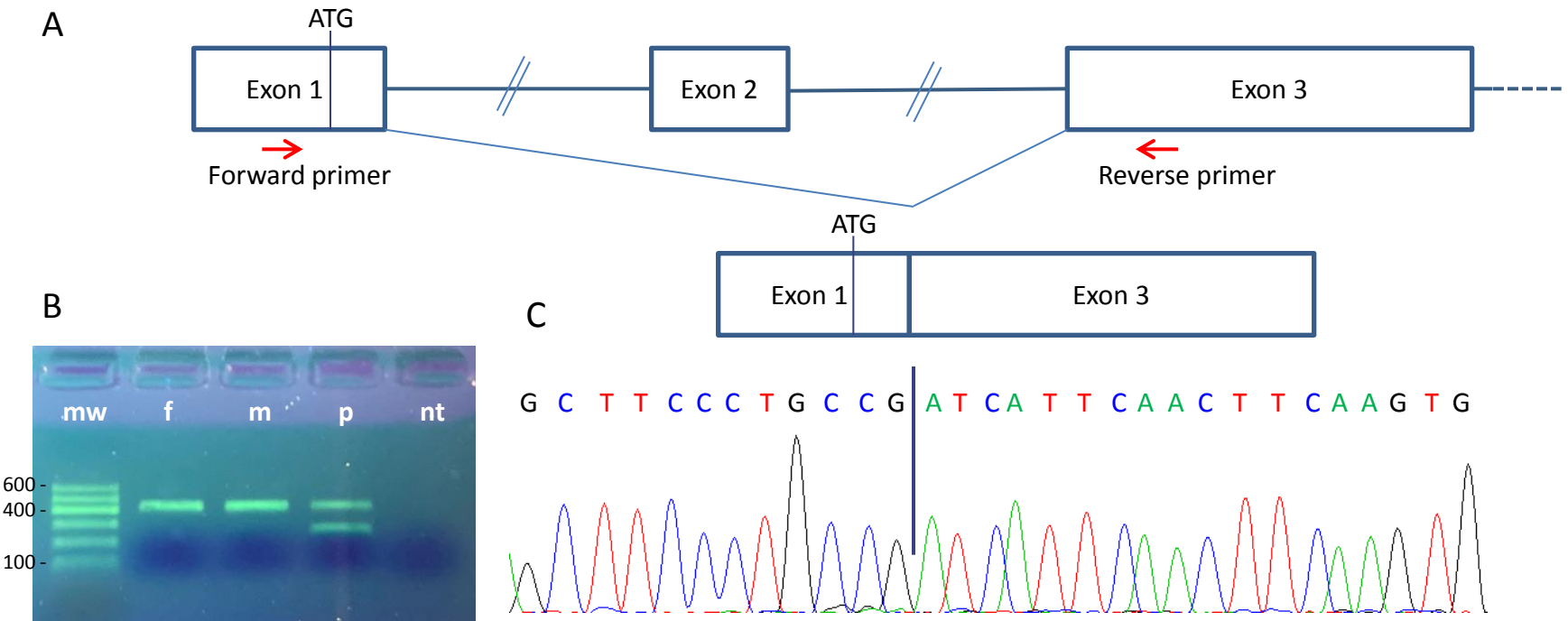
Table S1 - Sequence (5' -> 3') of the primers used for analysis of the *CEP85L* cDNA (and β-actin control)

CEP85L_cDNA_1F	AAGAAGCAGGAGGACGGAC
CEP85L_cDNA_1R	TGGTGTCAATGATTCCCTAAG
ACTB_F	GCAAGAGATGGCCACGGCTG
ACTB_R	ATCCACACGGAGTACTTGCG

Table S2. List of the 182 genes associated with brain malformations analyzed throught a targeted Next Generation Sequencing (NGS) custom panel.

ACTB	CELSR2	EIF4EBP1	KIF5C	PAX6	RPS6	TMEM237
ACTG1	CENPE	EML1	KIF7	PCLO	RPS6KB1	TMEM67
ADGRG1	CENPJ	EMX2	KNL1	PDE6D	RPTOR	TMTC3
AHI1	CEP135	ERMARD	LAMA2	PHC1	RTTN	TSC1
AKT1	CEP152	EXOSC3	LAMB1	PI4KA	RXYLT1	TSC2
AKT1S1	CEP290	EXOSC8	LAMC3	PIK3CA	SASS6	TSEN15
AKT3	CEP41	EZH2	LARGE1	PIK3R2	SEPSECS	TSEN2
AMPD2	CEP63	FAT4	MCPH1	PLK4	SHH	TSEN34
ANKLE2	CHMP1A	FIG4	MFSD2A	POGZ	SIX3	TSEN54
ARFGEF2	CIT	FKRP	MKS1	POMGNT1	SLC25A19	TUBA1A
ARL13B	CLP1	FKTN	MLST8	POMGNT2	SRGAP2	TUBA8
ARX	CNTNAP2	FLNA	MTOR	POMT1	STAMBP	TUBB
ASNS	COL4A1	FOXC1	MYCN	POMT2	STIL	TUBB2A
ASPM	COL4A2	GLI2	NDE1	PPP1R15B	STRADA	TUBB2B
ATRX	CRADD	GMPPB	NEDD4L	PQBP1	STX7	TUBB3
B3GALNT2	CSPP1	GNAQ	NFIX	PTCH1	TBC1D20	TUBG1
B4GAT1	CTNNB1	GPSM2	NHEJ1	RAB18	TBC1D23	TUBG2
B9D1	CUL4B	IER3IP1	NPHP1	RAB3GAP1	TBC1D7	VLDLR
C5orf42	DAG1	INPPSE	NPRL2	RAB3GAP2	TBCD	VPS53
CC2D1A	DCHS1	ISPD	NPRL3	RALGAPA1	TCTN1	VRK1
CC2D2A	DCX	KANSL1	NSD1	RALGAPA2	TCTN2	WDR62
CCND2	DEPDC5	KAT6A	OCLN	RALGAPB	TCTN3	WDR81
CDK5	DEPTOR	KATNB1	OFD1	RARS2	TGIF1	YWHAE
CDK5RAP2	DNAI1	KIAA0556	OPHN1	RELN	TMEM138	ZIC1
CDK6	DYNC1H1	KIAA0586	ORC1	RICTOR	TMEM216	ZNF335
CDON	EIF4E	KIF11	PAFAH1B1	RPGRIP1L	TMEM231	ZNF423

Table S4 - *In silico* predictions on how the different variants at CEP85L exon 2 donor site affect the canonical 5' splice site

NM_001042475.3:	MaxEnt (score)	NNSPLICE (confidence)	NetGene2 (confidence)	SPiCE (probability)	Patient #[†]
WT	9.60	0.89	0.79	0.02686	
c.232+1del	-9.38	0.00	0.00	1	1
c.232+1G>T	1.09	0.00	0.00	1	10
c.232+3G>T	5.61	0.46	0.00	0.89191	11, 12
c.232+5G>T	4.41	0.00	0.00	0.99519	20, 21
c.232+5G>A	5.54	0.00	0.00	0.98584	13, 14

†Case numbers refer to patients as presented in Table S3